CLARIFYING THE CONCEPT OF DISEASE: HOW SCIENCE DETERMINES THE BIOLOGICALLY NORMAL FROM THE ABNORMAL

BY

LOREN A. ZECH

DISSERTATION

Submitted in partial fulfillment of the requirements for the degree of Doctor of Philosophy in Philosophy in the Graduate College of the University of Illinois at Urbana-Champaign, 2011

Urbana, Illinois

Doctoral Committee:

Professor Robert Wengert, Chair
Professor James Wallace
Professor Jonathan Waskan
Doctor Evan DeRenzo, Center for Ethics, Washington Hospital Center
ABSTRACT

This is an investigation of the modern concept of disease and an explication of the concept consistent with scientific and medical usage. The core notion involved in the modern concept of disease (taken broadly as pathology) is biological abnormality in form or function, for a circumscribed biological system, contrasted against a theoretical sense of normal form or function. Chapter 2 presents an overview of science, its products and how these are produced. This sketch is limited and intended only to clarify my own view of science in as far as this is important for the main thesis. Chapter 3 discusses the concepts of normality and abnormality, particularly as it is used in biomedical science. I argue that science offers a theoretical sense of abnormality importantly different from statistical or conventional senses of abnormality. Chapter 4 begins with an examination of the normativist/naturalist debate on the concept of disease. I then offer my explication of disease and the criteria necessary for a disease claim to be legitimate. Chapter 4 closes with an extended discussion of interesting and controversial examples of disease claims aimed at clarifying and challenging my own position.
To Catherine, Cathy, and Lorene. Thanks for all the support.
ACKNOWLEDGEMENTS

I could not have completed this project without the support of the Philosophy Department at the University of Illinois. I appreciate the tireless guidance, friendship, and encouragement of my thesis advisor Prof. Robert Wengert. I also appreciate the time, effort, and advice from my committee members, James Wallace, Jonathan Waskan, and Evan DeRenzo. Peggy Wells also deserves thanks. I would also like to thank all my family and friends who encouraged and supported me while working on this project, in particular Catherine Zech, Cathy Monro, Lorene and Fred Alexander, Roger and Virginia Kerley, Sandeep Sodhi, Matt Olsen, and Celeste Yasunaga. I would also like to thank the Internal Medicine Residency Program at Washington Hospital Center for allowing me research time during my intern year to complete the manuscript.
# TABLE OF CONTENTS

<table>
<thead>
<tr>
<th>CHAPTER</th>
<th>TITLE</th>
<th>PAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>INTRODUCTION</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>SCIENCE</td>
<td>14</td>
</tr>
<tr>
<td>3</td>
<td>NORMALITY</td>
<td>65</td>
</tr>
<tr>
<td>4</td>
<td>DISEASE</td>
<td>126</td>
</tr>
<tr>
<td>5</td>
<td>CONCLUSION</td>
<td>218</td>
</tr>
<tr>
<td></td>
<td>BIBLIOGRAPHY</td>
<td>221</td>
</tr>
</tbody>
</table>
CHAPTER 1

INTRODUCTION

The topic of this thesis is the concept of disease. I am interested in what is being claimed when we say that certain problems that a person is having are caused by a disease. Claims about newly recognized disease are more frequent now that pharmaceutical companies can market directly to consumers via television commercials. Some of these claims about disease are controversial. In the United States, for example, a series of commercials have recently been warning men about a potential undiagnosed medical problem. One of these commercials states “Millions of men 45 and older just don’t feel like they used to.” It then asks if you, or someone you love, may be one of the sufferers, and also warns: “Don’t blame it on aging.” The commercial then advises those concerned to go to a website and take a short quiz to further clarify if they may have warning symptoms and if they should seek treatment from a physician.¹ The quiz consists of the following 10 screening questions:

1) Do you have a decrease in libido (sex drive)?
2) Do you have a lack of energy?
3) Do you have a decrease in strength and/or endurance?
4) Have you lost height?
5) Have you noticed a decrease in your enjoyment of life?
6) Are you sad and/or grumpy?
7) Are your erections less strong?
8) Have you noticed a recent deterioration in your ability to play sports?
9) Are you falling asleep after dinner?
10) Has there been a recent deterioration in your work performance?²

Answering yes to at least 3 of these screening questions, especially to question 1 or 7, may mean that low testosterone levels are causing a medical condition (i.e. a disease). This biological state is referred to by the commercials and the website as ‘Low T.’ The

¹ The commercial I am referring to is available on-line at http://www.isitlowt.com. I accessed it last on 8/27/11.
² The quiz is available on-line at http://isitlowt.com/do-you-have-low-t/low-t-quiz-test.html. I last accessed the quiz on 8/27/11.
website also advises people to be prepared to talk to their doctor about ‘Low T’ and offers a helpful set of questions to ask at your visit with a physician:

1) I don’t feel sick, I just don’t feel like myself anymore. What could be causing it?
2) Are the symptoms I am experiencing and my other conditions related to Low T? (Feel free to discuss your results from the Low T Symptoms Quiz.)
3) Considering my symptoms, should I be tested for Low T?
4) What medical treatment options are available if I have Low T?
5) What is the difference between the different testosterone replacement therapies?
6) If I have Low T, what happens if I don’t have it treated?
7) How does my diet, fitness, and lifestyle affect my testosterone?³

The skeptical may wonder whether ‘Low T’ is a ‘real’ disease or not. If some diseases are ‘real’ that implies some claims about a person’s problems being the result of disease may be fake or illegitimate in some way.⁴ Less skeptical viewers, particularly those who are feeling a little off, may make their way to the website and may wonder if ‘Low T’ explains their problems. If so, once diagnosed, medical intervention may alleviate the problems. Consider my case. I currently screen positive for more than 3 symptoms, and perhaps ‘Low T’ explains why I feel this way. However, I may be suffering from other sorts of problems. Over the past two years I have been completing a medical residency program. I have turned 40 during this time period. I have been working in a busy hospital, including, at times, 30 hour shifts without sleep. In addition to the 60-plus hours of work per week on duty, I have also been trying to read and study medicine in my off-duty hours, as well as occasionally working on this thesis. I have not been exercising as I was accustomed to as a graduate student, nor have I been eating a very healthy diet recently. The job is stressful. Perhaps my symptoms are from too much stressful work, too little exercise, and too much cafeteria food. Perhaps my problems are not symptoms of a disease. Perhaps they are a different kind of human problem, with a different kind of cause.

³ These questions to ask at one’s clinic visit are found at: [http://www.isitlowt.com/what-you-can-do/doctors-discussion-guide.html](http://www.isitlowt.com/what-you-can-do/doctors-discussion-guide.html). I last accessed the website on 8/26/11.
⁴ For the remainder of the thesis I will use the terminology of legitimate versus illegitimate disease claims for such controversies, and avoid the real vs. the fake imaginary dichotomy. It is not clear what un-real amounts to. I hope to specify what being a legitimate vs. illegitimate disease claim constitutes.
The example of ‘Low T’ raises several issues with the concept of disease. ‘Low T’ is actually being called a ‘medical condition’ that may be caused by another disease such as diabetes, hypogonadism, obesity, high blood pressure, high cholesterol, or chronic obstructive pulmonary disease. In fact, ‘Low T’ is merely the state of having a serum level of testosterone low enough to be outside the normal range of testosterone levels for (presumably healthy) men of a similar age, and it may apparently be caused by several other biological problems. It is a statistical abnormality and sign of disease. It may be another of the signs and symptoms that something is wrong medically. Discovering this underlying cause and reversing it may alleviate the symptoms, but if not reversible the symptoms may be alleviated by supplementing the testosterone levels back to the normal range. (In chapter 3, I shall discuss statistical abnormality as a warrant for disease claims.) To further complicate matters, assume that if tested, I would be found to have low levels of testosterone without any of the other associated medical conditions like obesity or diabetes. It might be that stress from too much work, too little relaxation and exercise, and all that cafeteria food caused my testosterone to drop. After all, I was supposed to ask my doctor about how diet, fitness, and lifestyle may be affecting my testosterone levels. Could I then claim that I had a disease brought on by stress that was causing low testosterone? Perhaps I have stress induced or idiopathic ‘Low T.’ Such an explanatory story might make the difference between whether an insurance plan pays for the testosterone supplements or not.

Compare this to less controversial disease claims. Take the case of a neoplasm of the thyroid gland. Consider a follicular adenoma of the thyroid. This often presents, when it grows large enough to be felt easily, as a solitary nodule in an otherwise grossly normal thyroid gland. It is due to increased growth of the follicular epithelium that makes up most of the thyroid gland. The neoplastic nodule consists of the cells that produce thyroid hormones as well as follicles or pools of stored precursors. These neoplastic growths are often silent as far as symptoms, but may occasionally overproduce thyroid hormone and cause the symptoms of hyperthyroidism (i.e. increased basal metabolic rate with weight loss, soft, warm, flushed skin, intolerance of heat, tachycardia, palpitations, possibly cardiac arrhythmias, tremor, hyperactive tendon reflexes, muscle cramps, emotional lability, stereotypical gaze and eyelid lag, etc.). These follicular adenomas are caused by
acquired genetic defects that cause the follicular cells to grow regardless of the molecular
signals that normally regulate growth, initiate cell reproduction, or halt it. Some specific
mutations are known, but many others are hypothesized and are yet to be determined.
Epidemiological data show that thyroid adenomas, although neoplastic growths, rarely
progress to malignancies that could metastasize. However, the clinical problem of
distinguishing the more common benign local adenomas from rarer potentially malignant
carcinomas of the thyroid is difficult without cutting out and examining the nodule
microscopically (see Kumar, 2005). This case of having a neoplastic mass of thyroid cells
that no longer is under the control of the body’s normal signals is an uncontroversial case
of disease. Interestingly, such a mass is a disease whether it causes the typical symptoms
of excess thyroid production or not. Some nodules do not secrete hormone and do not
cause the systemic symptoms. It is also a disease regardless of being discovered clinically
on examination of the neck with palpation of the gland through the skin. It may be
discovered only at autopsy after death or possibly never discovered. In addition to these
examples, there are paradigmatic cases of disease that must be accounted for on any
analysis or clarification of the concept. Examples include heart attack, insulin dependent
diabetes mellitus, and tuberculosis. I will discuss the example of heart attack in greater
detail in the introduction to chapter 4.

Questions about the nature of disease and about particular disease claims can be
motivated by different concerns or perspectives. The ‘Low T’ disease claims might
arouse fears that our contemporary society is medicalizing kinds of human problems that
are not primarily medical problems. The sudden epidemic of attention-deficit
hyperactivity-disorder (ADHD) affecting our society’s children may also be the
medicalization of problems that are not biological problems, perhaps not even
psychological problems. They may not even be problems, except in the eyes of certain
groups in the community who find certain behaviors abnormal and thus in need of
remedy. Historical examples of disease claims concerning masturbation, homosexuality,
drapetomania, or hysteria might also be seen as the medicalization of perceived
abnormalities that are not primarily biological malfunction nor best addressed in a
medical context. Sometimes the controversy over a disease claim stems from
disagreement over the extent to which the sufferer’s problems are within their own
control to change. The question may be whether a biological (or even psychological) explanation is the appropriate way to try and understand the nature of the problem. Alcoholism and addiction generally have been controversial in this regard. Obesity is a condition being claimed by some to be a disease, while others see this as medicalization. Sometimes the concern is whether the patient is malingering so as to get some non-physiological benefit from treatment, or from assuming the sick role, etc. Subtly related are issues about whether the patient’s goal is gaining some advantage or other desired outcome of being treated with biotechnology and not the alleviation of discomfort or a return to health. Perhaps some men seek not just feeling back to normal, but hope they will feel and perform better than normal once their “Low T” is treated. Furthermore, the claim that ‘Low T’ is a legitimate disease may also involve the difficulty of distinguishing normal aging and normal changes in the body from abnormal changes or pathology. When are aches and pains, or subtle changes in one’s endurance, abilities, memory, etc. just another part of normal aging? On the other hand, perhaps aging and death are the two most common and most dreaded diseases.

My own project combines intellectual curiosity and practical interests. As a physician I am interested in current usage of the concept in medicine and biomedical research. Physicians use the term regularly, and attempt to make the distinction for most patients between the biologically normal and abnormal. Many patients want to hear that their problem is not serious—that it is nothing to worry about—a benign or normal process soon to go away. Physicians are asked almost daily about what the cause is for the problem troubling the patient. Other patients care little about the cause and want to know that the troubling aspects of the process can be ameliorated, if not permanently, at least for the time being. The practice of medicine brings one face to face with issues of whether some problem is one of biology or something else, as well as questions of where normal variation or normal reactions to stressful or imperfect circumstances ends and subtle pathology begins. Interestingly, there is little discussion of the general nature of disease in medical school. The concept is learned by reviewing particular examples and by constantly contrasting the normal processes and functions of biological systems with the various ways that malfunction of malformation may occur. One looks in vain in respected medical textbooks for a general definition. However, I do believe that there is a
general understanding that physicians and biomedical researchers share. This thesis is an attempt to distill that concept into somewhat clearer form.

Practical concerns are also increasing the need to clarify what disease is, and which particular claims are legitimate and which not. I am interested in arguments and discussion about the moral concerns with using biotechnology for ends other than therapy. This includes using biotechnology for enhancement, self-expression, or abnormal longevity and survival. However, determining where the boundary is between therapeutic and extra-therapeutic uses of biotechnology rests on some demarcation of disease from other types of concern or problems that people may have. Therapy is biotechnical intervention for the purpose of curing disease or returning the patient to some approximation of normal. Extra-therapeutic biomedical intervention is intended not to cure disease or fix a malfunction, but fulfill some other human desire, usually those having to do with self-improvement, self-expression, or competition with others.

One frequently hears that the goal of medicine is to alleviate suffering. Even a short amount of reflection reveals this is either too broad a goal or medicine has been, from the beginning, aiming short of the goal. There are many kinds of suffering, and some of them do not primarily involve biology. Friends, parents, politicians, economists, engineers, lawyers, ethical counselors, religious authorities, etc. all have roles in alleviating certain kinds of human suffering. Medicine, particularly modern medicine in the Western tradition, is selective about the type of suffering and types of problem it sees as its province. A more accurate goal for medicine is the curing of disease. However, even given that narrower goal, the actual practice of medicine could be very different (and perhaps has been across epochs and cultures) depending on the conception of disease being assumed.

Medical practice is also changing because of its great success and increased ability not only to describe and understand biological phenomena but to manipulate and control them. The technology to do this is increasingly costly. Difficult decisions are having to be made regarding how much medical care we owe to each other as a matter of social justice versus which types of biomedical intervention should be available only to those who can afford it. Decisions have to be made about what social apparatuses should be arranged to cover the costs of those types of care we want to make available to all.
There are also ethical concerns about labeling some biological differences as disease, particularly because doing so might foster increased discrimination of those who are labeled as defective in form or function. In chapter 3 I shall discuss this in more detail.

Many of these issues can be conceptualized as boundary disputes. Controversy concerns where to draw a line distinguishing different types of phenomena. For example, some wish to distinguish between therapy and enhancement. Other boundary disputes include: medical intervention available to all versus optional or unnecessary intervention; the line where medical practice ends and other sorts of problem solving begin; and the distinction (if any) between psychiatry and the rest of medicine. I am interested in all these boundary issues, and think they are all interestingly related to the boundary between disease and non-disease. Driving much of the controversy in all these disputes is the distinction between the biologically normal and abnormal. For the person arguing that disease is an objective scientific term, the crucial question is how can science possibly distinguish the normal from the abnormal. How can science do so in a way that is largely free of personal and community judgments about what is preferable and desirable about human bodies, behaviors and performance? I argue that science can sometimes offer us respectable, unbiased descriptions of the biologically normal and abnormal. The view one has of science, its intellectual products, and how it produces these products importantly informs how one will conceive the concept of disease. Therefore I briefly discuss science and its products in chapter 2.

My project is clarifying or explicating the concept of disease as used currently by physicians, scientists and derivatively by educated adults in Western societies. I am interested in the scientist’s and the expert’s term. I am interested in defending a naturalist conception of disease as far as possible. Some now argue that the naturalist versus normativist conception of the problem of disease is not a helpful way to approach the issue (see Schwartz, 2007). I disagree. The normativist/naturalist debate has persisted because it continues to capture the fundamental tensions that people find interesting or puzzling about the concept of disease. Furthermore, although some crude form of naturalism is the position of most contemporary physicians and educated adults in my community, there have been surprisingly few naturalist theories of disease offered in the philosophical and humanities literature in the past 40 years. The predominant naturalist
position is that of Christopher Boorse (1975, 1977, 1987, 1997). The most often defended position in the literature, however, is normativism of one variety or another (see King, 1954; Engelhardt, 1996; Reznek, 1987). My project is to offer a naturalistic account of the concept of disease that captures the scientific usage of the term. I find much useful in Boorse’s naturalistic theory, and share his project, but ultimately find it in need of refinement, and discuss this in chapter 4.

There are other projects in the literature examining the concept of disease. These are worthy projects and offer valuable insights. However, they are not primarily my project. Let me clarify by dismissing some of these other perspectives and projects. Some projects are more pragmatic and explore the concept of disease in the context of what doctors actually do for patients. Some projects are sociological and explore disease as a classifier and describer of kinds of people and their behavior (those with illnesses and diseases) and explore the possible roles (sick role, doctor role, etc.) assumable within some community (see Parsons, 1971). Disease may have broader and narrower senses and be used differently across contexts and for different groups within a community. Projects may be more focused on semantics and linguistic usage (see Simon, 2007). In chapter 4, I will return to the issues about the actual usage of the term and issues of broader or narrower usages. Basically, there may be dispute about whether the word ‘disease’ actually describes both chronic and acute problems, both systemic and localized problems, active problems versus damage sustained from past trauma or currently inactive processes, as well as whether it applies to internal and external causes. Here it is worth noting that I am interested in the broadest sense of disease, a use perhaps synonymous with pathology. As such, the concept of disease may be a cluster concept involving many of the diverse elements that can be partially captured from the various narrower senses or the other perspectives and projects. The term seems to defy any attempt at definition by necessary and sufficient conditions. However, as noted above, my project is to see if a coherent and recognizable scientifically respectable explication of disease (as pathology) is possible that reliably explains uncontroversial paradigmatic examples, explains points of current controversy in a satisfying way, and potentially can be applied to future controversial cases.
Projects exploring the concept disease may also be primarily historical, and account for the changes in the concept across time and culture. Historical projects can be combined with any of the other types of concerns and projects above (see Engelhardt, 1996; Thagard, 1996). I have not primarily investigated the historical development of the concept of disease, although I have used historical examples. It is difficult to avoid the temptation to investigate past disease claims and see how they fare as contemporary claims. I have not addressed how the concept of disease may have developed and evolved across different cultures and in response to certain historical developments and events.

What I would argue (if this question of historical development were an important aspect of my project) is that biological (and psychological) abnormality contrasted with the biologically normal has always been a fundamental part of the concept of disease. Other core elements (in the possible cluster of elements) include those of incapacity, infirmity, loss of ability, suffering, and pain, and the perception of being afflicted with a burden beyond the control of one’s willpower. I would argue that as biological science has grown increasingly successful over the past 200 or so years, the notion of biological abnormality has assumed a predominant position at the core of the concept, while the other elements have increasingly been marginalized. As science has gained the ability to describe, explain, manipulate and control biological phenomena at increasingly remote (i.e. microscopic and ultrastructural extremes) it has been able to rely less on gross symptoms and signs in diagnosing and characterizing disease. Furthermore, explanation at the microscopic level has allowed meaningful distinctions between the mere signs and symptoms of a particular disease and the actual causal mechanisms constituting the disease. It is possible that disease claims from different historical periods and different cultures are incommensurable. It may also be the case that disease claims from different branches of medicine, or even different branches of biology, are incommensurable. I will be assuming that none of these possibilities is the case. We can recognize disease claims from other cultures and epochs as expressing commensurate fundamental core aspects of the modern concept. Plant biologists, veterinarians and physicians talk of disease and understand roughly the same thing by the term, as do psychiatrists and endocrinologists.

---

5 In chapter 3 I shall discuss the historical origins of the term normal and abnormal during the early years of modern physiology.
The core notion at the center of the modern concept of disease is biological abnormality. I think this has always been a core notion involved with the concept of disease, but it has come to dominate the concept as science has experienced exponential success in explaining and modeling biological phenomena. Biology has increasingly broken down organisms into subsystems, isolated these, and experimentally manipulated and described them. These descriptions necessarily involve distinctions between normal and abnormal processes and parts for the circumscribed systems being functionally decomposed. Smaller more carefully isolated systems are combined to offer larger more complex and dynamic models of metabolism, tissue function and malfunction, organ function and disease, and eventually models of entire physiological systems, such as the cardiovascular or endocrine system. The important question for the modern, scientific concept of disease boils down to how biological science can distinguish the normal from the abnormal and whether it can do so in ways that are not primarily or exclusively reflections of personal or social preference or values. Can science offer a relatively objective and primarily descriptive notion of abnormality and thus of disease? I argue that it can and does do this.

As mentioned, the continuing revolution in scientific explanation and exploration has also allowed for precise isolation and control of circumscribed biological systems. Thus disease is increasingly not considered a global phenomenon but a localized malfunction in a discrete localizable biological system. Interestingly, unlike disease, health is still most often a term applying to organisms globally. A negative definition of health would be the absence of disease, but many are interested in a broader, positive, welfare conception of health involving not just absence of disease, but some basic level of opportunity, capacity and flourishing (see Nordenfelt, 1987, 2006). It is unclear if science alone can ever deliver this sort of positive definition, or deliver pronouncements of such states of health, independent from political or ethical considerations. Medicine, however, may someday be able to offer negative pronouncements of health—essentially confirming that all one’s biological and psychological systems, to the limits of detection, are functioning normally.

---

6 Physicians sometimes do speak of a healthy heart or other isolated organ, but this is metaphorical and derivative upon the primary notion of the healthy organism.
My main thesis is that the scientific concept of disease, at its core, relies on the notion of biological abnormality. This is often biologically abnormal function. This is the conceptualization that best unifies the diversity of particular disease claims that we take to be legitimate and uncontroversial. Biological abnormality is also the notion that arouses most of the controversy when particular claims are disputed. Controversy arises over whether the particular type of problem, incapacity, or suffering in question is attributable to abnormal biological processes or parts. Consider the controversy over alcoholism. This can cause suffering and interfere with one’s ability to successfully contribute to the life of the community. The controversy about claiming this is a disease concerns whether this is an abnormality of the functioning of some biological system in those afflicted, or rather another sort of problem being erroneously medicalized. The other sort of problem here might be one of weak will, poor upbringing, poor choices, etc.\(^7\) Much of the controversy over ADHD revolves around whether this is primarily a problem of abnormal biological function or abnormal behavioral demands on normally functioning children. Determining whether biology is involved is sometimes very difficult, and I will look at some difficult cases along these lines in chapter 4.

The scientific models that distinguish normal from abnormal biological processes are human products. They are representations and as such can be more and less accurate. They can be more or less simplistic or detailed. They can be biased by personal or social agendas or prejudices, sometimes intentionally and blatantly so, but often their bias is more subtle and unintentional. But, as the original hypothetical model is scrutinized, challenged, and tested, it can be refined, and some models survive and evolve to be more accurate and less biased. Their evolution is effected not just by the social forces that are part of the scientific community, but by groups of scientists continuing to interact with the phenomena being modeled. These survivors become more sophisticated and intricate and increasingly branch out to help support other surviving models while simultaneously gaining support and elaboration from these neighbors. Scientific models and theories are

\(^7\) It is always a possibility that the problem of alcoholism is multifactorial, combining aspects of biological abnormality and weak willpower, poor upbringing, poor choices, etc. It is also possible that the biological factors predisposing some to alcoholism are not abnormalities but variations of normal biological function that happen, in certain environments, to be disadvantages for the individual in question.
human and social constructs—they are perspectival and fallible representations created to help us understand natural phenomena. They can be flawed, but they can also be wonderfully accurate and reveal to us surprising, previously unimagined aspects of the workings of nature. I shall argue, in chapter 2, that such representational models can have many different virtues (and vices) and that these various virtues come in degrees. Learning to recognize these virtues and evaluating for them in particular scientific models is something physicians and others do commonly. It is something that people can do better with practice. Such evaluation of the scientific models backing claims about disease is a crucial part of determining the legitimacy of these claims.

Another interesting theme that I shall repeatedly touch on has to do with the fact that biological science offers the most uncontroversial and convincing distinctions between normal and abnormal function for these isolated systems far removed from gross observation. The details of the normal and potentially abnormal functions and structure of the cells that make up the thyroid gland are not in dispute nor the target for claims that science is discriminating against those thyroid cells marked out as different. The issues are much more controversial when biological and psychological science distinguishes sexual behaviors as abnormal (e.g. paraphilias) or even when it claims that gross human structure is abnormal (e.g. congenitally conjoined twins or albinism).

This current chapter serves as an introduction to my thesis. Chapter 2 will give a brief sketch of my position on science, how it operates, what it produces, and how its products are tested, improved, and ultimately adopted as reliable and accurate knowledge regarding the phenomena being represented. The theoretical sense of normal and abnormal function (as distinct from other senses of normality and abnormality) is created simultaneously with other descriptive elements of particular biological models. This chapter contains the least original contribution on my part and borrows heavily from the work of others with whom I agree. Chapter 3 argues that there are different senses of normality and abnormality and in particular argues that modern biological science utilizes an importantly distinct sense of normality and abnormality in its descriptions of biological phenomena. It is important to be able to distinguish and keep track of different senses of normality and abnormality when considering controversial disease claims. Distinguishing different senses of abnormality provides insight into the allegation that
some disease claim is the ‘medicalization’ of a non-medical problem. It also provides insight into the allegations that some disease claims are merely ‘social constructions.’ Chapter 4 is the most important chapter. I discuss the dispute between naturalists and normativists about disease. I then offer ideal criteria for a legitimate disease claim, namely:

i. a specification of a circumscribed biological (or psychological) system
ii. a specification of the normal structure and normal function for this system (and/or its parts)
iii. a specification of a particular malfunction or malformation in the structure or function of the system (and/or its parts)
iv. these specifications are part of a scientific model (i.e. generated by those in the community recognizable as the authorities on the details of the biological or psychological system in question)
v. the scientific models involved are assumed or judged (by at least those recognized as authorities) to provide an accurate (i.e. having good ‘fit’) and relatively unbiased representation of how human biology or psychology normally functions and how it can in this particular way malfunction or be malformed.

I also discuss illegitimate and putative disease claims based on these criteria. In the last part of this long final chapter I apply my concept of disease, and my criteria for legitimate disease claims, to interesting, controversial, and difficult cases. I then conclude in chapter 5 with a brief summary.
2.1 INTRODUCTION

My main thesis is that a disease is an abnormality in the structure or function of a biological system. It is pathology in form or function. This is not a controversial understanding of disease among contemporary physicians. Furthermore, curing disease, along with reducing suffering, are the central goals of medical practice. One searches in vain, however, for a definition of disease in medical textbooks. During my own medical school education, a definition was never offered nor asked for. This assumption, that everyone has a clear enough idea what constitutes disease, is being challenged. Advancing technology and increased knowledge provides intervention and control of many biological processes, capabilities, behaviors and aspects of appearance. Many of these new biotechnological interventions involve managing concerns and addressing problems which traditionally have not been considered within the realm of medical practice. This creates an opportunity to ask if some issues that patients want medical help with are legitimate diseases. Is attention-deficit disorder in school children a legitimate disease or another sort of problem? Are the aches and pains of a depressed middle aged women normal or the signs of a disease like fibromyalgia? The wrinkles on an aging face or the acne on a young face are frequently problems brought to the medical clinic, but it

---

8 The controversy arises when one tries to carefully define disease and then considers examples to test the definition. It quickly becomes apparent that a set of necessary and sufficient conditions is not forthcoming if we want to capture all the examples of conditions currently called disease. The difficulty is compounded if we include historical examples.

9 An entire host of related questions may arise when one asks is this problem a medical problem or asks if the things troubling the patient constitute a ‘legitimate disease.’

10 When I have used the phrase ‘another sort of problem,’ people (particularly physicians) generally ask what I mean by this, as if diseases are the only real source of suffering or difficulty in life. I hope that our society is not so thoroughly medicalized that we cannot conceive of non-medical sorts of suffering and anguish, like a child being bullied at school or otherwise treated unfairly, or discovering that one’s spouse is having an affair. There are problems and difficulties that are not related to pathology or biological malfunction.
is not clear if they are pathologies. The increasing cost of providing an adequate level of medical care to most, if not all, members of society is forcing policy makers and medical providers to ask where the boundary between disease and non-disease lies. Simultaneously, pharmaceutical companies are increasing the number of products whose purpose is to improve the quality of life more than prevent death and disability. Presumably the boundary between legitimate disease and non-disease will serve as a starting point for discussions about what ought to be paid for by insurance and what sorts of services physicians should provide. What is needed is a way to distinguish legitimate claims of disease form illegitimate claims.

In addition to now being of some practical importance, the nature of disease is an intellectually interesting problem. It is one of the fundamental issues in the philosophy of medicine. Over the past 30 years, the main point of disagreement about the nature of disease has been cast as the disagreement between naturalists and normativists. Essentially, the normativist’s position is that disease claims are fundamentally evaluative in nature. Diseases are harmful or otherwise disvalued biological states. Saying that some biological process is a disease is much like saying that a certain animal or insect is a pest or vermin. It tells us more about our personal preferences and social practices than about the structure or character of natural phenomena. The main problem for normativism generally is that it undervalues or ignores issues regarding the differences in epistemic virtues of the explanatory biological models supporting particular disease claims. Naturalism is the position that disease claims are fundamentally descriptive, scientific claims. Diseases are biological malfunctions. Saying that a certain biological process or problem is a disease is like saying that a certain animal or insect is a member of a certain species. It tells us more about the natural biological phenomena than human preferences or values. The problem for naturalism is explaining the seeming teleology involved with designating something a disease. My own position, as will be detailed in Chapter 4, is something of a compromise, and might be called a modest or weak naturalism.

The current Chapter concerns a global sketch of science, what it produces, and how it does so. This is a very difficult task to set oneself, and I take the easy way out. I
rely heavily on the work of other philosophers of science with whom I agree broadly.\footnote{A disclaimer is warranted from the outset: I select only a few recent philosophers of science to draw on. However, these philosopher’s positions are often elaborations of earlier views and built on suggestions or ideas offered by previous workers in the field.} The reason for needing to give such a sketch is that one of the fundamental differences perpetuating the dispute between naturalists and normativists concerns their views of science, its products, and how these products are created. Basically naturalists tend to be pro-science and tend to defend an unrealistically positive view of science as delivering the bare facts about empirical phenomena. They frequently discuss cases of spectacular and uncontroversial scientific discovery and explanation, of which the history of medicine has many examples. Normativists tend to be very skeptical of science and the social and political authority it commands. They are critical of the epistemic claims offered by over-zealous defenders of science and its products. They frequently discuss cases where personal agendas and social biases were passed off as empirical facts supported by scientific investigation. Unfortunately, there are also many examples of this in the history of medicine. It is not too great an exaggeration to say that the simplicity of many of the positions offered on the nature of disease are related to the oversimplified and slanted visions of science on which they draw. Furthermore, my own explanation (or explication) of disease can be understood as an attempted compromise between naturalism and normativism, and my sketch of science also a compromise between the errors of over-simplistic praise or criticism. Science is a human endeavor, one open to bias and influence by the personal and social agendas or the people and communities pursuing empirical investigation, but it also has built up both technological and epistemic tools and practices, and institutionalized many of these, so that over time it tends to increase the accuracy and decrease the bias in its representational and explanatory models.

In this chapter I will speak of scientific models, and in particular I am interested in the explanatory models offered in biological science. I have a very broad conception of such models, and my meaning is close to what others may mean by ‘scientific theory.’ Scientific models are circumscribed representations of selected aspects of empirical phenomena. They are constructed by humans for use by humans. For instance, there are
models of how the human thyroid hormone has its effects on cells. For two excellent example see Figures 2 and 4 in Cheng, et. al., (2010). These figures are only simplified schematic representations of aspects of the overall model of the mechanisms allowing thyroid hormones to have effects on cells. Other aspects of the model are represented or described through text or via mathematical formulas. The ‘model’ is a complex combination of textual description, formulae, schema, etc. It is important to distinguish the simplified, generalized model of some biological system from any actual physical system that the model is representing. Models, like those involved with the thyroid gland, have been built up, tested and refined over generations. One can find the basic details of such models in textbooks dating back several generations. Other models are newer and perhaps less well verified and more likely to be substantially improved by continuing research, like those explaining how the nucleus accumbins forms part of the circuitry in the brain controlling feelings of reward and satisfaction (see Willuhn, et. al., 2010 and Gardner, 2011).

Models like these, of the function of human thyroid hormone, have very close interrelations with other models in this area of research. These groups of models lend each other mutual support. The scientific model of the function of thyroid hormone is closely integrated with models of how the hypothalamus works in regulating the thyroid and other glands in the body. It is integrated with models of how hormones are transported in plasma and interact with cell surface or intracellular hormone receptors (such as the interactions partially represented in Cheng, et. al., (2010), Figures 2 and 4). These are integrated with models of how hormones are metabolized by target tissues, etc. The more integrated a model becomes with larger collections of models, the harder it is to believe that the group of models, each worked on and constructed somewhat independently, and whose results dovetail with each other, could all be mistaken and in

---

12 This article and the figures that are in it are easily attainable online at no cost at either [http://edrv.endojournals.org/content/31/2/139.abstract](http://edrv.endojournals.org/content/31/2/139.abstract) or [http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2852208/?tool=pubmed](http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2852208/?tool=pubmed).

13 We can speak here of models or a model for any circumscribed system. Any model can usually be broken down into parts or component models, and any model can be combined with others to form a larger system. I freely change, for example, between talking of the models that describe the function and structure of the thyroid or of the model that describes this.
need of revision or replacement. Technology and medical intervention based on these models work as predicted. Scientists, physicians and students accept these abstract models as explanations for how individual, actual thyroid glands work and why they are located where they are and look the way they do, etc.

There is vertical integration as well as horizontal integration. The details of thyroid function are described in increasing depth. There are details of the normal pattern of arteries and veins bringing blood to and from the thyroid. There are wonderfully detailed models explaining the structure and function of thyroid tissue—specifying how follicular cells and C-cells are organized around follicles, or lakes, of stored thyroglobulin, and how such parenchymal tissue is organized by septae of connective tissue. Other more fine-grained models describe how these cells function to synthesize calcitonin and the two main thyroid hormones (T3 and T4). At lower levels of analysis there are models of how the enzymes in these particular cells are structured and can function to bind and utilize iodine in the synthesis of thyroid hormone, and how the concentration and activity of these enzymes are regulated by the interaction of local and systemic factors. At even lower levels of analysis are models detailing how genes and molecules interact to regulate expression and create and sustain the normal structure and functions of a human thyroid gland (as partially and simplistically represented in Cheng, et. al., (2010), Figure 2). It is granted that any actual token human thyroid gland may not operate exactly as the standard model details, but to a remarkable extent (that allows medical practice to be remarkably successful) the idealized model of normal function captures the important, reproducible aspects of the actual natural phenomena of any token thyroid.14

14 There are of course many complexities even with the idealized models that represent normal structure and function of the human thyroid. There is a description of how the thyroid develops embryologically, changes with growth and development, and how its function declines with age. There are also aspects of the model of normal thyroid function that explain how it operates under certain states of stress (like extreme illness or injury of some other part of the body, or during pregnancy, etc.). There are also aspects of the overall model of the human thyroid that take into consideration certain less significant differences in performance—differences within “the normal range of function.” Some amount of variety in actual function between token thyroid glands is not an insurmountable obstacle to a general model of normal thyroid form and function. The
The horizontally and vertically integrated set of models that explain the normal structure and function of the thyroid gland and its place in the larger human endocrine system can be considered a paradigm of an unproblematic, objective scientific explanation of normal biological function. Almost no one seriously worries that these models of ‘normal’ biological function are merely social values being passed off as factual so as to perpetuate some personal or political agenda. These models are assumed to be both an accurate representation of natural phenomena and an unbiased and objective description of normal biological function.

My main thesis is that legitimate claims of human disease are made in reference to unproblematic models of normal human biological function (e.g. the thyroid gland) along with models of the particular manner in which the system malfunctions or is malformed. A thyroglossal cyst (i.e. a remnant of thyroid tissue left in the midline of the anterior aspect of the neck along the path of migration of thyroid tissues from the base of the tongue to the area near the thyroid cartilage) is an abnormality and malformation and I would argue it is an unproblematic case of human pathology. The same is true for medullary carcinoma of the thyroid, follicular adenomas and carcinomas of the thyroid, papillary carcinoma of the thyroid, Hashimoto’s thyroiditis, DeQuervain’s thyroiditis, Graves’ disease, and for the various M.E.N (multiple endocrine neoplasia) syndromes that can be inherited, etc. These are human diseases because scientific models explain them as abnormalities of a biological system which can otherwise be explained as having a normal structure and function.

Let us turn to historical cases where claims about the existence of a human disease were made but now appear to be problematic. Appeals (either implicitly or explicitly) were made in these cases to scientific models intended to delineate abnormal from normal biological function or structure. Melancholia was a diagnosable disease of temperament for many centuries. It was basically either an acquired excess or inherited potential for excess of one of the four basic humors, specifically black bile. According to some versions of Galenic theory, black bile was produced in normal amounts by the spleen as part of the normal digestive process (see Garrison, 1929). Likewise, fever was issue of normality, normal function, and distinguishing insignificant variety from pathology will be discussed in Chapters 3 and 4.
also considered for many centuries to be itself a disease entity. Some models described fever as resulting from an abnormality in the normal balance of basic humors, namely an abnormal accumulation of yellow bile.\footnote{The excess of a particular bile in the body was usually due either to abnormally increased production or abnormalities in the means of excretion or conversion of bile. Here, the modern medical theorist is reminded of metabolic diseases such as gout.}

Drapetomania was a disease proposed in 1851 by Samuel Cartwright, a pro-slavery physician from Louisiana (see Cartwright, 1851). It was a theoretical mental illness that afflicted some southern slaves, and resulted in repeated attempts to run away from their masters. This disease was a result of improper care of slaves that arose out of a failure to properly understand the physiology and temperament of the African race and the psychology of the master-slave relationship. From our vantage point Cartwright’s disease claim was hardly more than an attempt to pass off his personal and political views as a scientific model of normal and abnormal behavior. Even Cartwright’s contemporaries must have seen the bias in his model. However, not all cases that we now recognize as illegitimate disease claims were so easily detected at the time they were proposed. Cartwright had studied with Benjamin Rush, one of the founding fathers of American psychiatry, a one-time chemistry professor at the University of Pennsylvania Medical School, a staunch abolitionist, and one of the most respected physicians of his day. Rush also described a disorder in African Americans that today we recognize as an illegitimate disease claim. At the time Rush believed he was observing evidence of a biological abnormality in a system that otherwise might operate normally. What Rush observed was a case of (what we now recognize as) vitiligo, an autoimmune disorder that results in patches of lost skin color. Rush believed what he was seeing was the partial cure of abnormally darkened skin. He proposed that darkly-completed persons must be suffering from a disease called ‘negroidism’ and for which a cure must be possible—after all, he was seeing evidence of its partial remission. He theorized that this skin disease was related to leprosy (a disease that was also known to cause hyperpigmentation), and urged a search for those factors that might cure it. Rush advised that those suffering from the disease not be treated as inferior, just because they suffered from a skin disorder.
However, he also recommended against mixed-marriage as children from such unions might help perpetuate the disease.

In hindsight, it is often easy to spot a putative disease claim backed with poor theoretical models of normal and abnormal function. Definitive rejection of such disease claims as illegitimate occurs as science slowly replaces the theoretical models such as those proposed by Rush with models offering more accurate representations of the phenomena being explained. Currently we possess much better models of the normal and abnormal functions of the biological systems responsible for skin pigmentation than Dr. Rush did. Today we understand more about how the skin acquires pigment and we have accurate (though still incomplete) scientific models of how the melanocyte-keratinocyte system works. We also have better (although not fully worked out models) of how autoimmunity destroys melanocytes in certain regions of skin in some individuals and gives rise to the clinical manifestations of vitiligo. It is easy to see now that drapetomania was a case of social prejudice being passed off as a descriptive theory of abnormality. However, even today the models that science offers concerning what constitutes ‘normal’ human behavior are not as well worked out, reliable or as uncontroversial as those for the human thyroid or the melanocyte-keratinocyte system.

As a community of inquirers, we are better positioned than Dr. Rush regarding whether changes in pigment are normal or abnormal biologically. However, we are not in any better position than Dr. Rush regarding current controversial claims of disease.\textsuperscript{16} Examples include chronic fatigue syndrome, attention deficit hyperactivity disorder, intermittent explosive disorder, pathological gambling, alcoholism, idiopathic short stature, obesity, pedophilia, etc. I will argue that whether these are legitimate diseases claims depends on whether or not these conditions can be explained as theoretical abnormalities of an otherwise normally operating and structured biological system. Importantly, legitimacy depends on whether the scientific models describing the system have the sorts of virtues that make them widely accepted as accurate and unbiased. Ideally, the scientific model backing a disease claim (and its corresponding claim about

\textsuperscript{16} It would be interesting to compare and contrast this episode with the more recent controversy over the publication of the book \textit{The Bell Curve} whose basic premise was that African Americans may be genetically less intelligent. In each case certain data are observed and a theoretical model of normal and abnormal biology is constructed.
normal human biology) would be known to be an objective representation of the natural phenomena in question with little or no human bias or mistaken interpretation involved, and would continue to be recognized as such by future generations of inquirers more enlightened and experienced than us.

The naturalism/normativism dispute that forms the core of the philosophical debate about the nature of disease hinges on whether claims of disease are naturalistic descriptions or social/personal evaluations. Questions about how scientific explanations are produced, exactly what is the nature of these explanations, and what kind of authority they should carry are never far from the surface in this debate. Stereotypical naturalists are overconfident about the products of science being unproblematic representations of the natural world and its character. For them science delivers the facts. For the stereotypical normativist, science itself is a complex of social practices and its models ultimately represent the perspectives, values, and pre-occupations of the community and people in question. Oversimplification in one’s view of science can lead to oversimplified positions on the nature of disease.

In this chapter I will sketch a vision of science as a complex human endeavor which attempts to understand and describe the natural phenomena around us and within us. I shall offer a sketch highlighting points important for understanding my main thesis of what distinguishes legitimate from illegitimate disease claims. Rather than a global acceptance or rejection of science, what is required to evaluate disease claims is the evaluation of individual cases of scientific explanation. Science, particularly biological science, has a decent track-record of producing accurate representations. However, as a human enterprise, bias can enter into attempts to describe and model the normal and abnormal functions of the human body. Science is not a purely rational process of producing true statements about the natural world nor merely a power play. It is complex and messy. It is difficult even to describe well. Alternatively it is not merely a means for the more powerful interests in society to perpetuate their position by clothing their subjective values in what appear as factual descriptions of the natural order.

Humans do assess the relative reliability of individual scientific theories and models. I do so on an almost daily basis as a medical student and physician. I frequently make assessments of models of biological function and dysfunction that support claims
about particular disease processes. Well-educated adults also make judgments on the accuracy and reliability of isolated scientific models specifying what constitutes normal human biology and behavior. Such models encountered when assessing or trying to understand claims about ADHD, depression, obesity, chronic fatigue syndrome, fibromyalgia, gulf-war syndrome, etc. must be evaluated for their reliability? How speculative are they? The factors relied on in such assessments are often not explicitly discussed nor sometimes even consciously acknowledged. As this chapter progresses I will try to highlight some of the factors or clues I think people should rely on when faced with determining whether a particular scientific model of biological normality and/or abnormality has the sort of qualities that make it likely to be reliable and accurate and make the related disease claims likely to be legitimate.

In many cases the scientific model in question is only a first speculative attempt to describe the functions and malfunctions of the system giving rise to the signs and symptoms suspected of being a disease. In these cases the answer is indeterminate and must await further work and more analysis. Often the observable signs and symptoms being described may be controversial, or already the subject of widely accepted conventional judgments. Whether masturbation, alcoholism, or an extremely large nose are signs or symptoms of a disease is controversial and any model of biological abnormality will compete and coexist with conventional judgments about the abnormality of these behaviors and attributes. In such cases the controversy is not merely about how accurate the scientific model is, but about whether science can or should replace, compete, or legitimate social judgments or naïve folk-theories about such signs and symptoms. Here, if the science being offered is only tentative or incomplete, the temptation will have to be fought to jump to the conclusion that an objective, descriptive divide between biological normality and abnormality has been established, particularly if the divide was previously just a conventional judgment about normality.

Ultimately the question of the legitimacy of the claim about disease is answered by assessing the accuracy and other epistemic virtues of the scientific model being appealed to in the disease claim. If the claim about disease cannot provide a reliable scientific model to back its case, then it is not yet a legitimate disease. If the model appealed to is not a legitimate and unproblematic scientific model, then it is not a
legitimate disease (although it may be a hypothesis or first approximation awaiting further research). This does not mean the physician cannot address symptoms and alleviate suffering in such indiscriminate cases, only that she may or may not be treating disease when doing so. This does not mean that the physician cannot address symptoms and alleviate suffering in such indeterminate cases, only that she may or may not be treating a disease when doing so.

2.2 GIERE

Ronald Giere (1985, 1999) argues for a ‘middle way’ between critics and defenders of science. He believes that both camps have internalized certain unsatisfactory assumptions of what science produces and how it does it. Giere calls this flawed vision the Enlightenment picture of science. Aspects of this picture include a belief in laws of nature (understood as universal generalizations), an a priori commitment to realism and naturalism, and a belief in a special kind of scientific rationality. Giere recommends moving beyond both criticizing and justifying this vision of science. Giere wants to retain the idea that scientific models and theories are representational, but move past the ideas that representation in science has to be linguistic in nature. “The winning combination, I suggest, is one that gives up the search for criteria of scientific rationality, abandons the attempt to separate the content and methods of science from the psychological and sociological reality, but preserves the view that science is a representational activity (Giere, 1999, p.44).”

17 A recent example of this Enlightenment picture and its assumptions can be found in much of the thinking of a school of thought Giere calls ‘Logical Empiricism.’ Giere says: “Within the English speaking world, the Logical Empiricist image of science, and the projects it generates, dominated philosophical though about science for a generation following World War II. Two fundamental aspects of this image are relevant here. First, scientific knowledge consists primarily of what is encapsulated in scientific theories, and theories are ideally to be thought of as interpreted axiomatic systems. It follows that the primary mode of representation in science is linguistic representation. Second, the reasoning which legitimates the claims of a particular theory as genuine knowledge has the general character of a logic. That is, there are rules which operate on linguistic entities, yielding a ‘conclusion’ or some other linguistic entity, such as a probability assignment (Giere, 1999, p. 119).”
For Giere, understanding how science works, or should work, does not require a special kind of rationality or logic of confirmation, and he calls this position ‘scientific judgment without rationality.’ He says:

Of course scientists make judgments regarding the fit between particular models and aspects of reality. And of course I do not claim that scientists’ judgments in these matters are ‘irrational.’ It is the conception of rationality that is at issue. The original Enlightenment dream was of universal, categorical principles of rationality that could guarantee the truth of natural laws meeting their conditions. By the twentieth century, this dream was generally reduced to a desire for universal principles that yielded the probable truth of natural laws. My view is that there are no such principles. One can have scientific judgment without rationality. Coming to hold that one model holds better than others is not a matter of pure reasoning or logical inference. Rather, it is a matter of making a decision. Effective decision making requires strategies for reaching desired goals. This applies to business and military decisions as well as to scientific decisions. If one wishes to talk of rationality here, it is a conditional or instrumental rationality, a matter of using effective means for reaching desired goals (Giere, 1999, p. 7).

Giere sees science as utilizing normal human cognitive abilities—cognitive capacities evolved for ‘mapping’ and manipulating our complex environment. Science proceeds by the same everyday human cognitive abilities involved in other complex social activities, and particularly, by developing and evolving special methods and practices that extend these ‘everyday’ cognitive abilities. Giere recommends an empirical study of how humans generally, and scientists in particular, go about creating scientific models and deciding which models to endorse, which to refine, which to abandon. He believes that science has already shown itself successful regarding such decisions, and has itself been developing the tools to study scientific decision making. “[W]e know that our capacities for operating in the world are highly adapted to that world. The skeptic asks us to set all this aside in favor of a project that denies our conclusion. And he does so on the basis of

---

18 Although Giere doesn’t explicitly discuss it, I believe he would want this empirical study to encompass both individual decision and the dynamic social phenomena involved in how groups of scientist, whether through formal institutions and practices or more informal means, shape which models are supported, sanctioned, and ultimately survive and which are modified, rejected, overlooked, etc. Later in this section I discuss Giere’s endorsement of building an explanatory model for ‘the scientific method’ utilizing evolution and selection as a metaphor instead of using a developmental stage metaphor. Surely such a process of scientific ‘evolution’ involves some of the dynamic social phenomena I have in mind.
what we claim to be an outmoded and mistaken theory about how knowledge is, in fact, acquired (Giere, 1985, p. 340).”

Even though Giere believes science does not use a special logic of decision, he still believes that science produces legitimate knowledge, and that this is knowledge of phenomena operating independent of our theorizing.

…there exists much genuine scientific knowledge. Moreover, I firmly believe there have been dramatic increases in scientific knowledge during the twentieth century... We have learned for example that inheritance is carried by DNA molecules with a two-strand helical structure. And even the continents have not always been where they now are. In stating these convictions I am not merely playing games with the meaning of expressions like ‘scientific knowledge.’ I intend such expressions in the relatively ordinary sense that scientific knowledge is knowledge of the world and that there is a difference between knowledge and mere opinion, even widespread opinion (Giere, 1999, p. 3).

Science is not merely the discovery of already categorized objects and relations. The categories we use are to some extent constructed by us. Nevertheless, scientists can sometimes legitimately claim genuine similarities between their logical constructs and aspects of reality. That makes me some kind of a realist rather than a social constructivist (Giere, 1999, p. 150).

Scientific models and theories do, more or less, represent aspects of reality. Disease categorization systems are constructed by humans but can with refinement represent aspects and patterns of the natural phenomena being represented. Giere calls to mind the American pragmatist school of thought, particularly on matters of science and naturalism, and in particular the work of John Dewey. “There was, for Dewey, no special sort of philosophical knowledge, particularly none that could provide any sort of foundation or ultimate legitimization for the sciences (Giere, 1999, p. 12).” Giere takes the current best science, its methods and results, as being paradigm examples of naturalistic explanation and knowledge.

Giere discusses naturalism as not only a commitment to avoid supernatural explanations and appeals, but also to avoid a priori ones. No transcendental or a priori arguments can be given for naturalism. This commitment to naturalism is more methodological then metaphysical, and Giere ultimately sees his own work as empirical.
One must be careful, however, how one characterizes naturalism. Presented as theses, it invites self-defeating attempts to construct transcendental arguments in its favor. Vigilance is required to keep arguments for naturalism within naturalistic limitations. A better strategy, I have suggested, is to focus on naturalism as a set of methodological rules for developing a comprehensive naturalistic picture of the world. Success in applying these rules gives comfort to those pursuing the program and encourages others to join the effort. Positively, that is the most a consistent naturalist can do (Giere, 1999, p. 83).

What the naturalist should be satisfied with is continuing to investigate the world by the best methods available, and building on the best models so far produced, knowing full well that these are fallible but also revisable. Belief that there is one, independent, describable world is one of many methodological assumptions that the naturalist perspective has operated under and should continue to do. But to argue over the truth or justification of these assumptions is a waste of time.

Another improvement over the Enlightenment picture is a more modest, but more sophisticated, realism. Giere believes that scientific models represent reality, but that the manner in which they represent needs to be reconceived. He describes his realism as constructive, perspectival, and a ‘realism without truth.’

[We should] reject the usefulness of the notion of truth in understanding scientific realism. I do not mean that we cannot use an everyday notion of truth, as when asserting that it is indeed true that the earth is round. Here truth may be understood as no more than a device for affirmation. Rather, it is the analysis of truth developed in the foundations of logic and mathematics, and used in formal semantics, that we should reject in our attempts to understand modern science. But if we reject the standard analyses of truth and reference, what resources have we left with which to formulate claims of realism for science? The answer is that the notion of linguistic truth is but one form of the more general notion of representation. What realism requires is only that our theories well represent the world, not that they be true in some technical sense. So we need a notion of representation for science that does not rest on the usual analyses of truth for linguistic entities. What might that be? A first step is to reject the analysis of scientific theories as sets of statements in favor of a model-based account which makes nonlinguistic models the main vehicles for representing the world, and places language in a supporting role. We may, of course, use language to characterize our models, and what we say of the models is true. But this is merely the truth of definition, and requires little analysis. The important representational relationship is something like fit between model and the world. Unlike truth, fit is a more quantitative relationship, as clothes may be said to fit a person more or
less well. Of course we can say it is true that the clothes fit, but this is again merely the everyday use of the notion of truth. Here I can offer no general analysis of the notion of fit, only a further analogy—maps. There are many different kinds of maps: road maps, topological maps, subway maps, plate maps, etc. And it can hardly be denied that maps do generally represent at least some aspects of the world... Maps have many of the representational virtues we need for understanding how scientists represent the world. There is no such thing as a universal map. Neither does it make sense to question whether a map is true or false. The representational virtues of maps are different. A map may, for example, be more or less accurate, more or less detailed, of smaller or larger scale. Maps require a large background of human convention for their production and use. Without such they are no more than lines on paper (Giere, 1999, p. 25).

The proceeding passage contains many points about scientific models and the idea that a notion of better and worse ‘fit’ should replace the issue of truth when it comes to the virtue of representational accuracy. Scientific models represent phenomena better or worse relative to the explanatory goal for which the model was constructed. Different models can be constructed for different aspects of a phenomenon and eventually they might be combined or otherwise enhance each other, just as different maps of the same terrain might each focus on different salient aspects and, combined, give an even richer account of the landscape or cityscape being represented. The metaphysics of representation no longer need consist in discrete objects grouped into sets and kinds by sharing properties and relationships specified by logical conditions. The real phenomena may be very complex and have properties that vary subtly and continuously, with the simplistic structure and relations of the model being an attempt to capture important and interesting patterns and correlations in the phenomena. Whether a model represents accurately is an empirical question.  

Giere says: “…[I]t is an empirical question whether a particular map successfully represents the intended terrain. If it does, we can reasonably claim a form of realism for the relationship between the map and the terrain mapped. I call this form of realism perspectival realism. Standard analyses of reference and truth suggest a metaphysics in which the domain of interest consists of discrete objects grouped into sets defined by necessary and sufficient conditions. Likewise, there is a metaphysics suggested by perspectival realism. Rather than thinking of the world as packaged into sets of objects sharing definite properties, perspectival realism presents it as highly complex and exhibiting many qualities that at least appear to vary continuously. One might then construct maps that depict this world from various perspectives. In such a world, even a fairly successful realistic science might well contain individual concepts and relationships
Perhaps only implicit in Giere’s vision is the point that when individuals dispute or question the details concerning some aspect of natural phenomena, they are most directly thinking of the available models. Models necessarily simplify and only partially represent the phenomena. These more simplistic representations are most often what the individual in question has learned and thinks of as the facts of the matter regarding the phenomena in question. When asking whether cardiac muscle has the same or different lactate dehydrogenase isoenzymes as skeletal muscle, most people will call to mind the details of the model as presented in a textbook, and to settle a dispute will go to such a text or other repository of expert opinion. Whether the particular model has good ‘fit’ is less often at issue. And even if it were at issue, outside a specialized laboratory set up to investigate such matters, verifying the accuracy of representation is not a possibility open to the average person. Again, the best that most people can do is consult authoritative texts and experts about how such things were discovered, how they are verified, and how such knowledge is reliably utilized. The same is true for claims about the details of normal and abnormal function of any biological system that might be disputed in discussion of a particular disease. The disputants will consult the details of the current, most authoritative model to determine what is ‘true’ of the model. The issue of whether the model accurately represents reality and if so, how well, are much harder and more complex issues and are not settled so easily. However, science as a community and even individuals often face just such issues of assessing ‘fit.’ More empirical investigation and careful analysis about just how such assessments are made, and how such assessment should be made, is needed.\textsuperscript{20}

Models can be nonlinguistic representations. Examples might include physical scale models, simplistic diagrammatic representations, textbook descriptions, or idealized formula explaining the relations of variables under specific circumstances. Some are complex highly detailed sets of interrelated models, such as those explaining the operation of the human cardiovascular system or the manner in which intracellular

\textsuperscript{20} I will return to this topic later in this chapter.
trafficking of proteins is accomplished, regulated, or disrupted. Models are whose structural elements are more or less similar to aspects of objects and processes in the real world.\textsuperscript{21} Science builds representational models, but the manner in which they represent is a matter of better and worse, and depends on the task at hand, and to some degree on the cultural values at hand.\textsuperscript{22} There is not one proper way to represent the world.

\textsuperscript{21} About representational scientific models and the idea of ‘fit’ Giere says: “One is tempted to ask: How do maps represent physical spaces? Asking the question this way suggests that the answer is to be found in some binary relationship between maps and places. A better question is: How do we humans manage to use maps to represent physical spaces? This way of posing the question makes it less easy to forget that making maps is a cognitive and social activity of humans. Part of the answer is that map-making and map-using takes advantage of similarities in spatial structure between features of a map and features of a terrain. But one cannot understand map-making solely in terms of abstract, geometrical relationships. Interpretive relationships are also necessary. One must be able to understand that a particular area on a map is intended to represent, for example, a neighborhood rather than a political division or corporate ownership. These two features of representation using maps, similarity of structure and of interpretation, carry over to an understanding of how humans use scientific models to represent aspects of the world… Here, then, is a suggestion for better understanding what it means for a model to fit the world. The fit between a model and the world may be thought of like the fit between a map and the region it represents. How further to understand that sort of fit is, unfortunately, not an easy question (Giere, 1999, p. 82).” Giere also discusses the possible distinction between hypothesis, theory and model, and advocates trying to stay as true to actual usage by practicing scientists as possible. “Claims about real systems, then, have the form: the real system is similar to the model. A pendulum with small amplitude, for example, is similar to a simple harmonic oscillator. I will call such claims ‘hypotheses.’ …The typical advanced text, then, presents the student with a cluster of models (really a cluster of clusters) together with a number of hypotheses about real things claimed to be similar to one or another of the models. For the purposes of developing a naturalistic theory of science, I suggest we understand the word ‘theory’ as indicating both the cluster of models and a broad range of hypotheses utilizing these models. Restricting ‘theory’ either to the models or to the hypotheses produces too great a variance with how scientists use the term. For all sorts of reasons, it is best to stick as closely to scientific usage as is compatible with developing an overall, adequate theory of science (Giere, 1999, p. 168)”

\textsuperscript{22} Giere says: “The question for a model is how well it ‘fits’ various real-world systems one is trying to represent. One can admit that no model fits the world perfectly in all respects while insisting that, for specified real-world systems, some models clearly fit better than others. The better fitting models may represent more aspects of the real world or fit some aspects more accurately, or both. In any case, ‘fit’ is not simply a relationship between a model and the world. It requires a specification of which aspects of the world are important to represent and, for those aspects, how close a fit is desirable. In this
Scientific knowledge, at any given time, consists in a structured set of the best models available and these models provide only a limited representation of some aspects of the complex, continuously variable qualities of the natural phenomena in question. There are grey areas at the frontiers of research where the ‘fit’ between model and the phenomena being modeled has not been well assessed yet. The combined biomedical sciences have built up a set of models that in sum give an idealized model of normal biological function and form of the typical human body. These models also include detailed accounts about various possible malfunction and malformation of these systems.

Giere acknowledges that interests, cultural values, and bias definitely influence model selection and the interpretation of theory, etc. “I think we will just have to live with the empirical possibility that, at any given time, our best science may nevertheless embody all manner of cultural interests and values in their very content. …I also believe that particular interests embedded in specific theories can be identified, and sometimes eliminated, by creating empirically superior theories (Giere, 1999, p. 18).” Any model will always be incomplete and partial—it will always necessarily be a simplification. This is the best we can do, but this sort of underdetermination or lack of completeness doesn’t yield an unacceptable relativism. Models are still tied to the reality they represent via observations and experiment and are always open to revision and supplementation. Investigators interact with the world, and their awareness that others can also interact with the world and test their model, and that actions and risks might be taken on the basis of their work—all these and other factors help constrain the content of scientific models. But creativity as well as cultural and personal bias also certainly influence the content of picture of science, the primary representational relationship is between individual models and particular real systems... (Giere, 1999, p. 93)” He also says: “The empirical question—the question of realism—is how well the resulting model fits the intended aspects of the real world. And here my central claim is that the fit is always partial and imperfect. There is no such thing as a perfect model, complete in all details. That does not, however, prevent models from providing us with deep and useful insights into the workings of the natural world (Giere, 1999, p. 6).”
some models. Such influences can be detected and the best policy is careful investigation of each particular case.\(^{23}\)

Giere adopts an evolutionary metaphor in his model of both the development of scientific disciplines and in explaining how progress occurs (i.e. the selection of better models). He says of scientific progress:

> There are several important negative lessons to be learned from the past thirty-five years of debate about ‘scientific revolutions.’ One is that we should abandon the political metaphor now embodied in the very term ‘revolution.’ Thinking of scientific changes this way makes them seem more social, and more arbitrary, than they in fact are. It unnecessarily encourages the idea that scientific facts might be just like social facts. In science, there is not only interaction among individuals and social groups, there is also causal interaction with the world. Although it cannot be claimed that this causal interaction uniquely determines the scientist’s pictures of the world, it does play a major role in the story. The task is to explain that role, not to ignore it or deny its existence. A second negative lesson is finally to abandon the developmental metaphor that underlies both Kuhn’s stage theory of science and its recent philosophical rivals. …Stage theories take a biological concept that applies primarily to the development of individual organisms and attempts to apply it to social groups. Taking an evolutionary analogy more seriously provides an immediate explanation of why developmental models do not work very well at the collective level. The variety of possibilities for evolution in a population is very great… Rather than trying to understand ‘scientific revolutions,’ we should be thinking in terms of ‘the evolution of science (Giere, 1999, p. 45).

The evolution of specific scientific fields or disciplines (or even science as a whole) can progress by selecting and refining the better models and rejecting and repairing worse ones—with part of this process being the detection and minimization of cultural, political or personal bias in the models. However, evolution in this sense does not guarantee moving forward towards increasing numbers of better and better models. Science, or one of its disciplines, can stagnate or actually regress in the sense of having increased

\(^{23}\) Giere says: “The main disagreement would simply be over the extent to which decisions about the fit of models to the world are influenced by interests rather than other factors, such as the outcomes of experiments. My view is that this balance varies from case to case and the actual influences can only be ascertained by examining the particular scientific episodes in considerable detail. In some cases experimental data may strongly influence theory choice; in other cases political commitments or professional interests might be dominant. Most cases are mixed (Giere, 1999, p. 61).”
amounts of cultural and personal bias influence models or undergoing a weakening of the pressures to be accurate in representation on individual investigators (either formal ones or less formal, more conventional pressures). At certain times and places, communities of investigators have suffered periods of such stagnation or regression. Scientific inquiry, however, often does make slow progress and does tend to select for better models over time. Such progress slowly ‘evolves’ and is achieved by certain pressures that emerge from the collective inquiry, examination, and competition within a community of inquirers. There is no guarantee that things will continue to progress. Stagnation and regression (from whatever perspective this is judged) are possibilities on the evolutionary model of scientific development. Contingency about which direction development takes, and the inertia of any path once chosen are also issues to consider on an evolutionary model.

Giere is also interested in families of models and theories, and their overall structure. He suggests that the structure of many families of models may be importantly similar to the hierarchical and webbed relations hypothesized by some psychologists to organize natural kind concepts. “Families of models may be ‘mapped’ as an array with ‘horizontal’ graded structures, multiple hierarchical ‘vertical’ structures, and local ‘radial’ structures. These structures offer the promise of important implications for understanding how scientific theories are learned and used in actual scientific practice (Giere, 1999, p. 117).”

The structure of sets of models can change across disciplines. Giere comments that many advanced textbooks for particular specialties present ‘clusters of clusters’ of models. I would call certain of these clusters the core of these disciplines, and suggest that different disciplines have a core cluster of models with different characteristics. This includes differences both in the qualities of the models that are within the core and differences in the interrelations between the models in the core. Giere seems to recognize that different disciplines have core clusters with very different qualities and interrelations.

---

24 The Lysenko affair and the atmosphere of respect for evidence within the scientific community in the early communist period in Russia is an example of such a period of regression. The success of eugenics in scientific and political communities in pre-WWII United States might be another example.
And these differences may require very different ways of understanding and studying different scientific disciplines or projects.

The model-based account of theories permits a theory of science which avoids one of the major methodological defects of much of social science—the demand for universality. If the empiricist model does not fit molecular biology, that does not mean it is worthless as part of a theory of science. The question is whether there are any major sciences, or long periods in the life of some major sciences, that fit the empiricist model [as described by van Fraassen and other strict empiricists]. It seems hard to deny that there are. Greek astronomy, thermodynamics in the late nineteenth century, and quantum theory in the twentieth century are obvious candidates. It may be more than coincidence that quantum physics is the science van Fraassen knows best. On the other hand, many contemporary sciences, including chemistry, molecular biology, and geology, seem decidedly realistic. Of course, many disagreements remain. For example, is empiricism or realism the dominant mode in contemporary science? I would guess that realism is dominant. Empiricism is the mode of sciences lacking a solid theoretical tradition, e.g., many of the social sciences, or of the sciences whose models are difficult to understand in spite of much empirical success, e.g., quantum theory. Another issue is whether having realistic goals tends to make scientists, or even science fields, more successful in discovering new results. Again, I would suggest that realism is scientifically more fruitful. Though difficult to resolve, these issues, happily, are more empirical than theoretical (Giere, 1999, p. 198).

Disciplines within natural and social science develop very differently regarding their methodologies and regarding the organization, success, and the characteristics and interrelations of their best models. This point is worth keeping in mind when considering scientific models that purport to distinguish normal structure and function from abnormal. The models that form the core of the basic biological sciences, the majority of what is reviewed in the first two years of medical school, usually deliver this distinction without any controversy. No one disputes that the normal function of the thyroid gland is to produce thyroid hormone. However, the psychological and behavioral sciences that are reviewed in the first two years of medical school are not as uncontroversial. Even the best available models of the normal sexual behavior of a human or the normal personality traits of a human (or even the range of normal personality traits) are not subjects accepted as obvious facts, even by medical students who are being presented the information as if it were on par with the models of the normal human thyroid. Part of the reason is that the
sciences of psychology and psychiatry have developed differently, had differing degrees and kinds of success, and have never fully developed stable core methodologies and models to the extent the other biological sciences have. Even non-experts in these fields have an intuitive hunch that the models of human psychology do not provide a description of normal human function as reliable as those of endocrinology. Part of the intuitive hunch probably has to do with the perceived reputation of these different fields, and part of the intuitive hunch comes from the fact that human behavior is observable, open to folk-theories and assessment via conventional norms and values and thus the explanation of such behavior has to compete with these in ways that the operation of the thyroid gland or immune system do not.

I agree with most of Giere’s alternative to the Enlightenment conceptions of science. I think that he is correct that scientific models are partial, perspectival, and often involve substantial non-linguistic and non-truth-theoretic means of representation. Something like a notion of better or worse ‘fit’ should replace the all-or-nothing assessment of true or false. Models are attempts to represent phenomena and those that are accurate are constrained by observation and experiments with the phenomena in question. I agree with Giere that science utilizes and extends general human cognitive abilities to map, represent, and model. Science uses pragmatic decision making. The rationality of science, if it can be generally described at all, is one of instrumentalism. This is not to deny that certain statistical methods for evaluating and weighing evidence have not been cultivated and encouraged within scientific communities.

I agree with Giere’s suggestion to discard the ‘political revolution’ metaphor and adopt the evolutionary metaphor regarding how science and its different branches develop over time. One issue that I believe Giere does not address adequately is the pressure that can be exerted by communities of investigators to retain and refine the better models while abandoning or correcting those that are inaccurate, poorly conceived,

---

25 Even within the non-psychological biological sciences there are important differences. For example many models in physiology are old and have been passed down over several generations, where as immunology is a relatively young science whose core methodologies and models have only recently (in the past 20 years) become stable. Students understand that the models of biological function being offered in physiology or anatomy compared with immunology are more reliable and unlikely to change much over the course of their career in medicine.
etc. I do believe that in the right circumstances there are such positive pressures and that part of what constitutes ‘science’ (and distinguishes it from other social institutions) is that, as a community of inquirers, certain pressures, methodological assumptions, techniques, etc. are perpetuated and attempts are made to verify, correct, adjust and in general increase the fit and accuracy of scientific models over time. How these pressures get balanced with opposing ones, such as individual career aspirations, political climate and pressures, etc. and how ‘science’ (hopefully) is able, over long periods of time, to self correct, are interesting and important topics. Certainly part of this discussion will involve how certain fields have at places and times done better or worse than other fields and how periods of doing a poor job regarding these matters can sometimes cause problems for future development in such fields. Another part of this discussion would be to try and determine what aspects of certain scientific projects or fields of study make them more susceptible to bias or to lack of regard for representational accuracy. This sort of investigation could shed light on whether disease claims from fields such as psychology and sociology should be treated with greater suspicion than disease claims backed by scientific models from fields such as physiology.

2.3 HAACK

Susan Haack has also presented a view about the nature of science that seeks a middle ground between extreme positions. Haack believes that science has had success explaining how the natural world works—it has produced legitimate knowledge.\(^{26}\) However, the way that science actually succeeds is complex, messy, and heterogeneous. Haack claims, there is no special epistemological strategy employed by successful scientists. There is not a special scientific method per se, just excellent and refined use of

\(^{26}\) Haack says: “The natural sciences have accumulated a vast and ever-growing body of knowledge about the natural world and how it works, patchy and incomplete to be sure, but increasingly well anchored in experience and increasingly integrated internally. …plenty of the knowledge scientific work has produced is trivial, boring, unimportant even from a scientific point of view. Nevertheless, some of the knowledge the natural sciences have produced is stunning; and the way it all fits together is extraordinary. In fact, the body of scientific knowledge accumulated even thus far is so extensive, and its internal interconnectedness so dense and complex, that I am a little at a loss to know how to illustrate the point at modest length (Haack, 2003, p. 301).”
the same cognitive skills that make for any good investigation or honest attempt to understand. She says:

The core standards of good evidence and well-conducted inquiry are not internal to the sciences, but common to empirical inquiry of every kind. In judging where science has succeeded and where it has failed, in what areas and at what times it has done better and worse, we are appealing to the standards by which we judge the solidity of empirical beliefs, or the rigor and thoroughness of empirical inquiry, generally. Often, to be sure, only a specialist can judge the weight of evidence or the thoroughness of precautions against experiential error, etc.; for such judgments require a broad and detailed knowledge of background theory, and a familiarity with technical vocabulary, not easily available to the lay person. Nevertheless, respect for evidence, care in weighing it, and persistence in seeking it out, so far from being exclusively scientific desiderata, are standards by which we judge all inquirers, detectives, historians, investigative journalists, etc. as well as scientists (Haack, 2003, p. 21).

Haack points out that part of what has made the sciences epistemologically distinguished is the development of ‘helps’ that extend our senses and our ability to gather evidence, stretch our imagination, and help increase respect for seeking out and carefully weighing evidence. Such ‘helps’ include experimental instruments, but also things like metaphors, statistical techniques, peer reviewed journals, etc. Furthermore, the success of science, with increased use of specialized tools and specialized vocabulary, make weighing the evidence difficult for the non-expert even though the underlying virtues of inquiry are not distinctly scientific. Specialized tools, methods and vocabulary have allowed investigations into areas that, on their face, seem anything but everyday empirical investigations. Consider experiments using the large hadron collider near Geneva. Nonetheless, science does not necessarily require a special epistemology to evaluate or understand the knowledge it produces.

Although Haack denies that the epistemology of scientific beliefs is different from other sorts of empirical reasoning, she is a career epistemologist and offers a theory of degrees of warrant based on the evidence available. For Haack, warrant is an objective relationship based on certain qualities (and the degrees of such qualities) of the evidence in question. By objective she means only that it is independent of whether anybody and/or everybody believes it to be warranted. Although the relation of a claim to evidence is objective, the judgment of an individual or group about such warrant is
perspectival and limited and depends on background beliefs specific to the context.

Haack explains her position as follows:

Complex and diffuse as it is, evidence is a real constraint on science. And though the degree of warrant of a claim at a time depends on the quality of some person’s or some group’s evidence at that time, the quality of evidence is not subjective or community-relative, but objective. However, it doesn’t follow that the objectivity of evidential quality is transparent to us. In fact, judgments of the quality of evidence depend on the background beliefs of the person making the judgment; they are perspectival. …When there are serious differences in background beliefs between one group of scientists and another, there will be disagreement even about what evidence is relevant to what, and about what constitutes an explanation—disagreements that will be resolved only if and when the underlying questions are resolved (or which may, as Max Plank famously observed, just fade away as the supporters of one side to the dispute retire or die off). What has been taken for paradigm-relativity of evidential quality is a kind of epistemological illusion; again as in the graphology example, whether evidence is relevant, whether this is a good explanation of that, how strong or weak this evidence really is, how well or poorly warranted this claim actually is, is an objective matter. Sometimes scientists know that they don’t have all the evidence relevant to a question; and sometimes they have a pretty shrewd idea what the evidence is that they need but don’t have. But sometimes, given the evidence they have, they may be unable to judge, or may misjudge, whether or what additional evidence is needed. They can’t always know what it is that they don’t know; they may not, at a given time, even have the vocabulary to ask the questions answers to which would be relevant evidence. Nor can they always envision alternative hypotheses which, if they did occur to them, would prompt them to revise their estimates of the supportiveness of their evidence. And so on. Since evidential quality is not transparent, and scientists can only do the best they can do, a scientist may be reasonable in giving a claim a degree of credence which is disproportionate to the real, objective quality of his evidence, if the real quality is inaccessible to him. Reasonableness, so understood, is perspectival (Haack, 2003, p. 77).

Haack is distinguishing the objective relationships that establish warranted explanation, and which are determined by the quantity and qualities of the available evidence. An expert in a field may evaluate and weigh this evidence and determine just how warranted an empirical explanation or model is at a current point in time. This would be the subjective (and perspectival) assessment of one expert. However, non-experts also might try to assess the value of the evidence and produce their own subjective judgment. Haack mentions some clues that might be used regarding such subjective assessments of the qualities of evidence for a particular explanation. Haack mentions, among others, degree
of supportiveness, degree of explanatory integration, comprehensiveness, security and independence, etc. I shall leave it to the interested reader to look into the details of Haack’s theory. Basically, individual scientists, as well as average citizens, are more or less aware of the sorts of qualities that make for more accurate, more virtuous models, but their perspective is limited and sometimes their interests are not purely focused on honest inquiry and accurate explanation. For any individual confronted with a model or ‘fact’ delivered as part of scientific research, there will be some process of evaluation that takes place. The intuitive process of evaluation will balance many factors, and for the scientist who is actively working in the research area in question, the process will involve very subtle points concerning the actual data available, the competing explanatory models available, and also knowledge of the reputation and honesty of the group producing the current model, etc. For those non-experts who are being asked to learn about the model or use it for a practical application, or even those needing to make policy decisions upon which the model’s findings are relevant, the evaluation will not be as subtle, and will involve trust of expert opinion. But even for such non-experts, reliance on the authority of scientists and definitive textbooks in the field are not the only factors available. Common sense intuitions about good explanation, relations to other tested and reliable models, etc. can be used by the non-expert. Furthermore, the ultimate evaluative process occurs over longer stretches of time, is carried out by a complex community of inquirers and the process stretches across space and time.

For Haack, certain qualities make certain explanations better. Scope, breadth of explanatory power, the capacity to unify apparently diverse phenomena along with positing specific causal mechanisms when possible, all, when properly combined, make for better explanations. Looking over the history of explaining the link between smoking and lung cancer Haack points out how the qualities of the explanation have improved over time. “Now there is a unifying explanation, combining specificity and scope in exactly the right way: an account of the damage caused by cigarette smoke, firmly embedded in our knowledge of the physiology of the cell and molecular biology, and tightly interwoven with what we know about heredity, evolution, etc. The present evidence is much more supportive of the claim than earlier evidence was (Haack, 2003, p. 131).”
Haack makes much use of a particular analogy to illustrate her points about the nature of scientific discovery, change and progress. She asks the reader to think about the process of working on a crossword puzzle that already has some entries filled in. Some of these previous entries are in areas well worked out and in which many of the entries are believed to be correct, while other areas of the puzzle are tentative and await further work and development.

Like crossword entries, reasons ramify in all directions. How reasonable a crossword entry is depends on how well it is supported by clues and any already-completed intersecting entries; on how reasonable those other entries are, independent of the entry in question; and on how much of the crossword has been completed. Similarly, what makes evidence stronger or weaker, a claim more or less warranted, depends on how supportive the evidence is; on how secure it is, independent of the claim in question; and on how much of the relevant evidence it includes. While judgments of evidential quality are perspectival, dependent on background beliefs about, for instance, what evidence is relevant to what, evidential quality itself is objective... Scientific claims are better and worse warranted, and there is a large grey area where opinions may reasonably differ about whether a claim is yet sufficiently warranted to put in the textbooks, or should be subjected to further tests, assessed more carefully relative to an alternative, or what. There can no more be rules for when a theory should be accepted and when rejected than there could be rules for when to ink in a

Haack says: “Picture a scientist as working on part of an enormous crossword puzzle: making an informed guess about some entry, checking and double-checking its fit with the clue and already-completed intersecting entries, of those with their clues and yet other entries, weighing the likelihood that some of them might be mistaken, trying new entries in light of this one, and so on. Much of the crossword is blank, but many entries are already completed, some in almost-indelible ink, some in regular ink, some in pencil, some heavily, some faintly. …In some areas many long entries are firmly inked in, in others few or none. Some entries were completed hundred of years ago by scientists long dead, some only last week. …Rival teams squabble over some entries, penciled or inked in and then rubbed out, perhaps in a dozen languages and a score of times… Someone claims to notice a detail in this or that clue that no one else has seen… From time to time accusations are heard of altered clues or blacked-out spaces. Sometimes there are complaints from those working on one part of the puzzle that their view of what’s going on in some other part is blocked. Now and then a long entry, intersecting with numerous others which intersect with numerous others, gets erased by a gang of young turks insisting that the whole of this area of the puzzle must be re-worked…I don’t mean to fob you off with a metaphor instead of an argument. But I do mean my word-picture to suggest, what I believe is true, that scientific inquiry is far messier, far less tidy, than the Old Deferentialists imagined; and yet far more constrained by the demands of evidence than the New Cynics dream (Haack, 2003, p. 93).”
crossword entry and when to rub it out; “the” best procedure is for different scientists, some bolder, some more cautious, to proceed differently (Haack, 2003, p. 24).

For the experts working on filling in parts of the puzzle, or for individuals merely learning or utilizing the entries already placed, there is the issue of how to assess the reliability of a particular entry. Some areas have been extensively filled in and the entries here have not changed much for long periods and have had many intersecting entries build up on top of them, while other areas are new and experimental. There are clues one can use to try and assess the reliability and representational accuracy of scientific models, and depending on which area of the puzzle one selects there may be more or less clues. When all else fails, then either the consensus of experts must be trusted, or a healthy skepticism adopted for the time being. This is the situation faced by anyone seeking to assess the legitimacy of a particular disease claim—such a person will have to assess the legitimacy and reliability of the specifications of abnormality and normality of biological function and structure found in the scientific models backing the disease attribution.

The crossword puzzle analogy also serves to reiterate that the progress of science is not simple or linear. Certain areas or inquiry experience periods of success and establish an entrenched core of models that are taken to be reliable representations of the phenomena being explained. Other areas do not achieve such rapid or sustained success. These areas tend to undergo periods of revision or ‘paradigm shift’ as new methodologies are adopted or new models displace older ways of thinking. When assessing a putative disease claim an individual (or even a panel of experts) will rely on the reputation and past track record of a field of inquiry. This is part of the reason why certain disease claims concerning behavior or personality are usually more controversial and considered less reliable (and more open to accusations of social construction) than those backed by models from scientific fields with entrenched models and methods, like physiology.

Although not a major point, Haack does discuss some of the issues surrounding the question of what to make of the behavioral and social sciences. Haack sees no reason why the study of behavior and social phenomena cannot be done in a way that embodies the respect for evidence and other values she believes constitute any good scientific inquiry. Furthermore, Haack sees the division between social and natural sciences as
artificial. However, she also concedes that there appears to have been less progress or success in the social sciences. “Why don’t the intentional social sciences seem to have made anything like the impressive progress of the natural sciences? For a host of reasons, among them that the ideal of respect for evidence is even harder to achieve in social-scientific than in natural-scientific inquiry, and that borrowing mathematical and other methodological ‘helps’ from physics in hopes of looking ‘scientific’ has sometimes proven counter-productive (Haack, 2003, p. 152).”

There are other reasons too. To stick with the crossword puzzle metaphor, the social sciences don’t have as many areas of previously well-worked out entries to build upon. Haack also points to the preoccupation in the social sciences with methodological matters, and this often covers-up the over-simplification of the concepts and categorization schemes being used. The categorizations in the natural sciences tend to be more robust. The more that human behavior is involved, the less well clustered and recurring are the properties being cited and the harder it is to pick out robustly recurring patterns characteristic of the phenomena being modeled. Social kinds are often very loose congeries of properties whose structure and organization does not reoccur with the frequency or generality that would make meaningful categorization easier. “The looseness of social kinds, and the local, contingent character of social institutions, is the source of some notorious pitfalls of social-scientific inquiry: taking the local and culturally specific… for something universal and inevitable; assuming whatever is true of one variant of a social kind… is bound to hold for other forms as well (Haack, 2003, p. 165).” Haack also points out that social phenomena are harder to study because social phenomena often involve intentional states like beliefs and desires. Haack is critical of the sociology of science also.

---

28 I will discuss some problems with categorizing people later in this chapter.
29 Haack believes there is a role for the sociology of science in studying scientific inquiry, but she is critical of much of the current work in the field. Radical sociologists of science often run together two kinds of relativity. An anthropological (or empirical) relativism claims that different communities and cultures have different epistemic standards. A philosophical (or conceptual) issue of relativity claims that it makes sense to talk of justification (or better or worse warrant) only relative to some culture. But a philosophical relativism, making objective cases of knowledge impossible, does not follow from the empirical fact of anthropological relativism. Besides this slip in
I agree with Haack that science uses souped-up versions of the same basic epistemological criteria, and often assesses the same virtues and vices, as does any empirical investigation or inquiry. Science, where it has been successful, is set apart mostly in the fact that it has a very impressive track record and that it has retained, refined and perpetuated specialized methods of inquiry, evaluation, and explanation peculiarly well suited to the disciplines or areas of study in question. I tend to think there could be objective relations between models and the appropriate evidence along the lines that Haack suggests, but I am also skeptical that we could have anything better than a good empirical model of what these might be, and that this model would be open to refinement and changes, and these relations would probably turn out to be context dependent. Judgments about how well supported a model is by the available evidence will always be perspectival and subjective as will judgments about such judgments. Basically I am less enthusiastic than Haack about the possibility of specifying just what are the objective relations that constitute being well supported by the evidence, although I do believe that with attention and practice people become very good at making judgments that do assess such relations, and other factors that go into claiming that a model is an accurate representation and is well supported by evidence. Such individuals constitute experts, but even non-experts regarding some field of inquiry can develop a sense of what makes for more and less reliable explanatory models in science. At the very least they might use clues that safely identify clear cases on either side of the uncertain middle reasoning, Haack finds too many thinkers in sociology of science succumbing to what she calls “the passes-for fallacy.” She says: “Perhaps it is because the social sciences are especially susceptible to the influence of political prejudice and bias that sociologists of science seem especially susceptible to a dreadful argument ubiquitous among New Cynics—the “Passes-for Fallacy: what passes for, i.e., what has been accepted by scientists as, known fact or objective evidence or honest inquiry, etc., has sometimes turned out to be no such thing; therefore the notions of known fact, objective evidence, honest inquiry etc., are ideological humbug. The premise is true; but the conclusion obviously doesn’t follow. Indeed, this argument is not only fallacious, but also self-undermining: for if the conclusion were true, the premise could not be a known fact for which objective evidence has been discovered by honest inquiry (Haack, 2003, p. 28).” Thus while conceding that the sociological study of science can be done well and yield useful knowledge, Haack concludes that too much current work in that discipline is conceptually sloppy and has unfortunate assumptions at its foundations.
ground. Also, I disagree with Haack concerning the need to continue talking about ‘truth’ and ‘natural laws,’ and agree with Giere that these are misleading.

Haack’s account is also lacking sufficient discussion of the practices and pressures that develop within the community of scientific inquirers (whether this is large scale or is on a smaller scale with investigators working on some narrowly defined problem). These are the pressures that somehow, over periods of time, select for models that represent more accurately, select for circumstances were respect for evidence and respect for honest inquiry are kept high. Ideally such pressures would also tend over time to discourage the production of models that are grossly inaccurate or biased, or are even mere formalization of some dogma. These are the pressures that would operate at differing levels in the evolutionary metaphor that Giere discussed as a model explaining the diversity of scientific developments and its tendency for progress. These emergent pressures and the procedures that communities of inquirers develop to try and keep inquiry honest and to ensure the quality of the explanations and models produced—these are components of what constitute ‘the scientific method’ or ‘the scientific spirit’ of inquiry for some particular community at some time and place.

I find most of Haack’s crossword puzzle analogy useful, particularly the ideas that science works piecemeal, that both the objective evidence and subjective confidence backing each model comes in degrees of better and worse. Judgments of such evidence depends not only on the data or clues from experiment, but also other minor clues including the reputation of the researchers producing the model, the reputation of the field producing the model, and how the model integrates with other models regarded as reliable and accurate. Success often breeds more success, and a core of useful models and useful methodologies can become entrenched. Other areas are more tentative, and/or more isolated, at any one time, form the most successful areas of research. I also tend to agree with Haack’s ideas about why social sciences have appeared to achieve less success than some of the natural sciences. Such differences in the reputations and perceived track records of success for different areas of science explain some of why psychological and sociological models that offer descriptions of normal and abnormal human biological
function and form are usually not as uncontroversial as models in molecular biology or physiology.\(^{30}\)

2.4 BROWN

Ted Brown, in a book entitled *Making Truth: Metaphors in Science*, agrees that to understand what scientists do, and the nature of the knowledge that they create, one must come to see that scientific thinking is inherently metaphorical. Much of the knowledge that scientists learn when being educated and the concepts and knowledge they use when observing, forming hypotheses, experimenting, and communicating their ideas, etc., involves metaphorical understandings of the phenomena in question. Metaphor has been neglected as an issue of investigation in traditional philosophy of science. Brown encourages those interested in science to pay more attention to the metaphorical nature of its models and the type of cognitive skills it takes to create and manipulate these models. He says:

…[M]etaphorical reasoning is at the very core of what scientists do when they design experiments, make discoveries, formulate theories and models, and describe their results to others—in short, when they do science and communicate about it. Metaphor is a tool of great conceptual power. It enables the scientist to interpret the natural world in wonderful and provocative ways… My intent in this book is to show that metaphor is essential to every aspect of science. It lies at the heart of what we think of as creative science: the interactive coupling between model, theory, and observation that characterizes the formation and testing of hypotheses and theories. None of the scientist’s brilliant ideas for new experiments, no inspired interpretations of observations, nor any communications of those ideas and results to others occur without the use of metaphor (Brown, 2003, p. 15).

\(^{30}\) There are other problems as well, and in the next chapter I shall discuss the difficulty that arises when science attempts to model behavior or sociological phenomena. Often these phenomena are open to non-scientific, non-controlled observation (in ways that the physiology of the thyroid or the details of cellular traffic of glycosolated proteins is not) and thus individuals and communities will develop folk-theories, norms and conventional ideas about what is normal and what abnormal for such observable functionality. The interaction, competition, or even the possible attempted merger of these conventional ideas with more formal scientific models purporting to describe what is normal and abnormal for humans is a major source of difficulty.
But science, as Brown sees it, is not special in this—most cognizing about abstract and complex theoretical matters involves the use of metaphors. The use of metaphor is particularly important to the kind of models that attempt to draw distinctions between normal and abnormal function. Specifically metaphors are often chosen that allow a rough usable notion of normal dynamics as well as suggests possible ways that a system might be perturbed.\textsuperscript{31}

When Brown speaks of metaphor, he intends a manner of conceptualizing, not merely a linguistic devise. In particular, metaphorical understandings are involved in the grasping of abstract concepts, such as love, time, inflation, etc. He cites Lakoff and the idea that we give content to such abstract notions by understanding them in terms of more familiar embodied experiences of the observable, macroscopic physical world. Humans understand new aspects of phenomena in terms of bodily and social experiences that become ingrained from interacting with the social and natural world on a daily basis. Another source of metaphors becomes available when certain areas of science achieve sustained success. Metaphors and structural models at work in the successful areas can be borrowed in part or in whole to model newer phenomena in other areas. For example, during the late nineteenth century and into the twentieth, the theoretical hypothesis of ‘hormones’ as chemical messages that communicate the need to initiate or terminate certain biological processes was developed. Later theoretical systems and actions for specific hormones were proposed and tokens were eventually isolated, first adrenal hormones and then pancreatic ones. The basic metaphors (i.e. messengers, keys, and locks) involved with creating the model for the concept of hormones has been borrowed many times in the twentieth century for activity within cells and at even more distant levels of analysis.

This borrowing of conceptions from one source in order to describe and organize other phenomena, is the sense of ‘metaphorical’ at issue. In the sciences, such a means of conceptualizing is especially important for describing and organizing phenomena and systems not grossly or directly observable. This is usually because our observational

\textsuperscript{31} For example, conceptualizing the cell membrane as a porous wall, or a boarder between countries, imports ideas about its normal structure and function and also ideas about how it might malfunction.
capacities are limited in various ways—the phenomena are too small, too large, or too complex, etc. Brown suggests thinking about the use of metaphorical conceptualizations of natural phenomena as mappings or borrowings from a source domain to a target domain.

In the process of such mappings a model is created for the phenomena in question by organizing the observations or data in question based on structure and/or the qualities of the source domain. The creation of a model is constrained by the evidence and data available, and also by what is already well known about related phenomena, but the scientist has some room to be creative in selecting a target domain by which to give order and structure to the observations. The normal function and structure of certain proteins manufactured in certain cells is modeled using the metaphor of ‘channels’ within ‘walls.’ The metaphorical model accounts for the observations in a way that makes the phenomena capable of being understood and questioned—i.e. capable of being mentally manipulated—in ways that are productive of further speculations, and possible experimental tests. It colors how further evidence is understood. It also suggests certain ways in which malfunction or malformation may occur. Brown illustrates the basic idea with the example of a ‘channel’ proteins that allow passage of ions or other molecules across lipid membranes:

During a long period in which many studies of channels were carried out, scientists could not see channels in cell walls, even with high-powered electron microscopes. The observations that formed the target domain included data on the rates at which ions pass into or out of cells under various conditions and correlations with the presence in the cell walls of various proteins that are thought to form the “walls” of the channels. These and other observations are complemented by models of what the cell wall proteins are like in terms of shape and other properties. Scientists then constructed a model of ion passage that captures as many characteristics of the observation domain as possible. A successful model must have structure, and relationships between its parts must map onto the corresponding elements in the observation domain. The model is a way for us to understand the observation domain in terms of more directly accessible experience in the macroscopic, everyday world. When we give the name ‘channel’ to the model, the word is being used metaphorically. The model itself is also metaphorical; it maps the properties of channels as we know them in the macroscopic domain onto a microscopic entity to which we do not have the same level of access as far as sensory data are concerned. Note that it is not just because the “channels” in cell walls are tiny and thus not directly visible that we
have a metaphorical connection. It is true, as we will see in later chapters, that our
only access to the structure of matter at the atomic and molecular levels is via
models that are themselves metaphorical. Thus, we do not have access to
something we could call a literal representation of a channel in a cell wall. But,
more fundamentally, the use of “channel” here is metaphorical in the sense that
we use our literal knowledge and experience of channel in the macroscopic world
to characterize not only structures but also functions and relationships between
entities in the microscopic domain (Brown, 2003, p. 21).

For Brown the typical spark of creativity involves selecting a particularly
productive metaphor that highlights crucial similarities. Naïve metaphors eventually
develop into what are considered respectable models as more structure and detail are
specified and as observational data tend to enrich and verify the usefulness of the
developing model. Other scientists realize the strengths of the model and eventually add
to its development, sometimes by criticism. Eventually quite specific hypotheses and
predictions about experimental results and future observations can be made. The details
of the model can be made more formal and mathematical equations can often be used to
describe the structure and relations in the model.

…a particular channel model may have the features that ions of the presumed size
and charge of calcium ions move preferentially through the channel under the
influence of an electrical potential. This feature of the model maps onto
observations of calcium ion transport into and out of [actual] cells. The model and
the story of how it matches, or accounts for, the observed measurements of ion
movement in and out of cells constitutes a theory of ion transport. The theory
might be made quantitative with mathematics. If it were, the terms of the
mathematical expressions, such as diffusion rates, channel diameter, and
interactions between charged species would match elements of the model. The
values of adjustable terms would be chosen to afford the best match of theory
prediction with experiment. In other words, the mathematical representation,
which some would call a theory [instead of a model] of ion transport across
membranes, is not really distinct from the model; it is an expression of it in
mathematical language. The theory is just as thoroughly metaphorical as the
model (Brown, 2003, p. 29, my emphasis).

So, even as a creative vision moves into a more structured attempt to account for the data,
and possibly into mathematical or other formal descriptions of relations and dynamics,
the evolving models remain metaphorical in nature.
This conception of scientific models highlights many important features of scientific activity and knowledge. Models (and the metaphorical structure that they rely upon) are always partial explanations. “Elements of the model map onto selected aspects of the observation domain. They are never complete descriptions of reality. In practice, no one model is sufficient to capture all the various observations that might be made. Thus, for example, we use distinctly different atomic metaphors to account for the physical and chemical attributes of matter (Brown, 2003, p. 73).” In particle physics differing metaphors, such as particulate versus wave-like, are often simultaneously deployed about the same phenomenon. In biology the heart can function within several metaphorical models—it can be a ‘pump’ in one model, or a ‘gland’ producing hormone signals in another model.32

Metaphorical models, like linguistic metaphors, are flexible and multivalent—always open to reinterpretation or recycling. Metaphorical models develop and evolve over time. Most models are often of isolated phenomena but can be combined with others to create more elaborate models. Eventually very complex sets of models for the same and related phenomena can be combined into even larger metaphorical schemes, and this is illustrated very well by examples in molecular biology, such as the cell as a city with production lines, regular consumption, policing of traffic, control of possible invaders, contingency plans for suspected invasion, import, export, communications and coordination with neighbors, remodeling, decay, etc. Metaphors are needed to create a descriptive framework in which model building and data interpretation can take place productively—they operate, says Brown, in many of the ways that Kuhn stressed were important features of paradigms. Even data or observations, especially if interpreted in light of prevailing metaphors, are rarely if ever purely literal statements of the properties in question.

32 The example with the heart here shows that the heart can be conceived of as part of multiple interrelated systems, each one represented in an accurate scientific model and each one assigning a different normal function to the heart within that system. What is ‘the’ function of the heart? It depends on how one isolates a particular biological system in order to represent the overall normal functioning of that isolated system. Different models may include the heart, and within different systems the heart may play different roles.
As the phenomena under consideration becomes more complex and especially when there are numerous diverse elements yielding emergent properties not easily described by simplistic mechanical or other familiar physical interactions, the metaphors needed to organize and structure explanatory models are often taken from social experience. Brown writes of ‘experiential gestalts,’ to designate these more complex, richer metaphorical schemas that contain numerous element and relations (often including as parts many simpler metaphors of the embodied and experiential variety). Just one of the examples in Brown’s book is the use of the complex metaphorical schema of human language to organize and structure models of DNA directed protein synthesis. Another example is the metaphors used to conceptualize the structure and dynamic functioning of cells:

Understanding the whole in terms of the multitude of parts can be achieved only if we have a way of conceptualizing the whole. The many individual sets of information can make sense only in terms of a mental model of the cell as an organized entity. For such models the scientist draws on experience with multifarious entities in the macroscopic world. The cell has inputs and outputs, in the form of substances that diffuse through the cell’s outer wall. It has budgets of various kinds; for example, the amount of matter entering and leaving must be kept in balance. Energy inputs in the form of food substances that provide energy sources must balance the cellular processes that consume energy. Metaphorical representations that reflect these cellular properties are drawn from the social domain, in which many agents with differing characteristics and goals go about

---

33 Brown says: “To conceptualize complex systems composed of multiple parts that interact with one another, with interdependences based on transports of matter, chemical conversions, and energy consumption and production, the scientist must draw on experiences in the macroscopic world that have those characteristics. Those experiences occur in the social domains of life (Brown, 2003, p. 158).”

34 Brown says: “The gestalts that consist of the complex of associations and ideas that make up our understanding and use of written language maps onto the molecular domain of protein sequence. Notice that this mapping does not involve a directly emergent physical experience but rather a human artifact in the social domain. This is an early example of an important and interesting aspect of metaphors in science: As the scientist attempts to understand systems of increasing complexity, metaphors based solely on embodied physical experience no longer suffice. In more complex systems, typically there are interactions between the components of the system. To deal with these interactions, which can be thought of as ‘transactions’ between components, the appropriate mappings increasingly derive from social constructs, with their attendant greater complexities (Brown, 2003, p. 126).”
their business, sometimes in competition, sometimes cooperatively, creating in the process a stable entity (Brown, 2003, p. 149).

The cell is conceived as a city with a complex, vibrant economy. The idea of normal functional capacities and tasks as well as ideas for malfunction and mistakes are often suggested by the metaphors.

One important feature to notice about the import of ‘experiential gestalts’ from the social domain is that it is one source of the transfer of teleological understandings and explanations into the sciences, particularly biology. Discussions of cells as factories or cities, the heart as a pump, the endomembrane system (i.e. the Golgi, endoplasmic reticulum, lysosomes, membrane receptor coated pits, etc.) as a manufacturing complex and import and export transportation hub, all involve just this sort of teleology. Paying attention to the metaphors involved in such models makes it easier to understand where some teleological thinking comes from and why. Even as scientists often realize that such descriptions and attributions are not literal representations of the phenomena, they are nevertheless indispensable for understanding and communicating the kind of science in question.

In a time in which biology has become relentlessly reductionist from an experimental perspective, metaphor has become a common tool in understanding observational data and communicating about them… Of course, no one literally ascribes purpose and human-like intent to molecules and cells. Teleological ascriptions of the kind exemplified here are simply metaphorical ways of conceptualizing complex systems in terms that correspond to human experiences. The important point is that such metaphors are a necessary part of the scientist’s understanding of the world under study (Brown, 2003, p. 158).

These sorts of metaphorical models lead easily to the ‘as-if-designed’ approach to investigating and understanding complex organic entities and their activity. This approach explains much about how functional and teleological explanation gets a foothold in scientific explanation. What Brown highlights, that is often overlooked, is that such teleology is widespread, important, and a necessary part of the content of models in the so-called ‘hard sciences’ just as much as in the social, psychological or the so-called ‘soft sciences.’ The reputation of various fields does not correlate with the use of metaphor or teleology. If anything, what attracts attention to the metaphors in some
social sciences is that they tend to undergo widespread change more frequently than, for example, in biological sciences.

Brown’s description of science is one based on how scientists actually work and communicate their results. He has no doubt about the existence of a reality that is structured and for which we can construct reliable knowledge. Scientific models are anchored to reality by their relation to experimental observations and confirmatory predictions. His concern over traditional philosophical debates about the nature of scientific knowledge, truth and realism is mild. The models that science creates, and refines are constructs, and they are a product, ultimately, of a community of scientists. However, they are the best we can do given the cognitive and sensory capacities that evolution has provided. He says this about truth: “In this way of looking at things, truth is the product of human reasoning. It follows that science does not proceed by discovering preexisting truths about the world. Rather, it consists in observing the world and formulating truths about it. …much of what we regard as scientific truth is metaphorical representation. This does not mean that science is capable of yielding only subjective results of uncertain reliability (Brown, 2003, p. 51).” One important feature that underlies the ability to have intersubjectively relevant representations, in addition to honestly produced observation and data, is that we all develop largely the same set of embodied, implicit knowledge of the physical and social worlds that we grow up in and occupy, and this is the common source upon which scientist (and non-scientists) draw to help understand, model, and explain more difficult-to-interact with phenomena.

I agree with Brown the metaphor is crucial to understanding the creation and success of many scientific models. Models are necessarily simplifications and partial, perspectival representations of aspects of the natural world. The observations of the macroscopic natural world and of the complex social, political and human world provide very rich sources from which to borrow organizing ideas to model the dynamics of phenomena harder to observe. Concerning my thesis on the nature of disease claims, Brown’s vision of science highlights that it is humans that attempt to understand patterns and structure within natural phenomena, by the best means at their disposal. The scientific models that specify biological normality and abnormality, and back disease claims, are in this sense social constructions. However, this is not to concede that they are
not also legitimate examples of empirical knowledge (i.e. accurate representations). In chapter 3 (on claims of normality in science) I shall examine the claim that the products of science are social constructions.

2.5 A BROAD SKETCH OF SCIENCE: COMBINING SOME OF THE POINTS MADE SO FAR

Science is a human endeavor and employs the same basic cognitive features that a person might use to investigate and describe any phenomena in their surroundings. It can be flawed for many of the same reasons a criminal investigation might be flawed, but the possibility of getting it right (more or less) also exists. Science has become a specialized type of inquiry due to the tools it has developed allowing phenomena to be isolated, controlled, and data to be collected even when the phenomena are distant from everyday human perception. It has also developed or incorporated special tools and procedures for analyzing data and evaluating its own models. Likewise a culture of respect for honest inquiry and a system of rewards and penalties has emerged and is perpetuated amongst inquirers. There are a series of pressures that the community of inquirers exerts on itself, and over time this has allowed better and more accurate models to be sifted from those that were flawed or inaccurate. At any one time, the products of science are of unequal reliability and accuracy, and furthermore, the processes which the scientific community uses to sort out the better from the worse often take time to yield results. Nonetheless, the results (i.e. current best scientific models) must often be assessed and relied upon by non-experts. For example, both physicians and grade school officials currently must assess and work with the scientific models that set the boundary between the normal and abnormal functioning of the neural systems that allow for concentration and attention. Presumably malfunction of these systems gives rise to the symptoms of ADHD.

In previous philosophical work on the concept of disease, the assumptions about the nature of science were often simplistic. A significant part of this oversimplification has come from the desire to situate scientific knowledge into larger epistemological projects. It is worth trying to move past the positivist vision of science as delivering a structured set of true statements about the physical world, but also recognizing that this
does not mean that scientific theory is totally adrift—merely interpretation without any mooring to natural phenomena.

My own position is that science is complex and delivers products of varying accuracy, and objectivity\(^{35}\) and at any one time it may not be possible to adequately assess all of the models that science currently offers. The implication for the status of the disease categorization scheme used by physicians is that it too is also a work in progress. Some diseases are supported by accurate scientific models accepted as reliable, objective and unproblematic by the community in question and for which the possibility of major revision seems unlikely. Historically we can also look back and see that the science involved in certain disease claims was clearly flawed or inadequate. In hindsight we realize these claims rested on flawed models of biological phenomena and were illegitimate disease claims. The most interesting cases, however, are disease claims supported by models that are obviously works in progress. Often such claims are based only on a few clues available about the malfunction or malformation involved, and even less is known about the normal functioning of the system in question. Schizophrenia might be an example. The behavior and cognitive function of people diagnosed with schizophrenia is clearly taken to be abnormal by most societies, even when the explanation involves demonic or spiritual possession, etc. Current work has yielded evidence that the dopamine neurotransmitter system of the brain is malfunctioning. But the scientific models of how this system works, and what its normal functions are, are still being worked out. For medical students and physicians and even for non-experts, the available model is accepted as somewhat reliable even while also understanding that future research could change our view of what is going on and change our view of the real nature of the malfunction involved. In such cases, where the details of the biological system in question are less clear, it is easier to imagine how new evidence or a better model might substantially change our view of the nature of the disease. When dealing with such putative disease claims, policy makers, physicians, and average citizens must use intuition and also a great deal of trust when it comes to deciding the accuracy of

---

\(^{35}\) By objective I just mean that the matter might be settled by investigation of the phenomena in question (not merely investigation of the communities views about the subject—i.e. that the possibility exists that the entire community could be wrong and the answer discovered eventually).
science’s claims about biological normality and abnormality. Concerns about the ‘legitimacy’ of such claims about biological abnormality are possible, especially from those skeptical or suspicious of science at the outset.

In some instances the best that can be done is to remain skeptical and await further developments. Historically, some communities have decided to ‘legitimize’ claims when caution would have been better. At any point in time, for any given community, there will exist a number of putative disease claims that cannot be accurately assessed and for which future inquiry and scientific work will be needed to establish or deny ‘legitimacy.’ But the existence of these putative cases does not necessarily call into question all scientific models of biological normality and abnormality.

Although I do not endorse the specifics of Haack’s theory of warrant, I do think her point about remembering the difference between subjective judgments of warrant versus objective relations between evidence is a good one. Legitimacy for a disease claim is similar. Models possess certain properties having to do with their accuracy and reliability as representations of natural phenomena. However, what users of such models deal with at any one time are there best judgments concerning the qualities of the model in question. If these scientific models back up claims about the legitimacy of a disease, then the legitimacy is directly related to the quality of the scientific models backing the claim. Legitimacy in this sense is at any moment going to be subjective, although, it too can be based on a set of objective relations.

A disease is a biological malfunction. A legitimate disease claim involves a set of signs or symptoms (or at least some detectable effects) explainable in a model as arising from the malfunction of some circumscribed biological system that otherwise has a normal function. My position transfers the burden of legitimacy onto the scientific model backing up the claim. A legitimate disease claim is one backed by good scientific explanation. This explains the obvious ‘legitimate’ cases (e.g. Hashimoto’s thyroiditis) while also explaining why many controversial historical cases are not ‘legitimate.’ However, a significant feature of my position is that it allows many cases of putative disease claims for which there is not currently a well worked out scientific model backing the claim, or the model does not carry the virtues needed to be accepted as an unproblematic explanation of the phenomena. My position does not provide the solution
to these controversies. However, it does suggest how they need to be resolved. Science needs to be allowed to build up the descriptive model of the phenomena to the point that the descriptions within the scientific models regarding normal and abnormal function are taken to be uncontroversial by the community. Virtuous scientific models yielding uncontroversial descriptions of normal and abnormal biological form and function allow legitimate disease claims based on such models.

2.5.1 Fit Is an Important Objective Quality That Makes Some Models Better

I believe something like the notion of ‘fit’, suggested by Giere, is the appropriate virtue to seek in scientific models being assessed for ‘objectivity’ and accuracy in representation. This quality is one of better or worse—a quantitative value—and not a dichotomous evaluation like true or false. Assuming that ‘fit’ could be quantified in some intercomparable units, models might be ordered as to better and worse fit. A model’s ‘fit’ is as an objective relationship (or set of relationships) that hold between a model and the phenomena in question, which is to say that it is possible that someone’s (or everyone’s) subjective assessment might be mistaken regarding the ‘fit’ some model has to the phenomena in question. The relationship of ‘fit’ holds (or fails to) independent of assessment of the relationship. I doubt, however, there is anything like a God’s eye view from which to accurately assess such objective relationships and the best we have are the evaluations, at any one moment in time, of careful and thoughtful individuals and groups. I believe individuals can and sometimes do make subjective evaluations of a models ‘fit’ that approximates the objective relation.

36 The notion of fit may actually be a combination of other relationships that hold between the model and the phenomena in question, and not a singular relationship in any simplistic way. However, for my thesis about disease, even if ‘fit’ is a shorthand for a more complex set of relation, it is specific enough.
37 I believe that ‘fit’ is the epistemological virtue of most importance regarding scientific models. Models are intended to represent accurately—that is their purpose and the reason scientific models are held by most to be knowledge about the physical world. I also believe this is ultimately the virtue most often being referred to when people claim a particular model is good or better than another.
38 For reasons beyond the scope of this essay I am skeptical such an intercomparable assessment and ordering could be made.
Even though fit is best determined over time by a complex selection process that I will discuss in the next section, individuals must assess scientific models over shorter periods of time. To assess whether a new model ‘fits’ the phenomena in question is probably rarely done by anyone except experts in a field of study, and even then consensus of committees or a group of experts is probably sought. This assessment would require expert knowledge of the phenomena in question, as well as related types of phenomena, and would require knowledge of competing models, the latest experimental data regarding the phenomena, the potential difficulties with past models and the unanswered questions most in need of explanation by any model of the phenomena in question. But even assessing older models—those for which the selective pressures of the ‘scientific method’ have been refining and verifying—is not an easy assessment. Such models would still have to be tested in some way in order to directly address the issue of fit to the phenomena in question. There are certain clues, however, about which models might have better fit that can be used by both experts and non-experts outside a specialized laboratory. Some examples of the kind of clues I have in mind would be whether or not the model has been around for a long length of time, and survived being tested with newer experimental equipment, and continued to make sense as newer data was gathered. This might be called a record of verification and survival. The model might integrate well with other models both within its own field and even with those in other fields. We could call this coherence or mutual support. If a model made predictions about future data or about the character of other models, and if the model’s predictions and description allowed successful technology to be implemented, this might be a clue to its representational accuracy. We could call this reliability. The more that a model was adopted by others, especially those outside the field of origin, outside the country or even the culture of origin—the more useful and accurate the model was found to be by diverse individuals and across space and even across generations—provides more evidence that its structure and details had represented certain aspects of the phenomena accurately. This along with directly surviving and overcoming challenges about being biased or limited in perspective, might be clues that the model was unbiased and had elements of accurate representation. A model might be more or less biased depending on relationships that hold between the model and certain cultural views held by those constructing or
interpreting the model. Interestingly, it might be the case the a model is simultaneously both biased based on its relations to prevailing cultural attitudes, but also a model with good fit based on its relation to the phenomena it explains. Current models of the genetic factors effecting human cognitive performance (i.e. intelligence) may be an example.

A model with more explanatory scope and capacity to explain multiple, previously unrelated phenomena with an elegant underlying causal mechanism will be attractive and intuitively seem likely to be verified and retained, especially assuming that the prior work it integrates and unifies is also held to be accurate. Another clue is that the model is complex and multifaceted enough to explain a phenomenon from multiple perspectives, or one that has multiple and diverse applications in technology. A model that is testable or makes testable predictions or claims can reasonably be assumed to be one that will be verified or rejected eventually. There are surely many other clues that are used in making evaluations of the relative reliability and accuracy of a scientific model.

These are only a few possible elements that might be clues used by people (in lieu of direct verification by experiment and interaction with the phenomena being represented) when assessing whether some model has a good ‘fit’ with the phenomena being modeled. Such assessments would be subjective, might be made with ‘gut intuition,’ and with limited perspective, especially if made by non-experts. But people are forced to make such assessments constantly. Better empirical sociological models are needed concerning how both experts and non-experts make such decisions concerning which scientific models to trust and which they decide to be wary of. As mentioned, the process of survival and selection of certain models over time, and the rejection or revision of others, is likely to be more accurate than the assessment, even of experts, given at a single moment in time. This process of selection will be the next topic.

2.5.2 The Scientific Process

Part of what people mean when they use the honorific term ‘science,’ is a body of models that are judged to have achieved a sufficient degree of ‘fit.’ ‘Science’ is sometimes considered to be the body of accurate representations about the physical, biological, and social world. However, ‘science’ can also refer to the spirit of inquiry and the process by which such models are developed, disseminated, critiqued, refined and
sometimes rejected. This evaluative process tends over time to select for those models with greater fit and select against those with poor fit. Such a selection process often occurs over large periods of time and the evolutionary metaphor suggested by Giere is insightful. Like evolution, such complex pressures emerge from the dynamic interaction of many individuals and the research ‘environment’ they operate within. The interactions influence the trajectory of research. However, unlike evolution via natural selection, the process of model selection can be greatly influenced by the good or bad intentions of individuals or groups, and by better and worse guidance from human regulators. It can be and is managed.

Another metaphor that offers insight into the selection of certain models over others is the selection process for successful businesses in a modern market economy. Many models may co-exist and develop, with occasional direct competition. There is also a certain level of external regulation and supervision of the activity of those monitoring the market. Both direct pressures, set by regulators, and emergent pressures, created by the market process itself, select for certain businesses over others. Over time, if the system is well managed, businesses are selected for having more efficiency regarding delivery of desired goods and services to discriminating buyers. Scientific models are selected for providing sufficient explanation of aspects of certain phenomena. When there is disagreement or competition the selective pressures for better explanations become particularly strong and models are more carefully scrutinized. Science makes progress by a complex process of many investigators working towards better explanations. It is the emergent community pressures and restraints as well as emergent incentives that help keep ‘science’ as a community of inquirers committed to accurate and honest explanation. Ideally these positive emergent pressures, as well as some amount of overall regulation and policing, help minimize the influences of cultural and political agendas on scientific explanation. Just as with business, at different times and places things may work better and worse as far as the propagation of the factors favoring the selection of the better from the worse. Also like business, science tends to preserve those techniques, methods, and arrangements that have yielded good results in the past, while also valuing and rewarding innovation.
It is important to reiterate that these pressures and constraints that help sift the better from the worse are a result of science being a highly social and competitive endeavor. A model that has survived much criticism and has survived the trial of time and the pressures of continued inquiry, and particularly one that also resides within the core set of models of a well established line of research, has a kind of authority—not the traditional appeal to some particular expert who gives his or her opinion, but rather the approval of the entire community that drives inquiry and perpetuates scientific knowledge. Science is a human activity. Scientific explanations are more like everyday reasoning, investigation, and explanation than they are like the formally reconstructed logical systems sometimes offered by philosophers to explain how science, at its best, possibly could and should operate. There is no special scientific method in the sense of a special logic or decision procedure. Scientific practice and progress emerge from creative human insights and constructs and from the inherently social aspects of scientific research, and not from any particular methods employed alike by all individuals engaging in ‘science.’ This is one area where the sociologists and historians of science have much insight to offer to the epistemologists of science. And it is an area where the pragmatist school of thought on science often has more insight than the positivist camp.

Science has become a special and honorific type of inquiry because of its ability to build on and to institutionalize behavior that leads over time to success and to the building of accurate and useful models. Peer review is one example. However, peer review cannot become too burdensome and should not be used as a means of protecting the status quo. This too must be balanced and continuously refined. Hopefully it will remain a positive force in the self-correcting system that has worked so well in developed societies for the past few hundred years. The pressures mentioned here and the level of institutionalization of the best practices and procedures will be different across disciplines and across cultures and research groups. Therefore the emergent properties in question, those properties that over time help guide the retention of the best scientific work, will also differ across time, from place to place, and between disciplines.

Fit should be the virtue that is sought in models of natural phenomena. The complex processes that allow the selection of models with better fit takes time—it is slow and not an orderly process. Even though it can be managed, just as in the case of large
economies, the management is difficult and often involves good fortune as much as careful calculation. Other virtues may also be selected for in certain times and places. Certain communities may value things other than the accuracy of representation. The Lysenko affair in Soviet Russia is an example of management (or mismanagement) of the scientific system such that qualities other than representational accuracy were sought.

2.5.3 The Evolution and Divergence of Scientific Disciplines: Social and Psychological Sciences

In this section I will give a general sketch of some factors involved in the development of different fields of scientific inquiry. This development produces fields of inquiry with different qualities. Specifically I am interested in differences between the psychological sciences and the more traditional biological sciences. The reason for interest is the issue of whether diseases in psychiatry are the same as diseases in the more traditional fields of medicine. Are disease claims backed by psychological sciences on the whole less legitimate than those for the biological sciences? I do not think they are. The claims are not fundamentally different. However, the qualities of certain scientific fields compared to others can influence the controversy or legitimacy a disease claim has. My sketch will be broad and brief.

I have suggested a model of scientific progress involving selective pressures and corrective mechanisms that emerge from the complex interaction of a community of researchers. At different times and places the conditions influencing selection are different, and the extent and attempts at management of the system are varied. Historically certain areas of exploration achieve success, and this success breeds more success and greater activity. The fields of anatomy in the sixteenth century, physiology in the nineteenth century, and molecular biology in the second half of the twentieth century are such examples in the biological sciences.

Areas that experience progress and increasing activity tend over time to collect a core of models that have survived and have been verified by continued investigation and scrutiny. These core areas are the well worked out, confidently inked-in portions of the crossword puzzle that Haack mentioned in her analogy. These productive areas of
research often develop well established methodologies and specialized tools. Successful areas of research can take their successful refined models and methodologies and perpetuate these to newer generations of researchers and learners in textbooks. The scientific work that forms the curriculum of the first year of medical school\textsuperscript{39} forms the core of empirical knowledge about the normal biological form and function of the human body. These fields share models, work across disciplines on larger sets of integrated models, and tend to produce mutual support and increase the likelihood of fit for the models involved. In these areas the claims about normal biological function do not raise suspicion or seem controversial. They do not appear ripe for being exposed as social constructions. It is the diseases of these systems that are detailed in the second year curriculum of medical school that are the uncontroversial cases of biological abnormality.

The evolutionary metaphor is an insightful one concerning the branching off into specialized disciplines and the differential rates of change, growth, and progress that disciplines achieve. While some areas thrive, some areas of inquiry proceed slowly or advance only in fits and starts. Some are popular or successful only for a period. Some types of research seem to be investigating phenomena that, so far, defy satisfying scientific explanations. Without a period of sustained success areas of inquiry cannot build up a core of well-established results and models. Without this core there is not a nucleus to build around. Models from such fields are often presumed to be less likely to have achieved as much fit as the models at the core of those fields having periods of sustained success. This problem usually accompanies the lack of a well-established methodologies and or techniques. Without shared methodology models may not get retested and refined as they would otherwise.\textsuperscript{40} In the worst case, each new generation of researchers may decide on its own preferred models and methods, only to have these

\textsuperscript{39} These include physiology, molecular biology, behavioral sciences, neuroscience, anatomy, embryology, histology, immunology, genetics, etc. Microbiology explains the normal form and function of microbes, whether these be pathogens or so-called normal flora of the human body.

\textsuperscript{40} Models developed with novel methods are often harder to initially evaluate. Models developed in disciplines that do not have a well established methodology are also harder to evaluate initially. Perhaps this is because the researchers in the discipline are not as closely tied together as those where a common, entrenched methodology exists and makes for more constructive criticism.
pushed aside by the next generation of researchers. Textbooks in such disciplines can not
draw on a well established core of results nor a common methodology and set of
techniques. Such fields instead have to organize textbooks on the theme of historical
development or on the central challenges or problems addressed by the field. Disciplines
with such problems might attempt borrowing methodologies from others disciplines
perceived to have established cores of successful models. Progress in these disciplines
might even come to be thought of as the establishment of newer methodologies. This also
creates opportunity for development of opposing camps along methodological lines, and
in the worst cases opposing camps each work with non-overlapping models and methods.
Groups of researchers or entire disciplines can be more isolated. The work in these
disciplines does not relate well nor seem relevant to other disciplines. There is less
exchange of ideas. In such disciplines there is often little sharing of models with
neighboring fields. The isolation tends to breed a sense that the discipline is special and
has very narrowly defined goals and objectives.

The social and psychological sciences appear to have suffered from various of
these problems more than the so-called natural or ‘hard’ sciences. These fields have not
tended to develop and retain a large core set of uncontroversial models or methodologies,
and tend to undergo something like Kuhn’s ‘paradigm shifts’ more often. Psychiatry has,
more or less, undergone shifts from physics inspired mechanistic models, through a
period influenced heavily by psycho-analysis, and now is influenced by models from
cell-biology driven mostly by the relative success of newer pharmaceutical interventions.
Part of the difficulties these fields face is the complexity of the subjects being studied and
the difficulty with isolating easily manipulated aspects of the phenomena being modeled.
The basic point for disease claims is that the models involved in specifying normal
human biology and behavior in these fields tend to be less well worked out, less well
tested over time, and less well integrated with models from other disciplines.

Some of the clues used by evaluators to attempt to assess whether a model is a
good description involve the reputation of the field in question. Some clues that a model
is likely to be retained and verified in the long run are that it comes from a field with an
established methodology and established, well-entrenched core body of results instead of
from a less successful, less well-established line of inquiry. A model from an isolated,
specialized field of inquiry is a more risky bet than one from a integrated discipline with models that have grown larger by having new aspects added through continued use and development. However, these are just clues and each model or hypothesis deserves careful assessment for its own virtues and merits as a representational model.

2.6 CONCLUSIONS

I have offered a broad brief sketch of science, what it produces, and how it selects better products from worse. I have very briefly alluded to the sorts of qualities I believe are involved in this selection process and also the sorts of qualities people use in making judgments about the virtues of particular scientific models. The reason for attempting such an ambitious task is because these qualities and these sorts of judgment are at root what determine better from worse disease claims and the amount of legitimacy that people judge particular disease claims to have. In the next chapter I shall turn towards discussing the notion of normal and abnormal generated by virtuous biological models and how this theoretical notion of normal differs from other possible notions of normal. In the final chapter of this project I shall turn directly to discussing what being a disease means and reiterate what makes for a legitimate versus an illegitimate disease claim. I shall also discuss controversial and interesting disease claims in light of my account.
CHAPTER 3
NORMALITY

3.1 NORMALITY

My main thesis is that, for certain signs or symptoms—certain human problems—to be the observable consequences\(^{41}\) of a legitimate disease, there must be a scientific model that explains how these observable problems are caused by abnormalities of a biological (or psychological) system. The model must also provide details of the normal function of that system. The legitimacy of the disease claim depends on the legitimacy and virtues of the scientific model. Ideally this model is an accurate unbiased representation of the biological system. In this chapter, I shall discuss possible equivocations across three different senses of normality. Biological models often specify a theoretical sense of normal and abnormal function or structure. However, conventional wisdom and mere measurement might also generate senses of normality. Equivocation about abnormality is especially likely when the signs and symptoms of a putative disease are open to non-scientific senses. Failure to recognize these different senses, and failure to recognize the different ways a judgment of normality might be generated, can lead to significant controversy surrounding certain putative disease claims. In this chapter I shall distinguish three importantly different senses of abnormality. I shall explain how the charge of ‘medicalizing’ certain problems can best be understood in terms of these differing senses of abnormality. I will also discuss an argument by Ron Amundson that the concept of abnormality ought to be avoided in biology. Finally, I shall consider the charge of ‘social construction’ of disease claims, and argue that it can be best understood in terms of differing senses of abnormality.

---

\(^{41}\) There are actually cases, on my account, where a disease process is occurring (an abnormally functioning biological system) but no observable signs or symptoms are present. It is possible to have a disease and not be outwardly ill or suffering. It is even possible that this disease persists chronically without any consequences perceived as adverse to the patient or members of her community. There is no necessary connection between being a disease and being a disadvantage or source of suffering to the person affected by the disease. I shall return to this topic and discuss examples in chapter 4.
3.2 HACKING ON NORMALITY

How any concept emerges and gets shaped over time is fascinating. Georges Canguilheim (1978) and Ian Hacking (1990) both discuss the emergence of the concept of ‘normal.’ The word ‘normal’ originates in European languages as a synonym for perpendicular or orthogonal. The Latin root refers to a builder’s T-square—a device for measuring right angles—and the Greek root refers to a surveyor’s rod. But even at that stage, notes Hacking, there was an ambiguity in the term.

On the one hand the words are descriptive. A line may be orthogonal or normal (at right angles to the tangent of a circle, say) or not. This is a description of a line. But the evaluative ‘right’ lurks in the background of right angles. It is just a fact that an angle is a right angle, but it is also a ‘right’ angle, a good one. Orthodontists straighten the teeth of children; they make the crooked straight. But they also put the teeth right, make them better. Orthopedic surgeons straighten bones. Orthopsychiatry is the study of mental disorders chiefly of children. It aims at making the child—normal. The orthodox conform to certain standards, which used to be a good thing (Hacking, 1990, p. 163).

Hacking traces the first usages of ‘normal’ in English back to the 1820s, when it was borrowed to describe French advances in physiology and medicine.42

The notion of the ‘normal state’ grew out of attempts by French physiologists to understand pathology and disease. Towards the end of the eighteenth century, newer concepts of health were being superimposed on older ones. Older notions utilized the metaphor of health as a mean, or balance of elements within the organism, while pathology was an imbalance (see Garrison, 1929). Disease was increasingly being understood as a process occurring not at the level of the whole body, but at the level of body systems and organs. Physiologists were engaging in the decomposition of the organism into subsystems. Pathologists and physiologists were explaining disease as malfunction in specific subsystems. They were working on explaining the mechanisms by which the body controlled and regulated its internal systems in the face of external

---

42 Hacking says: “The first meaning of ‘normal’ given in any current English dictionary is something like ‘usual, regular, common, typical.’ The OED dictionary says that this usage became current after 1840, and gives 1828 for its first citation of ‘normal or typical’. That was in a work of natural history alluding to the French writers (Hacking, 1990, p. 162).”
perturbations. The terminology of normality and abnormality (in English) had these specialized medical usages first, and only then expanded into more general usages. But even from the beginning this specialized terminology contained the possibility for equivocation and misunderstandings. Hacking, rather poetically, calls attention to this problem:

As a word, ‘normal’ …acquired its present most common meaning only in the 1820s… The normal was one of a pair. Its opposite was the pathological and for a short time its domain was chiefly medical. Then it moved into the sphere of—almost everything. People, behavior, states of affairs, diplomatic relations, molecules: all these may be normal or abnormal. The word became indispensable because it created a way to be ‘objective’ about human beings. The word is also like a faithful retainer, a voice from the past. It uses a power as old as Aristotle to bridge the fact/value distinction, whispering in your ear that what is normal is also all right. But also… it is a soothsayer, teller of the future, of progress and ends (Hacking, 1990, p. 160).

It was during the birth of what we recognize now as modern physiology that the term ‘normal’ was developed. The notion that a disease was a malfunction of a biological abnormality, however, was not new. Rather, what was new during this period were advances in our understanding of both organic and inorganic chemistry—and the dissolving of the organic-inorganic dualism. It was discovered that the chemical processes that allowed living beings to function were the same as those that could be carried out in a flask. This allowed different kinds of models to be built—different kinds of explanations concerning how systems functioned. It also made decomposition of larger complex systems into smaller simpler systems a fruitful strategy.

During the nineteenth century formal statistics was also being developed. The concepts of determinism and of mechanistic laws of nature were increasingly displaced by ideas of variation about a mean, and the mathematics of chance. Things once seen as unmeasurable and outside the scope of science were now fair game for the new techniques of measurement and analysis. There was an explosion of measurement and quantification in nineteenth century biology. The concept of ‘normality’ that we inherit from the nineteenth century involves elements of both its statistical and physiological heritage.

These themes of variation about a mean, along with increasing efforts to functionally decompose and measure complex processes, were picked-up and exploited
creatively by intellectuals outside physiology. Thinkers like Comte and Durkheim sought to expand the notion of normality into the analysis of the complex ‘organism’ called society. Such efforts re-ordered the priority of investigation. Whereas in physiology, malfunction had taken center stage and normal function was often defined as the absence of such perturbations, social thinkers increasingly focused on fleshing out the normal. Hacking says:

Something was normal when it was not associated with a pathological organ. Thus far the normal would be secondary, defined as the opposite of the primary notion, the pathological. But then what Comte called the great ‘principle’ of Broussais turned this around. The pathological was defined as deviation from the normal. All variation was characterized in terms of variation from the normal state. …Note the two parts of this ‘principle’: (a) pathology is not different in kind than the normal; ‘nature makes no jumps’ but passes from the normal to the pathological continuously. (b) The normal is the centre from which deviation departs. …The idea of continuous deviation from the normal came from pathology, as interpreted by August Comte. …He made it a basis for his social science and it became part of his political agenda. …Broussais’s ‘normal state’ might have made its way into language unattached, but it was the enthusiasm of Comte that gave it elevation and status. The idea that the pathological is not radically different from the normal, but only an extension of the variation proper to a ‘normal organism,’ was, he wrote, an ‘eminently philosophical principle whose definitive establishment we owe to the bold and persevering genius of our illustrious fellow citizen, Broussais.’ The important point was that all the characteristics of a thing were defined relative to the normal state. Explicitly: ‘The law of Broussais subordinates all modifications to the normal state.’ Broussais wrote of physiology, but his principles must be extended to ‘intellectual and moral functions’—and then, as my epigraph continues, to the whole study of society (Hacking, 1990, p. 166).

The concept of normality, expanded to do work in areas such as psychology, sociology, statistics and politics, was something to be discovered independently of any particular pathology. As its usage expanded a tension between two conceptions of ‘normal’ was growing—a tension that continues to cause problems, inspire rhetoric, and cause misunderstandings. Pulling in one direction were conceptions of ‘normal’ as the typical, average, even mediocre—something to improve upon if possible. Pulling in another direction were conceptions of ‘normal’ that stress it as a balance, equilibrium, or
a preferred state of affairs—the ‘natural state of affairs’ lacking perturbation or disruption. Hacking discusses how such tension became solidified:

Comte thus expresses and to some extent invented a fundamental tension in the idea of the normal—the normal as existing average, and the normal as figure of perfection to which we may progress. This is an even richer source of hidden power than the fact/value ambiguity that had always been present in the idea of the normal. The tension makes itself felt in different ways. If we think ahead to sociology and to statistics, in the modern comprehension of those terms—that is, if we think ahead to the work encrusted around names such as Durkheim and Galton—we feel the tension acutely. On the one hand there is the thought that the normal is what is right, so that talk of the normal is a splendid way of preserving or returning to the status quo. That’s ‘Durkheim’. On the other hand is the idea that the normal is only average, and so is something to be improved upon. That’s ‘Galton.’…The tension in these aspects of the normal will not dissolve just by noting that there are two ideas, one of preservation, one of amelioration. The former carries within it fondness for origins, youthful good health, an ideal condition to which we should be restored. The latter lusts after teleology, of ends that we may choose for the perfection of ourselves or of the race. Two kinds of progress (Hacking, 1990, p. 169).

This unresolved tension between amelioration and perfection remains under the surface in controversies about using biotechnology for altering or effecting human appearance and performance. This same tension continues to be felt today in debates about the concept of health.

The concept of ‘normality’ is currently ubiquitous, and has continued to cause confusion and controversy as science attempts to describe aspects of human life and behavior that were previously the domain of politics, religion or morality. Each of these perspectives on human events and behavior (science, politics, religion, etc.) are capable of describing what is normal and what is abnormal. It is possible to use the concepts of normal and abnormal with primarily either physiological, sociological or statistical senses in mind. However, the meaning behind such uses, as well as the intention and authority to credibly make such claims, are importantly different across different notions.

3.3 VACHA ON NORMALITY

Jiri Vacha (1978, 1985) has written on the ambiguity in the term ‘normal’ as it is used in biological science. He says:
At first sight, its meaning is clear to everyone, but a quite superficial attempt to define normality will suffice to reveal its deceptive polysemy. Normality is the elusive Proteus whose countenance changes according to biological branch and philosophical conviction of authors. The universality of this term suggests that it can hardly be dispensed with in biology, or at least that the majority of biologists are convinced of this; yet one would in vain look for the elucidation of what is meant by the word ‘normal’ in standard textbooks of ‘normal’ anatomy and physiology (Vacha, 1978, p. 823).

Vacha provides seven slightly different conceptions of the term ‘normal’ used in biological science. He offers these on a spectrum which moves from the less intricate and less value laden, to the most intricate and value laden. This spectrum also moves roughly from statistical conceptions through functional and into ideal conceptions.

The least complex notion on the spectrum is normality as commonness or the run-of-the-mill. Next is normality as maximum frequency or as the mean. Then comes normality understood as typicality—as the defining features of some type. The catalogue continues through normality as adequacy (being within some defined interval or range), normality as optimality, and normality as ideal performance. Finally, Vacha considers the possibility that one concept might capture all these aspects of normality. He says of this super-conception that it requires a presupposition “that the various meanings of the word ‘normal’ have a common basis, that some sort of normality exists in a general sense, combining the aspects of frequency, functionality and normativity (1978, p. 843).” Vacha is skeptical that the various meanings can be reconciled, and ultimately advises that scientists drop its usage for more precise terms.

I agree that there are importantly different notions of normality. Careful examination of these different senses of normality allows them to be distinguished and untangled in particular cases where there is ambiguity or overlap. Furthermore, science usually utilizes a sense of normal that is importantly different both from the predominately statistical notions describing the average or typical, and from an overtly

---

43 Vacha (1985) has also discussed the changing ideas about disease in the eighteenth and nineteenth centuries. The integration of mathematical conceptions for measuring variation caused various problems for scientific categorization. As an example he discusses the development in the 1920’s and 1930’s in Germany of constitutional theory—or how to define gross anatomical normality for human beings and how to catalogue abnormalities.
normative sense that equates normality with what is preferred or correct. Science creates
models that attempt to describe how things actually operate and function, not describe
what we value or how we wish the system would operate. Nor are these models merely
reports of the average performance on some measure. They are more complex and often
present the system as goal-directed and as-if-designed, even while ignoring or being
agnostic about the origin of this appearance of design or purpose.

The statistical notion of normality was mixed with the physiological notions of
normal function in the nineteenth century. The expansion of the concept into explaining
social arrangements and cultural phenomena added further complexity. Currently there
are many contributing currents running in the concept of normality. I will argue that out
of these influences it is possible to separate three importantly different but easily
confused conceptions. Keeping track of these separable conceptions of normality,
especially when they are mixed together in putative disease claims, can clarify some
points of controversy. Furthermore, being clear about what is meant by normality in
certain contexts allows more fruitful debate about utilizing biotechnology for non-
therapeutic changes to human biological systems (i.e. medical interventions not meant
primarily to treat disease).

3.4 THREE NOTIONS OF NORMALITY

I agree with Hacking and Vacha that the concept of normality is filled with
tensions and ambiguity. I believe this ambiguity perpetuates the hope that descriptions of
the physical world can inform our judgments about what is and is not of value. Some are
hopeful about finding an objective guide to human endeavor, and find the richness of the
concept of normality encouraging. Such hopes also drive philosophical exploration of
the notion of function and whether this notion creates a natural teleology (see Wright,
1973, 1976). Others are suspicious of claims of normality (see Dreger, 2004 and

44 I am not a proponent of a fact/value dichotomy, although I do believe talking of facts
and values, or descriptions versus value judgments is sometimes appropriate or useful.
Even our best simple observations, not to mention our best theoretical descriptions of the
world, are infected with being made from a perspective, and these perspectives involve
background beliefs and judgments involving epistemic and other values. Likewise value
judgments cannot be made in a vacuum completely uninfected by beliefs about how the
world or people in it are constituted.
Amundson, 2000). If we focus less on the historical roots of this tension and more on the current configuration, especially in regard to the issue of disease and health, I think we find three largely separable notions of normality. These would be (a) conventional normality, (b) statistical normality and (c) theoretical normality. Distinguishing the three and looking for entanglements of these notions in particular cases can be helpful in understanding why many putative disease claims are controversial. Such a distinction will also offer insight into what is really being claimed in charges of ‘medicalizing’ people’s problems or when a disease is described as merely a ‘social construction.’

One notion of normality is the conventional. This is the main sense of normal appealed to in the claim that the marriage of two adults of opposite sex is normal. It is what is meant when someone says that it’s abnormal for women to wear men’s clothes, or cut their hair short. It relies on reference to a social norm or convention within a particular community or group. Claims about something being normal in this sense are claims about human preferences or practices. To observe that something is normal in this sense is to observe that it is in accord with the preferences or practices of some specific group or culture. The phrases normal and abnormal in these uses can often be replaced by the phrases ‘proper’ or ‘expected.’ This sense captures the side of Hacking’s tension that has to do with expectations of the way things should be—to correct the abnormality is to go back to how things ought to be, were expected to be, or were legislated to be. Different actions can be achieved by claiming something is normal in this sense. The statement can be purely a description of the preferences, norms, or standards of a group. It can be intended as merely an observation without any implied endorsement or criticism. It could even be part of a scientific explanation of certain social practices, customs, and preferences at some place and time. However, the statement that something is normal in this conventional sense is more often intended to have evaluative force as

---

45 After writing the first draft of this chapter I became aware of the article by Wachbroit (1994) which also makes a three-part distinction somewhat similar to the one I am making. However, Wachbroit’s article focuses on arguing that one notion is special to the biological sciences (as opposed to the physical) and also that it is conceptually prior to the notion of function (in that notions of function, to be understood, must already assume the sense of normality in question). Wachbroit doesn’t give a very detailed idea of what this third kind of ‘biological normality’ amounts to except to say that it is relied on in allowing the distinction between function and malfunction.
People often cite something as normal, in this sense, in order to express their preference for the standard or norm, or to endorse it, and to criticize those falling outside what is normal. Putative disease claims that depend on or involve this sense of normal and abnormal often arouse suspicion that those making the diagnosis are also expressing approval or disapproval as well.46

The second sense of normal is the statistical. It is the sense of normal captured in the claim that it is normal for humans to have 10 fingers or one heart, or that the normal height for an adult male in the U.S. is between 4’10” and 5’10”. This is the sense in which it is abnormal to weigh 480lbs or to have 36 teeth. But it is also the sense in which a normal RBC is between 6-8 microns in diameter or a normal water molecule is 25 Angstroms. The phrase normal in this usage can almost always be replaced by the phrase ‘average.’ This notion of normal does not carry with it an underlying notion of things being the way that they are for a reason or purpose. States of affair are merely found to be this way. This sense of normal rarely carries with it a strong sense of teleology. However it often does carry with it a sense of being natural—of being the way that nature is, regardless of how any community might like it or want it. Normal in such contexts can acquire a sense of being a balance or mean along a ‘naturally’ ordered (i.e. merely found in the world) spectrum of variation. This sense of normal is closely tied to techniques in the field of statistics. This sense can come to be applied to anything that can be quantified and measured. This includes not only macroscopically observable things but also things that can only be characterized by using data—both things so large scale and complex that they cannot be directly observed or things too small to directly observe. Statistics and other formalized techniques of analyzing variation do ultimately apply human judgments. For example, humans decide how to measure where to set the cutoff for the ‘normal’ range.

The third sense of normal is particularly important to debates about disease. It is the sense captured in the claim that it is normal for the heart to pump blood and produce a

46 An example might be those claiming the behavior diagnosable as ADHD is a biological abnormality. Those critical of these sorts of disease claims might suspect that what is really being claimed is that such behavior is not valued or preferred by the community in question at the time in question. Perhaps in a largely agrarian economy in the past such behavior would not have been conventionally abnormal.
particular wave pattern on a standard 12-lead EKG machine. It is the sense in which having type I diabetes (i.e. autoimmune destruction of beta cells of the pancreas) is abnormal, while having a pancreas that secretes insulin in response to elevated serum glucose and/or elevated levels of certain amino acids is normal. The phrase abnormal (in biology) can almost always be replaced by ‘diseased,’ or ‘pathological.’ This sense of normality is an inherently theoretical notion. It is an integral part of the scientific model that generates the notion. The postulation of normal and pathological states is built into the scientific model of the system in question. The designation of, and details of, the system’s function and the various possibilities and mechanisms of dysfunction are a crucial and central part of the explanation of many models in biology and psychology. Claims of theoretical normal function are made in reference to a scientific model. This sense of normal captures something different than merely a measured mean or a conventional norm. It is not something that just gets ‘read’ into or onto the model by interpreters or by ‘loose’ and imprecise discussion. It is not merely a scaffold or aid to the creation of the model. The specification of the normal function of the pancreas is central to what the model is describing.

Some might argue that this theoretical sense of normality (or something like it) is special to the biological sciences. I doubt that such a sense of normality or the type of models that generate it are particular only to biology, although I do admit biology has made conspicuous usage of these kinds of models and that this probably has to do with the fact that the phenomena in question seem well suited for this type of explanation and that ultimately this type of explanation has delivered satisfying results. Part of what

47 Not the only, but the most important result, would be that these types of models have good fit with the phenomena. Do the phenomena being modeled actually and ‘really’ have normal and pathological states (as opposed to being merely our interpretation)? In considering how one might determine the ‘real’ structure, character, or function of certain biological systems, the only means to do so is our best available scientific investigations of these systems. This makes me a proponent of some variety of naturalized epistemology. These are complex philosophical issues, but what I can say here is that to the extent that our best and most virtuous, well fitting, models explain that these phenomena have built-in goals or states that are arranged or designed, to maintain or achieve, then, to that extent, such claims to normality are natural and objective. The issue about bias and social or political preferences or opinions influencing the determination of the normal or proper state of affairs is an important issue. But the model can be assessed
makes these types of explanations seem special or different is that they generally take a broad metaphorical stance towards the phenomena being modeled, namely the ‘as-if-designed’ stance. Phenomena are approached as if they were machine-like in their operations, and models are created that incorporate this understanding. This approach often generates, as part of the explanation, the specification of states of normal function (the accomplishment of some goal or task) and the specification of pathological states (where the goal in mind is not accomplished as well). Furthermore such models are often part of even larger models or groups of models, with a hierarchy, explicit or assumed, of lower-level normal functions or goals contributing to the attainment of the functions of higher-level systems. Think of the example of the human thyroid from the last chapter. It is difficult in many cases to specify the ultimate goals or functions of the system as a whole, or even sometimes to determine just what constitutes the final or ultimate system and investigated for evidence of its fit and virtue as an objective and accurate representation of the phenomena and also as a model whose design, features or claims may be biased or influenced by conventional norms of some kind. It is possible for a model to be both accurate as a representation and biased. This fact may often be overlooked due to the relatively greater attention given historical examples of model that were very poor regarding fit or accuracy and clearly biased or based on conventional norms. Less attention is given models that are infected with bias and conventional thinking, but also have some degree or accuracy and good fit. Such biased but relatively accurate models usually get refined and improved without much notice to the original bias or conventionality with which they were at first infected. The cases in which such things are overlooked, but then revealed by careful historical scholarship are very interesting.

This is just one of many broad metaphorical stances that might be adopted—to approach the system as if it were a designed artifact. Another one would be to approach complex dynamic phenomena as being like complex social or economic phenomena. This metaphorical approach to modeling has been described sometimes as ‘reverse engineering’ or ‘functional decomposition.’ It is a common and widely used technique employed in modeling complex phenomena. Depending on the context, however, there are different understandings of why and how something has come to be such that it behaves like a designed artifact. Sometimes goals such as survival or homeostasis are appealed to. But the ultimate goals of such hierarchies of functional systems are often vague or poorly defined. One can appeal to some other line of inquiry, such as evolutionary theory or theology to provide the content and the explanation of how and why the system is machine-like or was in some sense designed. The reason for this vagueness has to do with the fact that when deploying the decomposition-into-integrated-systems approach it is not crucial to know or define exactly what the highest order goals of the system in question are, or

48 This is just one of many broad metaphorical stances that might be adopted—to approach the system as if it were a designed artifact. Another one would be to approach complex dynamic phenomena as being like complex social or economic phenomena. 49 This metaphorical approach to modeling has been described sometimes as ‘reverse engineering’ or ‘functional decomposition.’ It is a common and widely used technique employed in modeling complex phenomena. Depending on the context, however, there are different understandings of why and how something has come to be such that it behaves like a designed artifact. 50 Sometimes goals such as survival or homeostasis are appealed to. But the ultimate goals of such hierarchies of functional systems are often vague or poorly defined. One can appeal to some other line of inquiry, such as evolutionary theory or theology to provide the content and the explanation of how and why the system is machine-like or was in some sense designed. The reason for this vagueness has to do with the fact that when deploying the decomposition-into-integrated-systems approach it is not crucial to know or define exactly what the highest order goals of the system in question are, or
at the top of the hierarchy. Furthermore, it is a controversial whether each system has one specific and dominant function, or can have many functions. A closely related controversy is the creation of a sense of teleology in these hierarchies of specified functions.\textsuperscript{51}

Take the example of schizophrenia to highlight the three concepts of normality. Certain patterns of behavior are characterized as abnormal and become the criteria for making the diagnosis.\textsuperscript{52} Such behavior can be seen as a statistical abnormality in that approximately one percent of humans (across nationality and cultures) behave and have experiences in ways consistent with the DSM criteria (APA, 2000). Because the DSM criteria are in terms of necessary and sufficient conditions, the numbers meeting the criteria (and those not meeting the criteria) can be measured. Being normal in the statistical sense means only that one does not meet the criteria. The behavior of those diagnosable as schizophrenics is also a conventional abnormality in that most people don’t expect or prefer humans to act that way. Relative to my culture (and many others), with its standards and norms for behavior, schizophrenics are abnormal. However, even if

\textsuperscript{51} Again, to the extent that our best scientific descriptions of the world create a sense of teleology or purpose for some systems, this is acceptable. The crux of the issue here is whether a specified system has one dominant function or main purpose that explains why the system exists, or whether the function has any relationship to survival. What is clear is that science describes what already exists, and in many cases it assigns function without much information on ultimate ends or the origins of the systems it models. Science builds models of locally specified functions with the goals being determined by the investigator creating the model. There is a branch of biology that seeks to understand how systems that look as-if-designed came to exist, but many models in physiology and medicine are constructed without any input regarding evolution.

\textsuperscript{52} This is actually by design in psychiatry. The DSM is a scheme for diagnosing mental disorders based on signs and symptoms alone without any necessary reference to the underlying scientific (or non-scientific) explanations of the cause of the behavior. The advantage is a relatively theory-neutral means of diagnosis that members of different theoretical camps can agree upon.
diverse cultures find schizophrenic type behavior abnormal, the content of the contrasting notion of conventionally normal behavior is most likely different across different communities.\textsuperscript{53}

The behavior of schizophrenics, however, can also be seen as a consequence of underlying malfunction. This is what it means for schizophrenia to be a putative disease, namely for the behavior to be conceived of as caused by the malfunction of a biological system. It can be understood as a malfunction of neural circuitry, or dysregulation of certain neurotransmitters or of populations of specific receptors. These circuits (and the cells, neurotransmitters, and receptors instantiating them) all have normal functions specified (or potentially specified) in theoretical scientific models. It is these models that characterize what is normal, and what constitutes malfunction. Normal and abnormal, here, only make sense in reference to some particular model of some biological system. It is the model—admittedly a model constructed for human purposes and with an eye on explaining what may also be a conventional or statistical abnormality—that creates this additional sense of normal and abnormal function.\textsuperscript{54} Whether the models are speculative and not yet well worked out, or are well tested, reliable, and uncontroversial, will determine whether the community in question accepts the disease claim as a putative or speculative one, or a legitimate and unproblematic one.

Interestingly, those models implicated in schizophrenia, and that explain the functions of various dopaminergic neural pathways in the brain, are still being developed. An accurate model of their normal functions is being sought, even though most experts and psychiatrists are confident these pathways are the ones malfunctioning in the disease, and are the ones most affected by partially successful chemical intervention (see Eisenberg, 2010). On my explanation, schizophrenia is still a putative disease. The models detailing the biological systems being disrupted are still being worked out. However, the observable consequences are so strongly perceived to be conventionally

\textsuperscript{53} Some cultures may find the behavior of schizophrenia not pathological but exceptional, perhaps elevating those exhibiting the typical behavior to the position of high priest or spiritual advisor.

\textsuperscript{54} The theoretical abnormality explains the observed conventional abnormality in behavior. The disease explains why someone suffers the signs of symptoms that can be grossly observed.
abnormal, and there has been so much progress in the last 20 years understanding the systems believed to be malfunctioning that it is a rather uncontroversial putative disease claim. It is interesting to compare the situation with the putative disease claims of ADHD or fibromyalgia, whose observable signs and symptoms are not so clearly perceived as conventional abnormalities. Even as some researchers attempt to build models of the underlying systems being disrupted the general perception is that having aches and pains all the time or being hyperactive at school are not all that abnormal. Public opinion is not unanimous on perceiving these as abnormalities or on treating such problems medically.

In summary, I do believe there are three senses of normal that can be separated from each other. One sense is relative to measurement, one is relative to conventions or preference, and one is relative to theoretical scientific models. These senses of abnormality can in various ways overlap, support, or interact with each other. In actual scientific theorizing, investigators often start out by taking a conventional or statistical abnormality and building a theoretical model that explains (in mechanistic details) why we observe the conventional aberrations or statistical distributions. The received wisdom a community perpetuates about some conventional abnormality can have important consequences for how the attempt to offer a theoretical explanation is viewed. Conservatives might view attempts to offer biological models of malfunction as the ‘medicalization’ of a non-medical, even a moral, problem. Progressives might push for a medical perspective to be adopted on what were previously thought of as character flaws, moral weakness, or social ills. Progressives might push for a medical solution to problems previously considered merely the natural or normal course of life—such as the slow deterioration of function with advancing age.

3.5 OVERLAPPING SENSES OF NORMAL AND MEDICALIZATION

The intermingling of the three senses of abnormality is often involved in controversies concerning putative disease claims. Figure 3.1 is a simple Venn diagram of the overlap of the three sense of abnormality. I have placed certain putative disease claims on the diagram to demonstrate both the difficulties and benefits of trying to untangle the three senses. I do not intend the assignment of cases to regions of the diagram to be taken too seriously. The point is to see how certain human conditions or
behaviors can draw on different senses of being abnormal, and to see how intuitions about being a disease versus another sort of abnormality might fluctuate as these senses become tangled. Failure to recognize the different senses of abnormality simultaneously at work creates many of the puzzles and controversies concerning whether or not a condition should be considered a ‘disease’ versus some other sort of problem. Note also that this diagram does not offer any assessment of the virtue and accuracy of the scientific models generating the purported theoretical sense of abnormality.

FIGURE 3.1: Overlapping sense of abnormal

Focusing on the overlap between statistical abnormality and theoretical abnormality one might consider type I diabetes, AIDS, as well as other infectious and autoimmune diseases. Both have been statistically rare, but AIDS is growing in both prevalence, and could move out of the statistical circle as it becomes more prevalent.
Currently both enjoy a synergy between being abnormal in the sense of being a well-worked out and accurately modeled pathology and being uncommon. This area will contain many uncontroversial classic diseases—theoretical abnormalities that are uncommon and, importantly, don’t have to compete with conventional wisdom in the same way that many behavioral or anatomical abnormalities do.\textsuperscript{55}

Coronary artery disease (CAD) and atherosclerosis have been and continue to be epidemic in the wealthy developed world. These conditions affect many people and produce much morbidity and mortality. There is pressure to find ways to intervene and influence the outcome for as many people as possible. Accurate scientific models are being developed to explain these as theoretical abnormalities of human physiology and metabolism. They can result in abnormal, even catastrophic disruptions of function (i.e. MI or heart attack). These scientific models also explain much about the normal function of many metabolic and physiological processes involved. Phenomena in this area of Figure 3.1 (i.e. in the theoretical abnormality circle alone) are not rare and often develop slowly over time. These phenomena often require that the scientific models of abnormality, at least originally, rely on multifactorial explanations. Epidemiological data on genetic, environmental and behavioral risk factors are gathered and used to guide and stimulate model building.\textsuperscript{56} Often these models involve the additive and interacting

\textsuperscript{55} There are a small and dwindling group of people who believe AIDS is not caused by HIV and thus reject the idea that this is an accurate theoretical model. Some people believe AIDS is just a ‘social construction’ and as such is merely a conventional sense of abnormal being used to segregate people for political or economic reasons. I do not know of anyone who claims that the model for type 1 diabetes is inaccurate or that the science offered is just a way to legitimize social and political power relations, however it would not be surprising to find these sorts of claims becoming a ‘hot’ topic as concern and publicity increase about the epidemic of type 2 diabetes in racial minority populations (African Americans and Hispanics) in the U.S.

\textsuperscript{56} This is not the whole story—in many cases there are also isolated genetic metabolic defects that produce early onset and aggressive versions of the disorder in question. These isolated varieties draw attention and often allow accurate models of the systems in question to be produced. These serve as important parts of explaining the normal functions and possible routes of abnormality in those physiological and metabolic systems that govern the more general, more multifactorial versions of the disorder. For atherosclerosis and heart disease the discovery of patients with a mutation in their LDL receptors causing these to malfunction, and causing these patients to die of heart disease in their teens, was a crucial part of the ongoing processes of developing the broader
contributions of many components that are hard to isolate and control completely, even in experimental animal models. Essential hypertension is another example. Acne and tooth decay fit in this area as well, although one might argue these conditions are considered conventional abnormalities for which theoretical explanations are being sought.

The fact that a condition is not rare may lead some to believe that it is not a disease. If most people have these conditions, are they not part of the normal behavior of the biological systems in question? This problem arises from conflating statistical and theoretical abnormality. Many diseases that effect lots of people involve issues surrounding what constitutes normal vs. abnormal aging and the decline in function in most biological systems with age. Some rate of deterioration, and some rate of atherosclerotic plaque formation, as well as some amount of acne and tooth decay, is probably part of normal development and aging. However, certain cases are not normal. The issue is just how much deterioration in function at any given age is in accord with normal aging. Finding this balance point produces controversy and unique issues generally for many abnormalities in these areas of Figure 3.1.

In situations where the normal state or function is difficult to distinguish from normal aging and deterioration, there is a propensity to confuse the ‘ideal’ with the ‘normal.’ Recall the example from chapter 1 concerning testosterone levels. In the case of atherosclerosis and heart disease this confusion can be seen in issues about what level of serum cholesterol is normal. There surely is a ‘normal’ value that science could determine if it had a very well worked out model of human aging and deterioration in function with age. However, medical advice and intervention seems aimed at an ideal level of serum cholesterol, especially now that means to technologically alter this value are available. There is controversy about defining the ideal, although scientific investigation of the systems and phenomena in question could yield an answer on this as well. Ultimately, the ideal might be the lowest amount of serum cholesterol (and cellular stores) that an otherwise normal adult could tolerate. But there is a difference between the ideal level and the normal level of serum cholesterol for a male or female of a particular

---

multifactorial explanations of what goes wrong for the more run-of-the mill person who develops heart disease.

57 Some cases are clear cut diseases. They are extremes. They are malfunctions of the system that otherwise functions normally.
age, just as there is a difference between the normal and ideal amount of atherosclerosis (or acne) for an otherwise healthy human given gender and age. The theoretical normal amount has nothing to do with our preferences while the theoretical ideal amount necessarily requires human preference to determine.

Science can handle these challenges. An accurate and authoritative, although complex, model of human aging (with specification of normal functional and structural changes and abnormalities) is possible, and certainly some models and details are already available. Ultimately science and its models can become as complex as necessary to handle such issues. Currently, however, theoretical models of human aging are not well worked out or extensive. This void where a satisfying theoretical explanation of normal aging could function is filled by explanation or speculation from other sources, like religion, politics, culture, etc—i.e. conventional senses of normal aging. Tooth decay and acne also run into problems due to the fact that scientific models of normal aging are not well worked out. It is all too easy for notions of conventional normality or even ideality to slip into the void. Some amount of acne as an adolescent is normal, but how much is not well worked out yet.

Currently masturbation is a pure conventional abnormality—there is no current scientific theory about it being a behavioral malfunction, nor do we currently believe it is statistically rare. It is only abnormal relative to norms of behavior for particular communities at particular times.\footnote{There is a rather embarrassing history of medical treatment of the disease of masturbation, especially when diagnosed in adolescence. See Engelhardt (1986).} There are other examples in the literature such as drapetomania—the disease ascribed to antebellum southern slaves who repeatedly ran away from their masters (see Cartwright, 1851). Even at the time of its introduction as a possible diagnosis, the abnormality should have been recognized as merely conventional.\footnote{There are some historical examples that were conventional abnormalities, and covered a broad group of individuals with similar appearance, behavior, etc. but among this overly broad group of ‘abnormals’ were subgroups of sufferers of what we currently recognize as legitimate diseases. This is probably particularly true of many historical cases of putative psychological diseases.} There are perhaps some abnormalities that tend towards purely statistical, like rare eye color.
On Figure 3.1 the abnormal behavior of a schizophrenic might be placed in the middle, as it can be interpreted as an abnormality in all three senses. Certain anatomical ‘defects’ that are held to be undesirable by the culture in question, are statistically rare, and also have a well established, respected, and accurate model explaining how such malformations occur, could also be placed in this area of triple overlap. Some of the various types of cleft palate serve as an example. Being born with either one less or one more finger than the average of five per hand, may be included in this group, although the embryological malfunctions that cause such cases are less well worked out than for many cases of cleft palate. There are potentially a large number of anatomical features of people that we can establish as statistical abnormalities and which our community considers abnormal. Whether there is a reasonable model explaining them as arising from the malfunction of a biological system determines if they make it into the area of triple overlap or stay in the area of double overlap to the right on Figure 3.1.

This area of double overlap containing statistical abnormalities that are also conventionally abnormal produces many problematic putative diseases claims. Homosexuality, shyness, chronic fatigue syndrome, abnormally large or small breasts, feeling certain that one is actually the opposite gender than what one’s external genitalia might testify to the world, etc. could all be examples. The phenomena being examined in this overlap are often prone to becoming medicalized. Some doctors and their abnormal patients begin to look for scientific theories that explain the problem in question. They seek explanations that will allow the misfortune to move into (or at least closer to) the area of triple overlap. Medicalization is the search for a theoretical, scientific explanation for an abnormality, and is particularly controversial when the abnormality is already clearly perceived to be a conventional and statistical abnormality. The most pejorative accusations of medicalization also contain the assumption that the problem being medicalized should not be understood as a disease, or handled by the intervention of medical practitioners.

It may be that part or all of the cases of conventional and rare abnormalities have, forthcoming, accurate explanation as malfunctions of otherwise normally operating biological systems. However, during the period in which scientific research is ongoing, it is still an open question whether the behavior or shapes of bodies in question can be
explained as a scientifically respectable biological malfunction or malformation. The behaviors that we now recognize as signs of the disease of schizophrenia had to make this move from conventional abnormality towards theoretical—and it is still making the move—and there are probably still some people who consider such behavior a sign of moral weakness or spiritual unrest instead of a legitimate, medical disease.

If one looks at the many putative disease cases that could be placed in this area of double overlap, another common feature emerges. Many of these abnormalities, or the symptoms of the abnormality, are themselves grossly observable. Because fields like psychiatry and psychology deal with problematic human behavior and are searching for theoretical models that explain the difference between the pathological and the normal, these fields are particularly prone to criticisms of medicalizing problems. ADHD has proven a controversial example regarding searching for a scientific model to explain conventionally problematic behavior as a result of a theoretical pathology. Such pathology should be contrasted with a theoretically normal state of cognition and brain function. The fact that certain medications seem able to ameliorate the problematic behavior helps direct the search for the desired scientific explanations, and ultimately the search for legitimacy as a disease claim. Culturally disreputable or odd behavior tempts psychiatrists (amateur or otherwise) to medicalize. Similarly, rare anatomical variations that society finds abnormal are also tempting targets for medicalization. When it comes to anatomical variation, the scientific fields of research that are called upon to explain normality versus abnormality are those of human development and performance, including embryology, some developmental pediatrics, endocrinology, kinesiology, etc. Science has models of normal human variation concerning gross anatomical structures and internal organs, etc. but these are incomplete and their detail only extends so far. The

---

60 Many of the problematic cases of purported diseases in the overlap between statistical and conventional abnormality consist of grossly observable behavior or anatomical variations. This is no surprise because conventional judgments about what is normal and abnormal (unlike theoretical or even statistical claims) usually only apply to what members of the culture can and do observe.

61 In psychiatry the DSM is constructed to be atheoretical and lists disease diagnosis merely as sets of necessary and sufficient conditions. But much debate and research occurs in the background and concerns attempts to legitimize certain diagnosis and determine which will get added and which dropped from each DSM revision.
situation is similar to scientific models of normal human aging and its lack of extensive satisfactory explanation. In many ways the definitive model of normal human aging (along with possible abnormalities) and the model of normal human development from a zygote to a adult would just be parts of the same overall theory of normal development, normal shape and function, and normal decline. But various parts of this complex model are not yet well worked out.

Figure 3.2 shows another speculative, but interesting exercise in trying to organize along a spectrum of anatomical variations judged to be conventional abnormalities by most cultures. The upper end of the spectrum represents development in accord with normal variation as determined by some model of embryological development and for which there is not a specific model of pathology. The bottom end represents those whose development is a theoretically abnormal variation—a malfunction—backed by some model of pathology of embryological development, endocrinology, etc. These cases tend to blend together and present real problems for determining what is a disease versus what is merely a medicalized bias against certain variations. The point I want to highlight is that a strong influence on intuitions about whether these cases are diseases or not is the belief in (if not the actual existence of) unbiased and accurate explanations of the condition as a theoretical biological abnormality.

Height is a grossly observable anatomical variation and extreme shortness is an example of a culturally undesirable statistical abnormality. Certain cases of shortness (like certain cases of atherosclerosis) have successfully been supported by a molecular biological explanation of its cause—for example, the lack of human growth hormone or malformation of some receptor or component in the hormone’s signaling cascade (see Rosenfeld, 2002 and Kamboj, 2005). Such cases of shortness have been made consequences of theoretical abnormalities of endocrine function.62 These cases of pathology causing short stature are cases of the successful move from the right-hand area of double overlap to the triple overlap area on Figure 3.1. Other cases of shortness have

---

62 Shortness is just a symptom of the disease, but it is the reason that the scientific model in question was worked on and elaborated, and it is ultimately the reason for medical intervention.
no theoretical explanatory models and tend to remain merely statistical abnormalities. Such abnormally short people are considered healthy and merely at the lower end of the distribution of heights. That some individuals will be labeled as suffering from a disease due to their height and medically treated, while others of the same height will be labeled normal and not helped, is controversial and *prima facie* unjust.\(^{63}\)

---

**Figure 3.2**

```
Variation within accepted models of normal development

Abnormality in reference to accepted models of normal development
```

- Ears that stick out too far
- Small breasts
- Shortness/obesity
- 6 fingers
- Missing limb
- Cleft palette
- Spina bifida
- Conjoined twins

Increasing availability of the means to alter biological systems and especially to alter gross anatomical structure, function, and behavior, will create even more pressure to

\(^{63}\) See Buchanan, et. al., 2000. My own interpretation of this would be that for some cases the theoretical model of malfunction and normal function is well worked out and for other cases it is not yet available. The boundary between theoretically normal and abnormal usually has a significant area of indeterminacy. It also seems to me that not all cases of short stature can be symptoms of a theoretical abnormality. The decision to treat short stature should be made, however, on more than the fact that it is or is not a symptom of a legitimate disease.
medicalize certain problems. Often these cases will involve claims about normality and abnormality—often claims in reference to statistical and conventional conceptions—and a search for a theoretical explanation that legitimizes medical intervention. Being aware of the possible equivocation across the concepts of normal and abnormal, and about the pressure to create a theoretical abnormality out of conventional ones, is an important part of analyzing these controversies, especially during periods where a consensus regarding the accuracy or reliability of the scientific models appealed to is still unavailable.

3.6 LEVELS OF ANALYSIS

The medicalization of problems and the social construction of diseases are more likely in cases involving certain mixtures of the three senses of abnormality. I noted above that claims about anatomical variation or behavior are particularly problematic in these regards. It is no coincidence that scientific models in fields of research that deal with such grossly observable phenomena often arouse more controversy. At certain ‘higher levels’ of analysis there is more intermingling and competition between the different senses of normal.

In biology differences might exist in explanations and models as phenomena are described at different levels—ascending from the atomic and molecular, to the cellular, tissue level, organ level, organism level and finally community and population levels. Different methodologies are involved in investigations at different levels. In evolutionary theory one might talk about selection occurring at the molecular level of genes and proteins, at the level of whole organisms, or at the level of populations of organisms. In the hey-day of support for ‘functionalism’ in cognitive science, research was distinguished by whether it described cognitive operations at the experiential/behavioral level, the functional level, or the instantiation level (see Pylyshyn, 1984). Different vocabulary and different sorts of explanation, as well as different sorts of criticism, were appropriate at different levels of abstraction. Talk of ‘levels of analysis’ arranged from those phenomena closest to those farthest from simple human observation is somewhat crude, but helps highlight that certain phenomena at one level of analysis might require
quite different kinds of scientific explanation than phenomena at a higher or lower level of analysis.\textsuperscript{64}

As one moves ‘up’ and out of physiology, and into either gross structural and functional anatomy of the entire organism, or into human behavior, cognition, or social structures, the three senses of abnormal often become entangled. At these higher levels, even when science does offer virtuous models, the phenomena are observable and scientific models frequently have to compete with folk theories. At these higher levels there are also direct influences on scientific modeling from conventional notions of what is normal for human anatomy, function, behavior, and cognition.

On the other hand, as one moves ‘down’ in levels of analysis, increasingly towards molecular interactions and particle physics, the postulation that there are pathological states becomes less prevalent in model building. At these lower levels the as-if-designed metaphor is employed less frequently. Some phenomena don’t seem appropriately explained as having a normal function that can be disrupted in various ways. For these systems, aberrations in the behavior of the system don’t seem so much pathological as they seem merely other possible states of operation consistent with different conditions. Such models often postulate large numbers of random interactions, governed by general rules, giving rise to mathematically predictable emergent states of affair. At these lower levels of analysis the statistical sense of normality is the dominant one. Even when some states are called normal—such as equilibrium states—this designation does not carry the same sort of teleology that we observe when a biological system is classified as functioning normally in opposition to states that are abnormal.

There is speculation and debate about whether functional analysis is only applicable to certain kinds of phenomena or at certain levels of analysis. Furthermore, some argue that biology is a special field with special kinds of explanation due to its

\textsuperscript{64} Rather than placing the phenomena observable by biologically endowed human sensory capabilities at the top end of the spectrum, it is more accurate to make it the middle. There are all sorts of large-scale phenomena (both large scale in space and time) incapable of being directly observed by normal human sensory capabilities. For example, levels of analysis ‘above’ those of gross human observation are required for economics and weather forecasting. These are, like very small scale phenomena, more difficult to gather information about or to control experimentally.
Perhaps there is a middle level of abstraction where functional ascription is most appropriate and theoretical normality is least problematic. These are very interesting and complex issues. There is not a line where one passes from models that create a robust functional sense of normality to those that rely mostly on a statistical sense. Likewise there is no clear-cut boundary where conventional senses of abnormality stop having influence. I would defer to examples of our best theoretical models to help resolve these issues.

In assessing the concept of normality, Jiri Vacha, points out both the ubiquity of the concept and its vagueness. I agree that there are different broad senses of normalcy and abnormality, and many subtly different variants of theoretical abnormality across scientific contexts. Different kinds of theoretical normality are generated by different kinds of models in science. Vacha advocates dropping usage of the term ‘normal.’ The situation is not as hopeless as Vacha and others indicate. Evaluating particular cases carefully can clarify what is being claimed and why certain problems are occurring.

Points to keep in mind when evaluating individual usages of normality in biology are: a) the three broad senses of normal specified above; b) that these senses are often intermingled; c) that the level of analysis at which a model is operating at can influence this intermingling; d) that the statistical sense of normality often does not generate as strong a normative force as the other senses; e) that the accuracy and objectivity (as well as the character of theoretical normality being generated) is related to the quality and details of the scientific models making those claims—accurate well verified models, in agreement with other core results in that field, and in a field that has a well entrenched and respected core of results and established methodologies, will often generate senses of theoretical normality that are unproblematic and unquestioned; f) that particular cases of theoretical normality can be different as they are relative to the model generating it and the details of how the model explains the phenomena it is representing; and g) that the politically or ethically controversial aspects of certain claims of normality often involve the conventional sense and that these are often cases of grossly observable behavior or

---

65 I believe this is actually a reasonable explanation of why teleology seems more or less prevalent in certain areas of science, namely whether the predominant sense of normal at work is statistical versus functional.
anatomical variation. In order to demonstrate how easy it is to overlook these considerations, I will discuss an article by Ron Amundson (2000), entitled Against Normal Function.

3.7 AMUNDSON ON NORMAL FUNCTION

Ron Amundson is a well-respected philosopher of biology, who has written on both the history and theory of evolution and developmental biology. His essay, Against Normal Function, argues against labeling individuals as ‘abnormal’ because doing so unfairly marginalizes them by “use of a falsely objective criterion.” He argues that the concept of ‘normal function’ does not reflect a ‘biological reality’ in the same way that concepts like species or sexual phenotype (male/female) do. Normal function is more like the concept of race—a categorizing concept that represents the social practices and conventions of the community more than it represents naturally occurring patterns. Thus ‘normal’ and ‘abnormal’ function are social constructs. Amundson argues that biological research makes the notion of ‘biological normality’ look very suspect. Moreover, it playing a role in justifying discriminatory practices against those with variant modes of function (i.e. the disabled). Regardless of being ubiquitous in scientific model building, debates about the nature of disease, and debates about the distribution of health care, he argues we should avoid the term in biomedical science.

The details of Amundson’s argument are as follows. If one looks closely at biological phenomena, whether evolutionary biology, developmental, or examples from anatomy or physiology—not to mention studies of human performance—one finds that, for many given functions, there is much variation in both the mode of functionality and

---

66 There are people who argue that both the concepts of species and sexual phenotype are not objective, scientific concepts—that they too are importantly similar to the concept of race. I believe that both the concept of species and sexual phenotype are complex but that our best current scientific models explain these as objective categorizations, even though (according to these models) borderline cases exist and processes occurs that work on small variations and produce new species.

67 Later in this chapter I shall explain what I think the charge of social construction, especially as it relates to diseases, amounts to and in terms of the different senses of normality.

68 Amundson distinguishes the level of functional performance that might be accomplished from variant modes of function. Two individuals might transport
the level of functional performance. These empirical biological facts are counter to any
theory of significant uniformity of human biological functions. There is no evidence for
species typical functions or normal functions for the species. The notion that variant
functionality is abnormal is also counter to the facts of biology. Normality and
abnormality are not objective biological categories. They are social constructs like race or
religious sect. Because they have been used to justify discrimination against variant
modes of function (e.g. preferring lip reading and attempts at verbalization over sign
language use) they should be avoided in careful scientific explanation. 69

The formal notion of normality that Amundson has in mind he calls ‘functional
determinism.’ He identifies this theory with Christopher Boorse (1997). Functional
determinism implies that each species has a sufficiently narrow distribution of
performance on certain species-defining functions to justify a dichotomy between normal
and abnormal species members. 70 In other words, labeling a subset of species members as

69 Amundson is a normativist, and not a naturalist, about the concept of normality. He
says: “The topic of biological normality is related to a philosophical debate on the
concept of disease. Naturalists consider disease to be a straight-forward, non-evaluative,
theoretical concept within the sciences of medicine and physiology. Normativists
consider disease concepts to embody evaluative judgments of the conditions designated
as diseases. Much of the present paper is an argument for the normativity of the concept
of functional normality, at least as the concept is currently used… My purpose is to show
that the normative taint is not avoided in current discussions of biological normality
(Amundson, 2000, p. 34).”

70 Amundson says: “Boorse’s two definitions imply that natural species have a certain
statistical characteristic: the variations in function among their members is sufficiently
narrow to justify a dichotomy between normality and abnormality based on the
distribution alone. Obviously not all species members function in exactly the same way.
We can treat them as if they do by labeling as abnormal any non-conforming species
members. This labeling is statistically justified only if the bell curve of functional design
‘normal’ will only be justified if the Gaussian distribution of performance for the pertinent set of functions is a narrow one—i.e. “that there are many uniformly designed individuals and only a few scattered individuals with novel functional design.” This is a statistical claim that species tend to have minimal variation in the performance of some set of pertinent biological functions. Being biologically normal is to be within the narrow limits on these pertinent measures. Add that this statistical claim represents some objective fact about biology and we have the target of Amundson’s criticism. He says: “…[F]unctional determinism states that functions take place in a uniform mode at a relatively uniform performance level\(^{71}\) by statistically distinctive portion of the members of a species. These are the normals (Amundson, 2000, p. 36).”

To support the premise that actual biological phenomena do not provide evidence for the doctrine of ‘functional determinism,’ Amundson gives examples. He argues that variation is the norm among members of a species. It arises not just from genetic variation, but also in variations due to developmental plasticity—i.e. the interaction of genetic predispositions and environmental factors as complex organisms develop form zygotes to adults. Amundson reminds us that evolution could (and probably does) create species whose members grow to be as alike as machine-made paperclips. It also allows for more complex multi-tissue organisms whose adult members have great variation in functionality due to built-in options allowing development to mold structure and function so as to meet the demands not only of the environment, but also the demands created by the interaction of widely variant co-functions mixed together. “…[F]unctioning adults can develop in an indefinitely large number of ways. The goal directedness seen in developmental plasticity renders the concept of species design highly suspect. Development yields adults that function, but not adults that function identically.

\(^{71}\) Amundson makes a distinction between mode of performance and level of performance. Functional determinism requires lack of variation in both. For example, assuming vision is one of the pertinent functions humans perform, particular humans would need the same level of performance in visual acuity and need to accomplish this via the same mechanisms.
Functional diversity is a product of developmental plasticity (Amundson, 2000, p. 39).” He also says: “Biological ‘types’ are unified not by the functional identity of their eventual phenotypes, or the common blueprint from which they were built. Rather they are unified by their shared developmental processes. These processes generate phenotypes that are functionally diverse, both between and within species (Amundson, 2000, p. 40).” Amundson admits that ultimately this is an empirical question about evolution and speciation, but he believes there is already enough evidence to make functional determinism not the case for many species, and especially not for complex organisms like humans.

He cites an example of a goat born without its two front limbs, and with several other, apparently compensating, anatomical variations, such as an atypically broad neck, S-shaped spine, and atypically positioned and developed musculature. This goat could walk bipedally and had adapted other functions to allow it to operate in its environment. He also discusses patients with hydrocephaly whose brains only develop one hemisphere, or are greatly reduced in size, but which function quite well due to neural plasticity. Amundson discusses the fact that nerves in primates can be grafted onto new target muscles and adapt to function quite well after such transpositioning. He also cites the functional diversity of disabled wheelchair basketball players. Even players with what appear to medical experts to have the same level of disability often display quite different levels of performance and talents on the court. He recalls the history of how expert medical assessment of ‘handicapping’ had to give way to actual performance based ‘handicapping’ of players. Sign language as a functional variation on spoken language is also discussed. Amundson sums up these examples by saying: “…the goal directed processes of biological development are not finely tuned towards the production of functionally identical species members. Their inherent flexibility can be expected to generate a rich diversity of functional modes (Amundson, 2000, p. 43).”

For my purposes, examples from anatomy and physiology are of particular interest. Amundson says:

In fact there is a multidimensional continuum of states of health. The health/illness and normal/abnormal dichotomies are illusions. A high degree of variability exists among individuals on any physiological measurement, with even
the most extreme values found within healthy individuals. Extreme values of physiological parameters, associated with disease in some individuals, are compensated for in others. Indeed, the constellation of other parameters in an individual may directly require the extremeness of a particular character for good health. …Functional integration at the physiological level gives rise to a range of differently functioning, but comparably successful physiological systems. …One would expect the large variability of muscle and tendon positioning in the hand, for example, to correlate with the level of strength or dexterity in certain kinds of manual tasks. Do better violin players tend to have a common configuration of hand musculature, or one that is unusual? Do people with the best and worst penmanship tend to have certain configurations? These questions seem meaningful, but they do not draw us towards a robust concept of ‘normality.’ We always knew that people varied in their manual abilities, and now we know that they differ in musculature as well. …Skills in penmanship and musicianship are so various that no one seriously thinks there are ‘normal’ ranges here. There is no reason that discovering a biological explanation for variations in functional performance should cause us to declare certain performances abnormal. …If medical textbooks emphasize average or typical cases, there may well be pragmatic reasons to do so. It would be a mistake to infer from this that diversity constitutes abnormality (Amundson, 2000, p. 45).

He further illustrates his points by drawing attention to a very interesting internet based project that documents human anatomical variation called the Illustrated Encyclopedia of Human Anatomic Variation. This is basically a repository of reported variations in the anatomical origins and insertions of muscles, and also in the course, branching and targets of nerves, arteries, and veins.

Of note, in the quote above, Amundson implicitly endorses an older, Aristotelian notion of health as a balance of parameters. Of more importance, Amundson discusses what is essentially a statistical notion of normality and abnormality—a notion based on the variation of measured parameters or performances. Amundson argues that variation is so ubiquitous that selecting some individuals for the label ‘abnormal’ doesn’t accurately represent biological phenomena as much as represent human judgments of approval and disapproval. He also mentions the possibility that basic sets of functional elements at lower levels of analysis might combine in different ways to achieve similar levels of performance at higher levels. Amundson advises we abandon the terms

---

normal/abnormal and health/illness in precise discourse and talk of variation of function instead.

3.8 RESPONSE TO AMUNDSON

I disagree with Amundson that the science of biology should do away with the concept of normality. For example, I see no possibility that the model of the thyroid could do without distinguishing between normal and abnormal thyroid functions. The fact that the gland has a normal function and structure is precisely what the model of the thyroid is explaining. The model abstracts away from the infinite amount of detail that could be described and generalizes to a stable representation of basic function. This includes normal ranges of functional performance. My main thesis is that the content of the concept of disease (taken broadly to include pathology and trauma) is basically theoretically specified abnormal form or function. The legitimacy of disease claims rests on the accuracy and reliability of the model (or rather the judgment, by the community in question, of the model’s virtues). Are we mistaken that ‘normal function’ is a concept essential to explaining biological systems? Presently the concept of theoretical normality is both ubiquitous and seemingly indispensable to medicine and biological science. It is taught in medical schools, discussed with patients, and appealed to in policy debates. Regarding empirical and practical issues, the model of thyroid function works well in medical clinics, hospital wards, and research labs. It is difficult to even imagine what biomedical science would be like without use of the concepts of biological normality and abnormality. The burden is on those who believe science could do without the concepts.

Amundson fails to properly take account of the different possible senses of normality. At times, he seems to understand that there is room for slippage between different senses of normality. He says:

In this paper I will use the term ‘typical’ not as a synonym for ‘normal,’ but in the colloquial sense of common, usual, or frequent. On my usage a ‘typical’ trait may be merely the least unusual, and an atypical trait need not be abnormal. This convention allows the discussion of typical and atypical traits without assuming that they are respectively normal and abnormal. It should also be noted that, as with other quasi-statistical uses of the concept of normality, abnormality is usually to be read as subnormality. Better-than-average function is not usually labeled as abnormal even though it is statistically atypical (Amundson, 2000, p. 35).”
Here Amundson seems to be aware that there is a statistical sense of ‘typicality’ that does not carry the same amount of normative force. However, he takes particular aim at a formalized statistical conception he calls ‘functional determinism,’ and which he finds in the work of Boorse and other philosophers in the naturalist camp. When he begins describing the discriminatory practices associated with the labeling of some individuals as normal, and when comparing the concept of normality with race, it appears the target is more of a conventional notion of ‘normal persons.’ This failure to carefully disentangle at least the three senses I have outlined produces equivocation.\textsuperscript{73} That ‘functional determinism’ is not true of biological phenomena does not necessarily mean other notions of normality are problematic or poor representations. Likewise, just because some conventional senses of abnormality are discriminatory, or poor representations of biological phenomena, does not mean that all (or any) theoretical senses of abnormality are suspect. The thyroid gland and pancreas have normal and abnormal functions and these are described in excellent scientific models with little bias. These models are not used to discriminate against those with atypical modes or levels of performance.

In the philosophy of medicine the biostatistical model of disease (Boorse, 1977) is the most widely discussed conception of biological normality and abnormality. However, this does not mean that there is one conception of normality in biomedical sciences. The sense (or senses) of normal generated by the models in question must be analyzed on a case-by-case basis—just as they must be examined individually when assessing

\textsuperscript{73} Amundson could grant the three senses I outlined, and could argue that there is no theoretical sense of abnormality, but only various conventional senses, and that science is better off by abandoning the concept in favor of talk of differing levels of variation or uniformity. The issue of how to distinguish theoretical senses of abnormality form merely conventional ones is difficult, but would involve appeal to the scientific method and the process of sifting out those models with little bias and with accurate representations (i.e. that have good fit). Theoretical senses of normality are produced in scientific models with the purpose of describing natural phenomena objectively, and there is a process of evaluation that attempts to assure their accuracy. This issue is one of the main points of contention involved in disputes about the extent of social construction in medicine. Strong claims about global social construction in medicine hold that there is nothing but conventional senses of normality. In the next section I shall discuss the charge that a putative disease is merely a social construction. Here it is enough to realize Amundson does not fully acknowledge the different possible senses of normality.
representational accuracy. A theoretical model specifying normality can involve varying degrees of statistical normality—in some models, as part of the explanation of the phenomena, normal parameters are determined, often so that normal function can be assessed by proxy. My point is that not only are there broadly different senses of normality as discussed in the previous sub-sections, there are also many different types of theoretical normality depending on the details of the model. Selecting one definition of biological normality and criticizing it does not necessarily implicate any others.

The fact that there can be significantly different notions of normality across different modeling contexts in biological and psychological science, also answers the challenge posed by Jiri Vacha earlier in this chapter. Vacha assumed that because there were differing uses across contexts in the sciences, that the term out to be dropped. The broad conception of theoretical normality in biology is flexible and will reflect the needs of a particular model in describing a normal state of function or form for a system.

By attacking ‘functional determinism,’ Amundson is attacking a particular statistical notion of normalcy. His argument is that biological phenomena demonstrate too much variability of function to allow this statistical sense of normality. He makes normalcy dependent on a lack of diversity in both mode and level of performance. His examples are meant to show that such a lack of diversity does not exist. It is a difficult issue deciding how much diversity and variation in function amongst species members (or anything for that matter) constitutes too much to prevent a claim of statistical normality. Just how narrow must the distribution be? How many outliers are too many? Does a bimodal distribution, even if the component means are close, rule out the possibility that everything within 2 standard deviations of the composite mean is normal? Why 2 standard deviations and not 1? Ultimately these decisions are determined by those doing or applying the research. Boundaries of what to consider normal and abnormal, even in a statistical sense are determined by the interplay of researchers, the reasons for examining the phenomena, and the process of model building and model refinement.

74 There are countless examples in clinical medicine—many of them involve looking for levels of certain proteins, sugars, or electrolytes in the blood. One example is the use of so called ‘liver function tests.’
Different contexts are allowed to have more or less variation. But even if Amundson is correct that ‘functional determinism’ is unlikely because of his examples, this is just one sense of statistical normality from the philosophical literature and its refutation does not affect other notions of statistical normality that might be more complex. The success of Amundson’s argument has no impact on the majority of uses of theoretical functional normality created by scientific models in biology and elsewhere.

Amundson examines variation in the actual phenomena. He discusses variation in measurable aspects of the phenomena. Theoretical normality is in reference to the simplified model. The model is an abstraction and often a simplification of the many aspects of the phenomena that might be described—it is perspectival. This is a subtle point, but an important one. Statistical normality more directly refers to measurable aspects of the phenomena. Variation in the phenomena may prove too much for a statistical sense of normality to be plausible, but scientific models abstract and simplify, or expand and become complex, to accommodate variation.

Science can handle variation. Sometimes variation is noted, explained away as largely unimportant, and the model simplifies over this variation. In other cases, models account for the fact that several variants of ‘normal function’ are possible, perhaps with each variant predisposed to malfunction in different ways or with each variant at risk for specific malfunctions to different degrees. The negotiation between generalizing across or cataloguing variation is a tension felt in most models of complex biological systems, but negotiating this tension successfully is part of what allows scientific models to organize complexity and serve as useful explanations. There is a series of models

---

75 Similar situations arise in determining diagnostic parameters in clinical medicine—controversy sometimes exists about whether a biomarker’s variation can be demarcated into levels or ranges that are ‘normal’ or correlate with the absence of underlying pathology. Prostate specific antigen is one recent controversial example.

76 This is a similar point to one made in the last chapter and a general point I will make again in the chapter 4, namely that oversimplifying a crucial concept for the sake of an easy definition, and for a definition that allows clear boundaries to be set, is tempting in philosophical analysis because it appears to solve some philosophical puzzles. However, the drawback is that the analysis is usually irrelevant when one turns to the much richer concepts actually being employed by practitioners or those dealing directly with the phenomena in question.
explaining red blood cell (RBC) structure and function. Some of these models handle variation by giving several normal possibilities—such as for the different ABO blood types, or even the hundreds of other rarer or less medically relevant blood types. There is also a theory to explain the variation—a theory of how these different blood types have originated in an immunological arms race with bacteria attempting to mimic the RBC surface proteins and thus evade immune detection. These different blood types may actually be distinct regarding immune function, yet still considered by the model to be normal variants. Currently, regarding these surface proteins there is no sense of normal blood type only more and less rare, but there are normal and abnormal types of hemoglobin and other enzymes in RBCs—the presence of abnormal hemoglobins leads to diseases such as sickle cell anemia. Eye color may be another example that is handled

---

There are statistical measures of the average size and other average properties of human RBCs and some of these statistical parameters can be measured in a CBC (complete blood count). This test is used routinely. Combinations of values outside the normal range, along with other abnormal tests or symptoms can lead to the diagnosis of underlying malfunctions (i.e. anemias).

As of 2008 the International Society of Blood Types recognized 30 different blood type systems of which the familiar ABO system is the most widely known and used (because it is involved in the explanation and prevention of the most dangerous transfusion reactions). See: http://www.isbtweb.org/working-parties/red-cell-immunogenetics-and-terminology/members-only/

But even if this microevolutionary model were inaccurate, it would not change the details of models explaining the normal antigens found on the surface of human RBCs. Things get very complicated. A number of pathologies (i.e. diseases) can occur when the wrong blood type is transfused into a person who cannot tolerate that blood type. The theoretical abnormality here is not the structure of the RBCs or the antigens on RBCs that elicit the reaction, but rather the contextual fact that the wrong type of RBCs entered the body of a person primed against these antigens. The antigens that are actually recognized as problematic by the immune system are normal. Furthermore the immunological reactions that cause the pathology are operating normally (i.e. as if a foreign invader was trying to contaminate the blood). What is abnormal is the fact that this immune reaction occurs in an attempted transfusion—it is an abnormality in reference to the model of how blood transfusions work (a mistake often on the part of the blood bank or physicians involved). Hemolytic disease of the newborn would be a variant of this same complex kind of abnormality, only here there is no human error—no context of transfusion—but one arranged by nature. Even in this setting, the abnormality involves normally operating immune reactions against normally structured RBCs—it just that the context for the reactions and the target of the reaction are abnormal according to the models of normal human childbirth.
by allowing variation without any sense of normal and abnormal, only more and less common.

Humans tend to grow and develop in remarkably similar ways, but also with variation. Medicine and biology abstract from this to create a series of integrated, hierarchically ordered set of models that specify normal and abnormal functions for the human body. This allows the models and theories of medicine to apply generally to all people. But medical students and physicians know that these models are abstractions and are perhaps even aware that the sense of normal and abnormal is generated by the models. Practitioners of medicine gain a feel for whether the models have good fit or need refinement, both globally but also in particular cases. Humans sometimes vary in form and function. On my theory this is no problem, and is even to be expected. Theoretical models can become as complex as they need to in order to describe the pertinent aspects of the phenomena in question. There are many examples in medicine where complexities such as this are built into existing models of normal and abnormal structure and function. Walking through the library while editing this chapter, I happened to see an example—a large text entitled: *Atlas Of Normal Roentgen Variants That May Simulate Disease, 8th Ed* (Keats, 2007). This book actually contains very little text and is 1200 plus pages of x-rays or CT images. Figure 3-277 shows three examples of variability in the size of adult cervical vertebral bodies, and Figure 3-278 shows marked variability in the size of the cervical vertebrae of a single individual. Many of these images appear to be the radiological evidence of an abnormality, but are created not by instances of that specific pathology, but rather by benign conditions. Figure 3-292 shows large transverse processes with attempts to form ribs at vertebrae C5 and C7, and Figure 3-293 shows long transverse processes at C6 simulating a fracture. These benign conditions (or at least more benign than the condition that the radiologist might suspect but be mistaken about) are often cases where no other known theory about embryological development, no theory about trauma or malfunction, makes the finding a pathology or an abnormality. In these cases, the findings, by default, are variants of normal. This default classification is open to re-interpretation as more information is gathered about
just what is causing these radiological findings, and how the biological system in question came to be that way.\textsuperscript{81}

What is important to gather from this example is that there is a tremendous amount of theory and iterated and interrelated scientific models appealed to in determining exactly what normal anatomy is, and how that normal anatomy appears on an x-ray or CT scan. Even the ability to read an x-ray to determine if it reveals a normal anatomy or an abnormal one requires a tremendous amount of theoretical knowledge. When the medical student, or even the expert radiologist, pronounces that an x-ray is ‘normal’ he is referring to a sense of normality that is generated by these theoretical models—the sense of normal refers back to these models—the individual patient being evaluated is being compared against the theory of normal human anatomy and being judged normal or abnormal. This is subtly but importantly different than when I see a paraplegic in a wheelchair and, compared against what our society takes to be normal modes of ambulation, and normal levels of personal mobility, I judge this individual to be abnormal. Because of the complexity of science, our society, and because of the difference in qualities of scientific models, there is room for blending of these two types of judgments and they can sometimes be difficult to untangle.

The frontier of research is always being pushed. New areas are explored and models proposed. Similar to the question about how much variation is too much to allow a claim of statistical normality, the issues of how much variation is required for pathology or theoretical abnormality is also difficult. But these decisions are made on a case-by-case basis and models which specify where normal variation ends and pathology begins are proposed, tested, and either refined or rejected, over time, by the scientific community. These issues often raise difficult challenges for specific notions of theoretical normality used in certain models or even used by entire fields. The fields of genetics and genomics labor under these difficulties currently. The human genome project is an attempt to model the normal human genome—but many questions have arisen about the distinctions between normal variation and pathology. This is all the more complex

\textsuperscript{81} Again, the situation can get very complex as theory about how three dimensional energy dispersion is turned into one dimensional pictures becomes involved. But the contrast between normal and abnormal remains throughout the escalating complexity.
because the genome is a historical repository of older changes, and also a theoretical object which undergoes changes slowly over evolutionary time periods. Built into the theory of the genome is the idea that variation serves as the building blocks for eventual shifts in what is considered the normal character and constituents of the genome itself. Furthermore, the model of the genome is an abstract theoretical entity that represents the general human genetic make-up, but was actually constructed from the DNA of only a small number of donors. Ultimately, however, the issues concerning variation in the genome will not differ from those already faced by anatomy.

The models that constitute human anatomy have many of the virtues that I claim it is possible for models and fields of research to have—it has been, and continues to be, taught largely unchanged for at least 300 years, and it squares well with practical experience in kinesiology, sports medicine, and surgery. It is also hard to imagine how the core of scientific models and explanations in anatomy and embryology could be altered in the foreseeable future. Examples include mundane facts about how many fingers a hand has; the gross structure and function of the heart; the number, shape, location, and age of complete ossification of bones; and what basic motions certain muscles can accomplish, where these muscles should be located (origin and insertion) and what blood supply and enervation they have. The field also deals with the location and functional organization of the organs. Anatomy atlases and textbooks illustrate and explain the anatomical structure of the theoretically normal human male and female, as infant, child and adult. Anatomy gives details about what muscles and nerves are required to be working properly if a person where to pick up a pencil off a desk, and it describes the effects of nerve, muscle, and tendon damage that would prevent the performance of such a task. These details are provided in terms of the range of motion and details about extension and flexion at a joint. But the core of the theory does not discuss things such as the variation in musculature in the hand or forearm that might make for better violin playing—although it potentially could. The standard theories of anatomy address human performance only at the level of very basic function.


In recent years study of how maximum athletic performance varies across different types of bodies has grown. However, these explanatory models in biophysics make few
Standard theory does record variation, such as innervation of muscles, and in the path and branching of blood supply, but such variations usually, more or less, follow one of a handful of possibilities—and the theory allows these so called ‘normal variations.’

The student learning normal human anatomy by dissecting cadavers is at first troubled by such variations. The student finds that each cadaver is not exactly the same. However, as the student learns the theory, she increasingly can recognize and demonstrate the normal structure and function of parts on all cadavers. Furthermore, as the student begins to be able to apply the theory learned in textbooks, the student can identify examples of normal variants in actual human bodies being dissected. Eventually, the experienced surgeon relies on these theoretical models of normal anatomy (including knowledge of normal variants) to carefully dissect through living tissues. The surgeon locates where she currently is cutting and where she is going to cut by using landmarks and relationships learned as theory and tested by experience. This allows the sacrificing of insignificant structures while avoiding damaging crucial nerves or arteries.

It is possible to measure certain aspects of human anatomy, functionality, development, or cognition and make claims about what is above and below average, but this is largely a statistical sense of normality. This is different from developing scientific models offering a theoretical sense of normal states of gross human anatomy, normal development, performance, and cognition. Of importance is the fact that such models do not extend very far into explanations of human aesthetics, functionality, and behavior. It is not impossible to imagine that someday scientific models extending into these areas might exist and have the virtues discussed in the last chapter. Scientific speculation in these areas, however, has not yet produced products good enough to completely displace religion, philosophy, politics, or popular culture as the authority in speculating on what is normal or proper in human appearance, behavior, cognition, etc. And in some areas, such as the normal and the abnormal shape of the human face, it may not be possible to develop a theoretical sense of normality completely devoid of conventional senses, even

claims about ‘normality.’ On the contrary, they examine extraordinary performance and look to build explanatory models of how such performance occurs.

84 A classic example is the ‘right dominant’ versus ‘left dominant’ heart, based on which coronary arteries predominantly supply the left ventricle.
if science develops descriptive explanations for what are the normal conventional views we hold and why our minds are designed to hold such views as opposed to others.

Most of Amundson’s examples are from higher levels of analysis—from grossly observable behavior, abilities, or anatomical structure. As discussed earlier in this chapter, any scientific model explaining phenomena at these higher levels will have to compete with conventional folk wisdom and prevailing social values in explaining what is and is not normal. When describing such phenomena one must be careful to untangle the differing senses of normal at work. Amundson did not do this with his examples.

To highlight some of the particular problems with anatomical senses of abnormality or disease consider the example of obesity. There are conventional norms about body weight in various cultures and societies. In the past few decades there has also been increasing focus by medical professionals about the adverse effects of obesity and the correlation between increasing weight and poorer outcomes for a number of diseases such as atherosclerosis, type II diabetes, and various kinds of cancer. Medical professionals have developed the concept of body mass index as a measurement, and certain intervals of this measure have been designated normal and others underweight, obese, and morbidly obese. These are basically statistical notions of normality (with the boundaries influenced by conventional notions). Research into why some individuals seem to be particularly predisposed to obesity has also generated scientific models of the hormonal communication between the digestive tract, adipose tissue, and the brain—communication that when working normally allows a person to feel satiated and no longer hungry because a certain amount of food is ingested or energy storage is achieved. These models have also detailed certain genetic mutations that disrupt this system (see Grimm and Steinle, 2011). There are theoretical models of pathology that result in morbid obesity, however there are also many examples of obesity for which there is not a scientifically well established explanation. Some people who have attained obese states (i.e. achieved a BMI over 30) have also started to view their condition as a disability and appealed for accommodation and even filed lawsuits to be protected from discrimination.

This case is one where an anatomical feature may be in various lights be considered a biological abnormality or related to an abnormality. It could be claimed that
having a weight far outside the usual measures is a statistical abnormality.\textsuperscript{85} Obese persons are often judged to have a body habitus less desirable than others, which is to say that many considered it an abnormality in a conventional sense.\textsuperscript{86} It could also be claimed that obesity is a treatable disease. What this claim amounts to depends on one’s interpretation of the nature of disease, but the intention behind the claim is most likely to represent obesity as a theoretical abnormality—an abnormality that is determined in reference to a presumed accurate scientific model. The problem with the claim that obesity itself is a theoretical abnormality (and not just the symptom of an abnormality) is that the scientific model in question would have to be one concerning human anatomy and build—a science similar to the ‘constitutional theory’ Vacha discusses being debated in Germany in the 1920s and 30s. Human anatomy as a discipline has collected much data on measurable qualities of human bodies and has built a basic model of human anatomy and function, but one whose level of detail is quite basic and reflects things like which muscles are involved in the basic movements of the body. Statistical data could be collected and a normal range might be selected (as has been done in the case of BMI), however this is not quite the same as creating a theoretical sense of abnormality that would make obesity an unproblematic and convincing case of disease in and of itself. What is needed is a scientific model that explains the operation and function of some circumscribed biological system in terms of normal operations and functions and various ways in which dysfunction and poor operation can set in. If all cases of obesity could be explained this way, and explained by malfunctions in the same system, then obesity could be said to be a disease—or more likely, a consistent, observable sign of a disease. However, only a few instances of obesity can be explained as by malfunctions in a known biological pathway or system. In these cases, obesity is a sign of disease. However, many other cases of obesity do not have such a theoretical explanation with a model denoting normal and abnormal function. The charge that a non-medical problem is being

\textsuperscript{85} However, if more and more people achieved a BMI above 30, the sense in which this was a statistical abnormality would be diminished even if we decided as a community of health care professionals to retain the theoretical sense in which BMI above 30 was unhealthy and an abnormality.

\textsuperscript{86} One could easily imagine contexts in which this was not undesirable; a community that found obesity a sign of wealth, success, happiness and divine blessing. Consider why Buddha is depicted as obese.
medicalized is common in such circumstances. The situation is similar to the issues with short stature and could exist for tall stature. One difference is that we believe (conventionally) that personal choices about diet/exercise can influence weight in ways we do not believe influence height. Low body weight is another anatomical condition potentially seen as disease or seen as the medicalization of a problem not directly caused by biological dysfunction. Anatomical features can be evaluated as statistical, conventional, and theoretical abnormalities.

There are also biological and medical sciences that explain theoretically normal development from zygote through embryo, fetus, infant, child, adolescent and into adulthood. Again the core body of models in this field do not extend very far past explaining what is the normal course of basic development, providing statistical data on the average or typical milestones and pace of changes, and highlighting processes that are disrupted and lead to identifiable syndromes and diseases. There is not a comprehensive theory of normal development yet, but slowly science is building on what is known. As this work is completed, explanations of what are normal aspects of development, including normal or innocuous variations, and details about abnormalities and diseases, will be part of these models. It is also not unreasonable to expect that science will slowly make progress developing models for normal aging and deterioration of function as one ages. The lack of theoretical models for age-appropriate deterioration of biological function and form is currently a problem for determinations of disease. Where to draw the line between disease and normal aging is difficult. Culture and statistical surveys currently have more to say on the matter than biological theory. Therefore, one must be careful in examining claims about normal and abnormal functioning regarding aging and one should attempt to separate out putative theoretical claims from conventional claims.

Another consideration about variation is the possibility of multiple realizability. Multiple realizability is sometimes at work in biological systems. Thinking in terms of levels of analysis helps keep this in mind. It may be that a higher level system like the cardiovascular system in humans can be described according to a general model of functionality with standard performance parameters, while at lower levels, details about the particular mechanisms that give rise to higher order ‘normal’ performance are different. Higher order, emergent phenomena may have little variation (under normal
conditions specified by the model) even while there is variation in both mode and level of performance across numerous lower level details of instantiation. Perhaps the only difference at the higher level between differently instantiated systems is resiliency in the face of certain unusual perturbations. Such situations are handled with models that account for such complexity and give an appropriate description of normal function as emerging from a possible diversity of contributions of subsystems or components. Models of blood pressure maintenance in the cardiovascular system are just one example. Many examples exist in physiology because the body has redundant mechanisms for maintaining homeostasis in the face of different kinds of challenge.

The majority of Amundson’s examples involve abilities or gross anatomical structures. Amundson states that he is interested in “disabilities, rather than in the episodic or life threatening conditions commonly called diseases (Amundson, 2000, p.34).” Most judgments of normal and abnormal, about such observable aspects of people, are really claims about social convention, or at least are mixtures of conventional and theoretical sense of normality. These judgments are often about the individuals who instantiate these abilities, behaviors, or gross anatomies rather than about biological parts or processes. The community judges as abnormal not the sins, but the sinner.87 This is the case for Amundson’s examples concerning discrimination. Individuals were labeled as abnormal for using sign language or wheelchairs. The community in question preferred that deaf children learn to read lips and speak words. It preferred to see those with disabilities of ambulation use modes that most closely resembled legs. It is no different than the stories of young left handed school children being physically disciplined until they switched to using their right. I would agree this was discrimination, and part of this campaign involved using the label of abnormality for those wanting to sign, use a wheelchair, or write with their left hands. Furthermore, there were historical attempts to pass off these community preferences as being scientific. There were cases of medical

87 It is interesting to consider disease being a subjective state that involves both the patient and the community judging some condition to be a particular kind of misfortune which permits the adoption of the ‘sick role,’ etc. I believe that one can easily make a distinction between theoretical normality as disease and conventional normality as perceived disease, and the distinction between abnormal biological function versus individuals currently perceived as abnormal.
authority being misused to force the preferences and social norms of some onto others in
to ways that clearly caused suffering. But it is better to be clear about exactly what was
happening in those cases—namely that conventional senses were mistakenly being
represented as theoretical senses of normal and abnormal. The remedy is not a ban on the
word ‘abnormal,’ but a careful investigation of why the label is being used and for what
purposes. When such investigations are done well we see the social construction (in a
pejorative sense) of a concept of abnormality or disease.

I don’t agree that all claims about normality are problematic in these ways, nor
that ‘normal function’ is always a social construction in this pejorative sense. I disagree
that medical science should abandon using the concept. The claim that the pancreas of a
patient who has type 1 diabetes is malfunctioning and is abnormal compared to the
normal function of a human pancreas (as explained in an endocrinology text) is not
problematic. It is not problematic in the ways that claiming certain shapes for human ears
are normal or abnormal. The normal structure, functioning, and possible malfunctions of
the pancreas are explained by scientific models built, expanded, refined, and verified over
many decades. These claims about normal and abnormal function accurately represent
biological phenomena and are not expressing social preferences or justifying
discrimination. In fact, the study of diabetes as an abnormality of the pancreas eventually
lead to accommodations that allow diabetics the ability to regulate their serum glucose
levels (i.e. achieve a good level of performance) via exogenous insulin (i.e. through a
socially less desirable mode often involving self administered injections).

Amundson’s example of the transplantation of nerves in primates and humans
(and the fact the body has enough plasticity to retain or grow back some functionality)
does nothing to show that theoretical normality as a concept is suspect. It is not clear
what this case is supposed to show. The nerves, after being moved to new target muscles,
and once they re-grow enough connections with muscle fibers, function as, or
approximate the normal function of, motor nerves as specified by the model of how
skeletal muscles are innervated. Models have been created to describe such transplants or
transposition procedures and contain descriptions of pathology that nerves used in such
procedures may be susceptible to. There is a theoretical sense of normal outcomes (i.e.
normal function) for nerves in such contexts. The statement that Mr. Smith’s transposed
ulnar nerve is ‘working normally’ would make perfect sense to both the surgeon who said it and the patient. And even though the patient might not know the details of what constitutes normal functioning in this very special situation, and even though both the surgeon and patient might be clearly aware that his overall manual abilities are conventionally abnormal, they would understand that normality here was in reference to the biomedical theories of how nerves should work after such medical procedures.

I have been arguing that by looking at the examples in Amundson’s paper it is clear that he did not do a good job of separating the different senses of abnormality from each other. He also did not consider issues concerning different levels of analysis and most of his examples are from one, particularly problematic, level. Furthermore, I have suggested that a lot of confusion can be avoided by looking at the actual scientific models generating or associated with a claim of abnormality. The particular details and the particular virtues and flaws of the model in question are important issues in deciding just what is being claimed and whether bias and discrimination are potentially being supported. What one finds is a great diversity of conceptions of normality in biology and across science, and that each claim of normality needs to be carefully examined and unpacked. By actually recognizing the complexity and variety of scientific models one realizes that it is very unrealistic to assume there is a universal notion of normality at work in biology. However, this is just what Amundson does. Such an assumption partially explains why Amundson focuses so much on variation as a problem for biological normality and why he believes that once he has shown that there is significant variety in human anatomy that the general concept of normality ought to be abandoned.

For Amundson, functional determinism is what is meant by biological normality. This may be the concept held by some prominent naturalists about diseases (see Boorse, 1997). Functional determinism is an attempt to give a statistical definition of normal function, referenced to members of species with appropriately similar sex and age characteristics.88 I agree that it is an empirical question whether functional determinism is

88 One could consider functional determinism to be a theory in evolutionary biology concerning the amount of similarity and variation amongst species members or a theory in medical sciences concerning the nature of disease. However, on both accounts it is largely a statistical notion of normal based on lack of variation in biological structures or in the performance of certain biological functions. It is ultimately an empirical question
true for tokens of any biological type. But, as mentioned, I do not believe that there is a uniform notion of normality at work across biology. At the fine grain level, there are many subtle variations in the sense of normal used by biological models. At the course level, there are at least three broad and distinct alternate senses of normality. Regarding theoretical normality, each biological model is able to generate a sense of normal according to how it explains variation and how it determines what is abnormal. Determining theoretical normality is more difficult when scientific models are created in an attempt to take a conventional norm or preference and create a theory or model to explain how the natural world actually produces or endorses the conventional sense of normality and abnormality in question. Whenever it appears as if some conventional norm or preference is being used as the basis for normality in a scientific model, that model should undergo very careful scrutiny.

Amundson attacks the view that diversity is abnormal, as well as the converse, that lack of variation is normal (i.e. functional determinism). He argues that there is no theoretical sense of normal, that empirically a statistical sense of normality is false, and all claims of normality are therefore conventional and have no place in science. Amundson’s main argument may be restated as a denial of any legitimate sense of theoretical normality—any scientific claim about normality is either a statistical claim about variation or is merely an attempt to make certain individual preferences and social practices seem as if they are unbiased and the natural order of things. As stated above, I disagree. Science can handle variation—it is good at telling us how much variation there is in some phenomena, and whether this is pathology or just a set of variants of normal. The real issue regarding controversial claims in biology is whether the models backing such claims of abnormality need rejection, revision, or are relatively sound. It is much harder, however, to evaluate individual cases than it is to reject the entire concept. Careful evaluation takes time. While the model is being verified and tested we are left in a difficult position. The best we can do in such a position is to investigate how the label

whether functional determinism is true for a given species. For such species each would be a ‘textbook example.’ But science clearly has a much less regimented, and less precise, definition of normal function, and this sense is created by and specified by the particular model defining normal function for the system in question. It is relative and specific to the model that creates it.
of abnormality is being used. Such investigation is the proper role of those who wish to point out examples of localized social construction.

3.9 OVERLAPPING SENSES OF NORMALITY AND THE SOCIAL CONSTRUCTION OF DISEASE

Many claims about the ‘social construction’ of diseases can best be understood in light of the different coarse senses of normality. A strong, global charge of social construction in medicine can be understood as the claim that the theoretical sense of normal is ultimately no different than the conventional sense. Proponents of such views believe that there is no theoretical sense of normality worth talking about. Biological models that designate normal states of affair are perpetuating and justifying social conventions and political preferences and as such ought to be evaluated in the same manner as more clearly recognized value judgments and norms (i.e. politically). On this view there is no issue about fit to the phenomena and representational accuracy.

A weaker, local version of the charge of social construction in medicine would be that some cases of what medical authorities try to pass off as theoretical abnormality (i.e. diseases) involves conventional or statistical senses of abnormality. And this weaker charge is clearly true both historically and currently. Some claims of disease involve naïve misunderstandings of the different senses of normality and some are attempts to codify and legitimize discriminatory or other power relations in society. An important issue for any claim about disease is whether the models that give rise to purportedly theoretical senses of normality are accurate representations—do they have good fit with what is being represented. Because this sometimes requires careful study and investigation of the available research or even the undertaking of new experimentation or data production, it is often convenient to get a sense of how respected and virtuous the model seems to be in the eyes of authorities on the subject. This can be a useful shortcut to determining if the model is reliable and worth taking at face value, but it can also lead
to the situation that critics warn against—namely merely accepting the judgment of those in positions of authority.  

Ian Hacking has written on the issue of the social construction of disease, especially mental disease (Hacking, 1999, 1998; 1995). Hacking has tried to frame the issue fairly and has made some useful distinctions. He stresses that the point of social constructionist talk is usually to raise consciousness—not merely to describe how things are, but to change perspectives, and possibly to see that what is thought to be inevitable or unchangeable, is in fact more contingent and open to modification than we expect.

Hacking recognizes that a very potent motivator for constructivist claims is the desire to deflate the authority and power of science. “There is a strong element of unmasking in the work of many constructionists. Their target is not the truth of propositions received in the sciences, but the exalted image of what science is up to, of the authority claimed by scientists for the work that they do… Constructionists want to unmask metaphysics as a bolster for the authority of science. They also want to show that the present state of science was not the only inevitable upshot of dedicated inquiry into the material world that surrounds us. We achieve a robust fit between theories and apparatus, but the fit that we achieve is not the only one we might have arrived at (Hacking, 1999, p. 95).”

For most constructionists disease models are internally coherent, but they represent our own judgments, cultural values and other contingent factors more than representing the fabric of reality. To claim that a disease is a social construction (such as AIDS) or that the very concept of disease is a social construction is to claim that rather than inevitable discoveries about the natural order of things, details about diseases and the concept itself are determined by social factors—factors that would have been very different if our contingent social environment had been different.

---

89 It is always possible that, on the one hand, the incentives and reasons to search for a model are politically motivated and worthy of criticism, while, on the other hand, the actual product has some degree of fit to the phenomena being modeled. The qualities of bias and good fit for a scientific model are not necessarily fully exclusive.

90 Hacking says: “…By constructionism (or social constructionism if we need, on occasion, to emphasize the social) I shall mean various sociological, historical and philosophical projects that aim at displaying or analyzing actual, historically situated, social interactions or causal routes that led to, or were involved in, the coming into being or establishment of some present entity or fact (Hacking, 1999, p. 48).”
Hacking offers a crude sketch of what is generally meant by claims that ‘X is socially constructed’:

(0) In the present state of affairs, X is taken for granted; X appears to be inevitable
(1) X need not have existed, or need not be at all as it is. X, or X as it is at present, is not determined by the nature of things; it is not inevitable.
(2) X is quite bad as it is.
(3) We would be much better off if X were done away with, or at least radically transformed (Hacking, 1999, p. 6).

(0) is a precondition. Inevitability is the main “sticking point” between constructivists and their opponents. Certain scientific classification schemes are assumed to be inevitable, or at least might converge on an inevitable form eventually, given that the natural world has some inherent structure to it. The inevitability of the categorization system for diseases implies that we would slowly, but inevitably, build up a description of normal human biological function, along with many corresponding malfunctions, very much along the lines of the ones we have, because our models are representationally accurate and have been guided by interaction with the phenomena being modeled. The inevitability that we would discover, with sufficient inquiry, that type 1 diabetes is a human pathology, is an example. Constructionists think that if the sociocultural landscape had been different, our model of diabetes may have been importantly different. The constructionist thinks that the details of the model, even its very existence, are significantly based on very contingent aspects of the sociocultural milieu in which it has developed. Hacking points out that even though (2) and (3) are often part of the claim about some X, they need not be.91 “One may realize that something, which seems inevitable in the present state of

91 A gradation for constructionist commitment can be arranged based on how strongly one endorses (2) and (3). The least demanding grade of constructionist, basically just committing to (1), is called historical. Things just could have turned out different had history taken different turns. The ironist, for Hacking, is committed to (1) but finds it ironic that despite clearly realizing that X is not inevitable, people still find it convenient to believe or act as if X were inevitable. Hacking has Richard Rorty in mind as an example of an ironist. Nietzsche might be another example. The unmasker is much like the ironist, but hopes as more people see the lack of inevitability regarding X that it will lose some of its hold on people. The unmasker is often committed to (2). The reformist is always committed to (2) and is even more optimistic that changes can be made regarding X. The constructionist that holds all 3 theses about X is rebellious, while someone who not only believes these theses, but tries to achieve change, is revolutionary.
things, was not inevitable, and yet is not thereby a bad thing. But most people who use
the social construction idea enthusiastically want to criticize, change, or destroy some X
that they dislike in the established order of things (Hacking, 1999, p. 7)."

On my view, the contingency that constructionists see in disease claims emerges
from believing that the sense of abnormality in question is a conventional sense.
Conventional senses of normality are influenced by the social milieu which gives rise to
and sustains their existence. The contingency at issues makes sense once one realizes that
it is conventional senses of abnormality that the advocate has in mind when they are
attempting to unmask the social construction of a putative disease.

Another fruitful distinction Hacking makes concerns what sort of thing is being
labeled as socially constructed. Hacking distinguishes ideas from objects, and both from
what he calls ‘elevator words.’ Trees, rocks, children, genes, quarks, bacteria, etc. are
all objects (if they actually exist). The Second Law of Thermodynamics, the harm
principle, the first amendment, etc, are ideas. Groupings, classification schemes and kind
concepts such as ‘disease’ are ideas, while the actual material entities or events being
categorized are ‘objects.’ Ideas are not free floating, but part of a community and culture.

Ideas do not exist in a vacuum. They inhabit a social setting. Let us call that the
matrix within which an idea, a concept or kind, is formed. The matrix in which
the idea of the woman refugee is formed is a complex of institutions, advocates,
newspaper articles, lawyers, court decisions, immigration proceedings. Not to
mention the material infrastructure, barriers, passports, uniforms, counters at
airports, detention centers, courthouses, holiday camps for refugee
children….This discussion of ideas and classification takes for granted the
obvious, namely that they work only in a matrix. But I do want to emphasize what
in shorthand I call the idea of the woman refugee, that classification, that kind of
person. When we read of the social construction of X, it is very commonly the
idea of X (in its matrix) that is meant (Hacking, 1999, p. 10).

92 Those concepts arising from semantic ascent, such as truths, facts, reality, knowledge,
etc. are elevator words. These are harder to define, but here Hacking has in mind claims
about the social construction of the very ideas of knowledge, facts, reality, etc. His point
is that it is quite a different thing to say that the notion of being a fact is socially
constructed, than to say that an object or even an idea is socially constructed. Such
‘elevator’ words are almost always evaluative.
Ideas are influenced by and in turn influence the matrix they exist within, and the scientific models specifying a disease claims are no exception. It is this dynamic matrix itself that is often what is being claimed to be socially constructed and whose current dynamic form, it is claimed, was not inevitable.

Hacking admits this three-fold distinction is coarse and involves much slippage and blending between the disjuncts. However, it is still very helpful to get clear on what is actually being labeled as a social construct. Hacking demonstrates such utility very nicely with his discussion of child abuse. The phenomena or events (i.e. the material acts) that we might call child abuse are very ‘real,’ and cause pain and suffering. The claim that these individual acts are social constructs is hard to understand and false. But the idea and definition of what counts as child abuse (i.e. how we as early 21st century Americans perceive it), and its place in larger classificatory schemes of behavioral interactions between child and parents, is something that Hacking shows is not inevitable, has not always been the way such events and behaviors were classified or perceived, and may not be the way that non-Americans perceive such behavior, etc. Similarly, there may be very ‘real’ symptoms and disability that a person experiences, but the idea behind this—the explanation of what is going wrong biologically—the scientific model explaining normal and abnormal function—this may be constructed. And not inevitable. Hacking says: “‘Social construct’ and ‘real’ do seem terribly at odds with each other. Part of the tension between the ‘real’ and the ‘constructed’ results from interaction between the two, between, say, child abuse, which is real enough, and the idea of child abuse, which is ‘constructed’ (Hacking, 1999, p. 101).”

Once any classification system is in place, if the things being classified are people, then even more complex and dynamic matrices occur. People often understand they are being classified. For example, consider the concepts of ‘woman refugee,’ ‘homosexual,’ or ‘cancer survivor.’ Such classificatory ideas are called interactive kinds by Hacking, as opposed to indifferent kinds (whose classified objects are unable to react).

We have seen that very commonly, when people talk of the social construction of X, they have in mind several interacting items [some objects and some ideas], all designated by X… [The objects grouped by interactive kinds] can become aware that they are classified as such. They can make tacit or even explicit choices, adapt or adopt ways of living so as to fit or get away from the very classification
that may be applied to them. These very choices, adaptations or adoptions have consequences for the very group, for the kind of people that is involved. The result may be particularly strong interactions. What was known about people of a kind may become false because people of that kind have changed in virtue of what they believe about themselves. I have called this phenomenon the looping effect of human kinds… Looping effects are everywhere… Think about the transformations effected by the notions of fat, overweight, anorexic. If someone talks about the social construction of… anorexia, they are likely talking about the idea, the individual falling under the idea, the interaction between the idea and the people, and the manifold of social practices and institutions that these interactions involve: the matrix, in short (Hacking, 1999, p. 34).

I have not defined “interactive kind,” but only pointed. Kinds that are subject to intense scientific scrutiny are of special interest. There is a constant drive in the social and psychological sciences to emulate the natural sciences, and to produce true natural kinds of people. This is evidently true for basic research on pathologies such as schizophrenia and autism, but is also, at present, equally true for some, but only some, investigators who study homosexuality (the search for the homosexual gene) or violent crime (is that an innate or heritable propensity?). There is a picture of an object to be searched out, the right kind, the kind that is true to nature, a fixed target if only we can get there. But perhaps it is a moving target, just because of the looping effect of human kinds? That is, new knowledge about “the criminal” or “the homosexual” becomes known to the people classified, changes the way that individuals behave, and loops back to force change in the classifications and knowledge about them (Hacking, 1999, p. 104).

For Hacking, the more interesting and sensible claims made in the constructionist vein have to do with claims that some classifying idea is contingent on the particular matrix it is imbedded within. Such categorizing ideas usually cannot be understood or evaluated outside this interactive matrix. When the objects being classified are people, or even the behavior of people, then especially complex interactions and ‘looping effects’ may occur. These complex dynamic interactions of classifying schemes utilizing interactive kinds (instead of indifferent kinds) are one feature that makes social and psychological science appear more contingent.93 Charges of social construction seem

93 Hacking says: “The ‘woman refugee’ (as a kind of classification) can be called an interactive kind because it interacts with things of that kind, namely people, including individual women refugees, who can become aware of how they are classified and modify there behavior accordingly. Quarks in contrast do no form an interactive kind; the idea of a quark does not interact with quarks. Quarks are not aware that they are quarks and are not altered simply by being classified as quarks. There are plenty of
more appropriate for the scientific models in fields that seek to classify types or kinds of people, especially if the boundary in question demarcates normal from abnormal.\footnote{In the end, the “real vs. construction” tension turns out to be relatively minor technical matter. How to devise a plausible semantics for a problematic class of kind terms? Terms for interactive kinds apply to human beings and their behavior. They interact with the people classified by them. They are kind-terms that exhibit the looping effect, that is, that have to be revised because the people classified in a certain way change in response to being classified. On the other hand, some of these interactive kinds may pick out genuine causal properties, biological kinds, which, like all indifferent kinds, are unaffected, as kinds, by what we know about them. The semantics of Kripke and Putnam can be used to give a formal gloss to this phenomenon. Far more decisive than semantics is the dynamics of interactive kinds. The vast bulk of constructionist writing has examined the dynamics of this or that classification and the human beings that are classified by it. Studies of Authorship, Brotherhood, the Child Viewer of Television, and Danger have, in their various ways, been connected with just that: the social construction of the idea of X, of X, of the experience of being X, and so on, and how these interact with each other (Hacking, 1999, p. 123).} There are important differences between attempting to classify the malfunctions and abnormalities of organ systems, tissues, enzymes, genes, etc. versus attempting to classify grossly observable abnormalities in behaviors, personalities, appearances—to classify at the level of the individual person. Amundson, above, ran into this problem because most of his examples dealt with people or their behavior being labeled as abnormal. The conventional and theoretical senses of normal are often complexly entangled at this level of analysis. On the other hand, there are very few conventional theories, norms or folklore about the pancreas and its normal function. A scientific model that explained certain behaviors or kinds of people as diseased would have to compete and interact with other non-scientific notions of normality and abnormality. It may also have to contend with certain ‘looping effects.’

Hacking also explores the dynamics of what he calls transient mental illnesses or “… an illness that appears at a time, in a place, and later fades away. It may spread from place to place and reappear from time to time. It may be selective for social class or gender, preferring poor women or rich men. I do not mean that it comes and goes in this or that patient, but that this type of madness exists only at certain times and places
Hysteria is a well studied instance of just such a transient mental illness, and Hacking examines the less well-known diagnosis of fugue states, as well as dissociative identity disorder. Hacking is interested in issues lurking behind whether such illnesses are ‘real’ or not, but he also is interested in how transient mental illness are possible—i.e. what allows them to flourish at particular times and places, and then eventually disappear. Such transient illnesses only flourish in specific matrices. He uses the metaphor of the ecological niche to stress the idea that a specific set of conditions allows particular speciation to occur. He could have also compared these transient mental illnesses to infectious disease epidemics.

The most important contribution here is the metaphor of an ecological niche within which mental illnesses thrive. Such niches require a number of vectors. I emphasize four. One, inevitably, is medical. The illness should fit into a larger framework of diagnosis, a taxonomy of illness. The most interesting vector is cultural polarity: the illness should be situated between two elements of contemporary culture, one romantic and virtuous, the other vicious and tending to crime. What counts as crime or as virtue is itself a characteristic of the larger society, and the virtues are not fixed for all time… Then we need a vector of observability, that the disorder should be visible as disorder, as suffering, as something to escape. Finally, something more familiar: the illness, despite the pain it produces, should also provide some release that is not available elsewhere in the culture in which it thrives. (Hacking, 1998, p. 2)

The transient mental illness has to be something that, from the current schemes and horizon of possibilities, looks like other recognized diseases. It would have to be what I call a putative disease. Cultural polarity means that the illness has to play off current concerns about human behavior. It has to negotiate a space between conventional senses of normal and abnormal, and between choice and fate. The observability requirement ensures that the condition is one that will stand out to authorities in a position to encourage medical interpretation and intervention. For example, the diagnosis of fugue-states was very observable in Fin-de-siècle France, where army-desertion, immigration scares, and mandatory identity papers for all travelers made a mental illness marked by observable travel stand out. In America during the same period the expanding frontier and the possibility of travel without interference or suspicion made such a ‘disorder’ less visible. Lastly a transient mental illness should provide a release for some desire—
usually a desire not easily attainable by the group particularly at risk for the transient mental illness in question. For fugue states this was the desire to take a ‘vacation’ from day to day life.

On my view, a major component in explaining transient mental illness is to recognize that initial models of pathology may contain much borrowed from conventional notions of what is normal and abnormal for humans and their behavior. However, as the sense of theoretical abnormality (and contrasting normality) is developed and refined, the contribution of conventional senses of abnormality may diminish. Furthermore, as the model is passed from generation to generation, and is developed and refined, the conventional senses that originally aided in the birth of the model may be abandoned by society. This is true of the case of fugue state. The ecological niche that allowed the diagnosis to be created ceased to be, but the scientific model of a behavioral and cognitive abnormality has survived, even if it is now diagnosed with less frequency. It is not surprising that such ‘transient’ disorders arise in psychiatry or those fields dealing with gross appearance or behavior and the associated conventional judgments of abnormality.

Hacking specifies three areas of real disagreement between constructionists and their pro-science critics. He calls these ‘sticking points,’ and in particular they arise in discussions of the extent to which the indifferent kinds specified in scientific models in the natural sciences are socially constructed.

A social construction thesis for the natural sciences would hold that, in a thoroughly nontrivial sense, a successful science did not have to develop in the way that it did, but could have had different successes evolving in other ways that do not converge on the route that was in fact taken. Neither a priori set of bench marks nor the world itself determines what will be the next set of bench marks in high-energy physics or any other field of inquiry. …If contingency is the first sticking point, the second one is more metaphysical. Constructionists tend to maintain that classifications are not determined by how the world is, but are convenient ways in which to represent it. They maintain that the world does not come quietly wrapped up in facts. Facts are the consequences of ways in which we represent the world. The constructionist vision here is splendidly old-fashioned. It is a species of nominalism. It is countered by a strong sense that the world has an inherent structure that we discover. The third sticking point is the question of stability. Contrary to the themes of Karl Popper and Thomas Kuhn, namely refutation and revolution, a great deal of modern science is stable. Maxwell’s Equations, the Second Law of Thermodynamics, the velocity of light,
the lowly substances such as dolomite are here to stay. Scientists think that the stability is the consequence of compelling evidence. Constructionists think that stability results from factors external to the overt content of the science. This makes for the third sticking point, internal versus external explanations for stability (Hacking, 1999, p. 33, emphasis added).

For the biological model of the thyroid to be a social construction its central features would be significantly influenced by cultural and social factors and could easily have had different content than they do, because the social and cultural landscape—the matrix of ideas in which they came to life—could have been different. One could also believe that the trajectory of what questions are asked—what sort of research gets done, what gets rewarded, what overlooked or ridiculed—is contingent. Such contingency of form or trajectory is a real issue, as outlined in chapter 2. However, it is possible, I believe, to assess representational accuracy and fit. Radical contingency, however, holds that there are no external constraints that inevitably lead some models, like that of the thyroid, to converge on increasingly more accurate representations. With enough time and with the accumulation of enough robust fit (and probably also some overlap in the interests and concerns that drive research and experiment) there would be (and has been) convergence in the content of specific scientific research programs. This is the sense in which the constructionist denies inevitability. Such radical contingency is especially understandable if one’s position on the second sticking point is that there is no inherent structure in natural phenomena to be discovered (or that any such structure is beyond our ability to describe).

The third sticking point concerns the stability of certain scientific models and categorizations. Some models require only occasional refinement. They have resisted refutation. Certain disciplines appear to have a collection of such stable models built up over several generations and spanning distinct historical periods and cultures. Anatomy and physiology are examples. We could, as I did in chapter 2, call the collections of such stable models the ‘core’ of an established disciplines—the sort of basic and fundamental findings that might organize a comprehensive textbook for the field in question. These core findings are stable because of a preponderance of evidence and from the mutual support of many researchers, and from their interrelations with other core models and findings both within and without their home discipline. These explanations of stability
are internal to the content of the natural sciences and the support and stability ultimately come from experimental data and the scientific method—from the process selecting those models with the best fit. Constructionists, on the other hand, do not think this stability comes from such sources internal to the methodology of the natural sciences. “The constructionist holds that explanations for the stability of scientific belief involve, at least in part, elements that are external to the professed content of the science. These elements typically include social factors, interests, networks, or however they be described (Hacking, 1999, p. 92).”

The idea of our scientific theories arriving at much different content even after sustained periods of investigation and revision is unlikely. In biology and psychology, debates about contingency and stability are better understood when one keeps in mind the difference between conventional and theoretical/descriptive sense of abnormality and normality. Scientific models are the constructions of human investigators, but to admit this does not commit one to their being necessarily contingent, nor even to their being determined more by the social and cultural landscape than by the salient detectable features of the phenomena being modeled. For any individual model this is an empirical question, and the ‘fit’ that the model has with the phenomena, as well as the possibility of bias or error, must be assessed. If the model is from a very reputable field that has an established methodology, a track record of accurate and useful models, and if the model under assessment has existed essentially in its current form for several generations, surviving criticism and challenge, and refining itself as new data and other related models have been developed, then, in that case, one might feel comfortable trusting an assessment that the model was a good representation and constituted legitimate empirical knowledge. The current models we have about the function and form of the pancreas and the thyroid glands, and diseases that result from their dysfunction, are examples, and it is very difficult to see how the content of these models could have been different had the social context or values of the community been different. However, there are other disease claims, backed by less well-established scientific models, for which the possibility of contingency is more concerning. For diseases such as ADHD or gulf war syndrome the explanations of the biological abnormality (as well as models of the normal function of the biological system effected) are not yet available. Here there is a grey area.
Should one trust that these are legitimate disease claims and that the models will not be revised or rejected? In such situations social construction in the pejorative sense can occur. What seems inevitable may turn out to be much more contingent once more information is available or even as we become less interested in some questions relative to others. During such periods of exploration and development the matrix within which models are constructed may lead to contingent content.

I believe that the natural sciences often produce models and categorize types of things in a way that avoids any problematic sense of being a contingent social construction. In the natural sciences, even those dismissive of social construction talk will grant that there is a history and a process behind the development of scientific models and theories. Such a process is social and is influenced by cultural and historical factors—factors external to the presumed content of the theory in question. The defenders of the accuracy and stability of some scientific models should clearly distinguish between the process of discovery and the time-tested products of ongoing inquiry.

In summary, I believe that claims of social construction regarding diseases can be understood in terms of the differing senses of normality. The main argument of my thesis is that to be a disease is to be a theoretical abnormality in the function or form of a biological system. Legitimate disease claims occur when the theoretical sense of abnormality is specified by a reliable, objective and uncontroversial scientific model. The best understanding of the claim that a disease is merely a social construction is that the sense of abnormality being conveyed is to a significant extent a conventional sense of abnormality. This explains the origin of the contingency or transience of such a socially constructed abnormality. Conventional senses of abnormality are relative to personal and social norms or value judgments. As mentioned, the most interesting cases are often those disease claims that contain mixtures of conventional and theoretical senses of biological or psychological abnormality. Such mixtures tend to occur frequently as one moves towards levels of analysis that take account of gross anatomical form and function, human behavior and cognition, and even overall assessments of individuals.

The looping effects that Hacking discuses are interesting to consider in light of the possible mixtures of theoretical and conventional senses of abnormality. Take the example of schizophrenia. The theoretical concept of abnormality at the heart of the
claim that it is a disease concerns details about various dopaminergic neural circuits in
the brain and about the neurotransmitters, receptors, and enzymes that control the levels
and activity of these circuits (and ultimately genes and environmental factors that
collaborate to create the potential for abnormality). However, the actual behavior that
serves as the symptoms of the disease, and the overall perception and experience of the
individuals who suffer from this abnormality in brain chemistry are subject to
conventional judgments of being abnormal. And these judgments of abnormality are
influenced in complex ways by the social matrix in which they exist. One need only think
of the different ways in which suffers of this disease (i.e. those exhibiting the typical
behaviors) have been judged in different social or historical contexts. In certain historical
contexts these individuals might have been taken to be divinely blessed oracles or
vehicles by which the gods communicated with the community. They may be considered
great artists of unique creativity. They might be considered possessed by demons,
impure, criminals, etc. These people may also be perceived as suffering from a disease
beyond their control and deserving of special care and accommodation. The view that a
community holds about these individuals might be internalized, rebelled against, or
embraced by the individuals so labeled or judged. The individuals in question can
understand what the community finds unusual or abnormal about them and this may
possibly change their behavior in light of this realization. However, across all
circumstances and regardless of what any particular community might hold, regardless of
anything the individual may decide, if and when there is a reliable and accurate
theoretical model of neural malfunction, the biological details being specified will not be
open to change or alteration due to the interplay of perceptions between suffer and
community. No matter how the community perceives the behavior and symptoms of
those with type 1 diabetes or congenital hypothyroidism (i.e. cretinism), the details of
what is abnormal about the function of the pancreas or thyroid is the same across
communities and individuals affected.

Consider the example of disability, as discussed in many of Amundson’s
examples. The reaction of the individual and the interplay of how the individual and
community deal with the behavior in question are contingent and malleable—the person
being judged abnormal may change their behavior in light of this judgment and the
community can change its interpretation and treatment of individuals who exhibit the behavior or the gross anatomy in question. Regardless of how the community chooses to treat or judge the disabled, the reasons for some of their condition, if specified by a reliable model are not contingencies. Where the appropriate scientific models exist, these are pathologies and the results of abnormal human biological form and function. The biology is not contingent even if the way we choose to deal with it and perceive it, as individuals or a community, is open to many possible perceptions. The looping effect is at work with conventional sense of abnormality, but not theoretical senses. To fully address discrimination, disability, or human suffering we must understand what the causes are, what can be changed, and how best we can effect changes. To adjust an impractically, inaccurate or unjust label of abnormality, we need to understand what sustains the claim and what forces, if altered, would reveal the problem and lead to a solution.

It is important to keep track of what sort of thing is being called a social construction. Ideas are human constructions, and thus scientific models are constructions—usually constructions contributed to by groups of researchers. Successful models are the result of a very social practice of sharing, challenging and revising the details. But what makes these models representations and not merely satisfying stories is the possibility of interacting with the actual phenomena—experiments can be reproduced and models challenged with new experiments, new apparatus, new results from research into related phenomena, etc. The actual phenomenon is not contingent or relative to the communities values, even if our initial models of that phenomenon, at any one moment in time, may be open to bias or influenced by contingent factors.

The global charge that all disease claims are social constructions is unconvincing. I see no interesting or meaningful sense in which the models of normal and abnormal function for the thyroid gland, the pancreas, or the heart are reflections of personal or group values or norms, nor are they intended to maintain certain social arrangements or discriminate. The process of discovery and the history of how and why certain models were proposed may be contingent, and originally models may contain bias and value judgments. However, as models are criticized, refined and increasingly improved to have better and better fit with the phenomena they describe, the factors lending to contingency
have less influence and the objectivity and representational accuracy of these models improves (at least in the successful cases). The stability and longevity of certain models in their basic form is evidence of their good fit with the phenomena they model.

Regarding the view of extreme nominalists about scientific representation, I would answer along lines similar to those of Giere as described in the last chapter. Realism is a methodological assumption that has worked out very well so far, and for which there currently is no reason to doubt this assumption. Whereas Giere had physical sciences in mind, I am focused more on the biological and psychological sciences. In the next chapter I will give my explication of disease and discuss some interesting and challenging cases.
CHAPTER 4
DISEASE

4.1 INTRODUCTION: THE PROBLEM OF DISEASE

Consider an emergency room doctor talking to a 52 year-old woman. He’s taken
the history of her complaints, discovered her reasons for coming to the hospital in the
middle of the night, noted her past history and what medications she is taking, and he’s
examined her. Some blood work has been done along with some bedside monitoring of
the electrical activity of her heart. The physician tells the patient that the shortness of
breath, feeling of pressure on her chest, and sweating that awoke her from sleep a few
hours ago, were due to a heart attack. Specifically she has had a myocardial infarction,
most likely secondary to a suddenly occluded coronary artery. Sufficient blood is not
going to the muscle that makes up the walls of the heart, and there is evidence from the
blood work and electrocardiogram (EKG) that some of that muscle may have died. The
evidence is strong enough to risk the recommended interventions. The patient is given
certain medications to prevent progression of the physiological processes involved in
occlusion and to reduce the hearts demand for blood. The evidence justifies risking a
procedure to find and unclog the occlusion. A catheter shall be inserted into her femoral
artery and passed up to her heart and into her coronary arteries. The procedure will search
for the suspected occlusion and, if found, reopen the arteries, possibly placing a metal
stent to help keep the occlusion from reoccurring.

From the physician’s perspective, the patient’s claim that she is suffering from a
disease is uncontroversial. The physician listens to her complaint, examines her and
collects some initial data via blood work and an EKG. He knows that many different
things can cause chest pain, waking at night short of breath, and sweating. He knows that
discovering the cause of the problems is crucial for providing the correct intervention.
He’s kept the possible causes in mind, ordering them regarding likelihood as more
evidence is collected. Based on that evidence, he believes clogged coronary arteries are
the most likely explanation. This is an acute coronary syndrome, and based on the EKG,
an ST elevation myocardial infarction (STEMI or MI). The percutaneous coronary
intervention (PCI) planned will definitively prove the diagnosis if there are occluded
arteries, and it allows several kinds of intervention during the same procedure. The emergency room doctor will have to make the case that a certain disease process is causing the symptoms to the cardiologist on call (who will perform such a procedure). The physician responsible for the primary care of the patient will eventually also want to know about the details of the diagnosis, because certain long term medications, lifestyle changes, etc. will be warranted to slow any further disease progression and lower the risk of a second heart attack.

The insurance company that is asked to pay for the emergency room visit, the PCI, and the hospital stay will be interested in whether this is a legitimate disease claim. Was the specific disease diagnosed the right one, and did it justify the procedures and care that ensued? If it had been merely a strained chest wall muscle causing chest pain and difficulty breathing, less costly intervention would have been justified. Furthermore, had the diagnosis been wrong and the patient’s symptoms had been caused by a dissection of the ascending aorta or a massive ‘saddle’ pulmonary embolism, death could have occurred while the wrong diagnosis and interventions were pursued. In those cases hospital administrators and lawyers would have been interested in the claims about what particular disease was at work and why it was or was not diagnosed correctly.

The patient perceives the situation a certain way based on the specific diagnosis and her previous understanding of such situations. The diagnosis will justify certain entities to pay for the intervention. The patient will be allowed to miss work without being disciplined for her failure to show up. For the next few months her entire life will be re-evaluated from the perspective of being a person who has suffered a heart attack, and whose life is threatened by the physiological processes that caused the current episode and potentially future ones.

The patient’s current condition is the result of one or more diseases. One of the central questions in the philosophy of medicine is specifying the nature of the concept ‘disease.’ The patient in the scenario above is suffering a heart attack. A myocardial infarction (MI) is a pathology and the clogged arteries causing it were produced by microscopic changes occurring in the structure and function of cells making up the arterial walls and occurred over many decades. Contributing to these physiological processes (let’s assume) were the further diseases of atherosclerosis, hypertension, and
diabetes. These were risk factors for the MI. But not all claims of disease are so uncontroversial. Is chronic fatigue syndrome a disease? What about alcoholism? The child who’s arm is broken by child abuse is experiencing pathology, but is the parent who did it also suffering from a disease? Drowning may be an accident, but is it a disease? Is the child who cannot sit still in class just ‘being a typical child’ or is he suffering from the disease of ADHD, such that without the proper medical interventions, his entire academic future may be in jeopardy? If there were a dispute about some claim of disease, how would it be settled? How would a physician, lawyer, or policy maker decide if a disease is causing the problem in question? How does one decide between legitimate and illegitimate disease claims?

Depending on what sort of scholarly project one was engaged in or what sort of perspective one took, disease might be conceived of differently. The term might be seen as justifying or helping to coordinate certain practices and social roles in the community (Parsons, 1971). A sociological project might conceive of disease as classifying certain kinds of people and behaviors. A pragmatic perspective could be taken, seeing disease as whatever doctors diagnose and treat. A more theoretical perspective might also be taken, seeking an answer to the question of whether disease is a natural kind or not (see Reznek, 1995 and D’Amico, 1995). Perhaps it distinguishes importantly different aspects of biological phenomena. Concerns about the scope of the term disease might be addressed. What relationship does ‘disease’ have to other terms, like ‘illness,’ ‘pathology,’ ‘injury,’ ‘affliction,’ ‘wound,’ ‘malady,’ ‘malformation,’ ‘defect,’ ‘syndrome,’ or ‘disorder.’ One might see these as a family of related terms. Some terms might be narrower (like wound) and some broader (like pathology). Some focused on acute issues (i.e. wound) and some chronic (i.e. malformation). Some examples might have ‘internal’ loci of causation (i.e. like genetic defects) unlike those that imply attacks from without (like infection or injuries) and might be the fault of another person or a dangerous environment. Some of these terms might highlight systemic malfunctions and imbalances (i.e. disorder) while others highlight more localized problems (i.e. injury). Some of the terms may refer to static states or structures (i.e. deformity or handicap) while others are dynamic processes (i.e. illness or disease). Some terms might emphasize causes (infections) while others emphasized consequences (wound). Our intuitions, based on linguistic usage, might be
that the term ‘disease’ overlaps with some of these and less with others.\textsuperscript{95} We might claim that getting hit by lightning causes a wound, and is a trauma and injury, but not an illness or a disease. Drowning may be a trauma and cause injury, but it does not cause a wound, and is not an illness or a disease. Being born with six fingers or as a conjoined twin may be a malformation and defect, but not a disease nor trauma. Hypertension may be an illness, affliction, and even a defect, but not an injury or syndrome. Amyloidosis may naturally be called a disease and a syndrome, but not an injury, like a broken leg. Being poisoned may be a misfortune and malady that causes pathology, and seeking medical help may be appropriate, but is it also appropriate to call it a disease? And what about malnutrition or vitamin deficiencies? Perhaps the only commonality between these examples is that they are all inconsistent with being completely healthy.

Different examples of diseases may motivate different conceptions. Examples and explanations from different historical periods may also motivate very different ideas about disease. The original explanatory model for Down’s syndrome is much different than the model medical science now offers. In the past, extreme depression and sloth (i.e. psychomotor retardation) had theoretical models very different than those currently offered by science. In the future our current models may look very incomplete and naïve. Different examples from different branches of medicine or biology may motivate different conceptions of what we mean when we say someone has a disease. The disorders of psychiatry may seem different in kind than those of endocrinology, infectious disease, or trauma surgery. Kinesiology and functional anatomy may have very different examples of malfunction than other fields. Diseases caused by single gene defects may seem very different in character than disorders with complex multifactorial causation (including environmental components) such as diabetes, hypertension, and atherosclerosis. The diseases affecting only newborns and premature babies might elicit intuitions different from those based on diseases of advanced age. Regarding branches of biology, medical physiology may give a perspective on what is normal and abnormal

\textsuperscript{95} A natural language analysis could be offered, although that is not my project or interest. My intention is to clarify what physicians mean by the term disease. My intention is to clarify the scientific term that is used as part of the practice of medicine and biomedical research, that is disease as pathology.
different from evolutionary or developmental biology. Disease examples in veterinary medicine or plant biology might elicit different conceptions than human medicine.

Many theories have been offered in the philosophical literature (see Simon, 2007 and Schwartz, 2007). Basically, on my reading, most authors can be pushed into one of two broad camps, namely the normativist or the naturalist position. As I was illustrating, differing perspectives and projects can motivate clarification of what constitutes being a disease. Normativists generally are more motivated by activism and ethical concerns. They are focused on aspects of the consequences of disease instead of the causes of disease (see Engelhardt, 1996; Reznek, 1987; King, 1954; Wakefield, 1992; Nordenfelt, 1987, 2006). They tend to see disease as more of a welfare or value concept than a descriptive or theoretical concept. A strong normativism basically holds that all senses of normality and abnormality are conventional, and thus involve value judgments. Realizing this allows us to better solve issues concerning the legitimacy of disease claims, because fairly applying a normative concept like disease will require some aspect of negotiation within a community and consensus building, as well as rational argument. Thinking that empirical research can settle claims about whether particular kinds of problems, such as those suffered in chronic fatigue syndrome, are legitimately called disease, would be mistaken according to the normativists. Research might tell us how we could ameliorate the problem with technology, but not whether we should see it as something best treated with medical intervention.

Naturalists tend to focus more on causes of disease and less on the social and personal consequences of being diagnosed (Boorse, 1997). The concept is a theoretical and technical term more than an evaluative one. The naturalist position is that disease is a descriptive concept. Naturalist tend to be motivated more by epistemological concerns than ethical projects or activism. For the naturalist, empirical research is the primary means for settling issues concerning the legitimacy of disease claims. Naturalism is the modern default position of most physicians. The normativist position, however, is the dominant position in the academic literature on disease. It would come as a shock to most physicians and patients that what distinguishes legitimate disease claims from non-legitimate is a complex process of social evaluation. Many academics would reply that discovering diseases are essentially social construction, although shocking, is what results
form careful reflection. Furthermore this insight is valuable for those having to deal with political or ethical issues involving disease claims.

What would be the consequences of ignoring questions regarding the nature of disease claims? Medicine seems to operate quite well while ignoring any ambiguity or indeterminacy in the concept. First, intellectual interest and clarity of thought are worthy goals on their own, and certainly enough to motivate a philosophy dissertation. After all, disease is a concept central to medicine and much of biological research. But beyond intellectual curiosity and rigor, should anyone care about the debate between naturalist and normativists? Only in the past few decades has the answer been clearly yes. This is due primarily to two factors that are driving demand for definitive answers to practical questions of legitimacy concerning specific claims about certain problems being diseases. One factor is the increasing knowledge and success of biomedical science. Humans now understand how many biological systems work and we can increasingly exert some control on some of these systems. The second factor is the impact that employing more and more biotechnology has on the economy, especially as the technology becomes increasingly expensive. There is a finite amount of resources that the community can spend on medical intervention. And because some of these resources are more scarce than others, issues of allocation arise. Increasing ability to intervene, but at increasing cost, produces demand for answers to the question “Is this problem one that ought to be treated with medical intervention?” Most people find the answer to that question involves asking if the problem is a disease or caused by a disease. The boundary between legitimate and illegitimate disease claims, it is hoped, provides an answer to where the boundary lies between what conditions should be treated medically and which should not.

How can the boundary between legitimate claims of disease compared to conditions and problems falsely being claimed to be human diseases be discovered? Normativists and naturalist offer different answers on how we must draw this line. Furthermore, normativists and naturalists see the relationship between legitimate and illegitimate disease claims and other boundary issues differently. As mentioned, one of

---

96 I touched on much of this in chapter 3 when I discussed the phenomena of medicalization.
these related boundary issues concern which sorts of human problems should be
addressed with medical intervention and biotechnology and which not. If a certain
problem is a disease, treating it is therapy and this could be distinguished from non-
therapeutic uses of biomedical intervention, whether this is preventative intervention, or
intervention for means of enhancement, self creation, self expression, social
standardization, etc. The boundary between what interventions and technologies will be
paid for as a matter of one’s health insurance versus those that should not be covered by
such pooled risk management mechanisms is closely related to the therapeutic versus
non-therapeutic divide. As mentioned many people are hopeful that legitimate diseases
can serve as a proper target for what to address with medical intervention and what ought
to be covered by insurance. For example, Norman Daniels (Buchanan, et. al. 2000;
Daniels, 1985) has written on the topic of how much health care we ought to provide
each other as a matter of equalizing opportunity. There is a level of health and
functionality to which everyone ought to be maintained or restored. He argues we owe
each other this as a matter of social justice, and sees some basic level of health care
provision as following from the theory of distributive justice offered by Rawls. Daniels
case rests on having a somewhat objective boundary between normal and abnormal
biological function, and this could potentially be determined by a naturalistic conception
of disease. The nature of disease might influence issues concerning the similarity of
psychiatry to the rest of medicine and about the similarity and differences between
mental illness and physiological diseases. There are interesting questions about the
relation between the art of medicine and the science of biology which will be influenced
by how one answers the questions about what counts as disease. Is medicine applied
biology the way mechanical engineering might be considered applied physics? Is
medicine fundamentally different due to its uses of pathos and ethos, as well as logos.
Similarly, the line between what physicians should treat and not treat, or what role
physicians should play in society, is related to the legitimacy boundary. Being diagnosed
with a disease is frequently interpreted as being labeled abnormal in some way. This
creates boundaries between the healthy and ill, and this is particularly important when
diseases carry with them a social stigma, such as HIV. People will often assume that they
can be or should be treated differently. Being clear on exactly what kind of abnormality
or misfortune is being claimed by having a disease is important (as are the specific
details). This clarity might prevent or mitigate certain kinds of prejudice and
discrimination.

The naturalist hopes that appeal to a descriptive, scientifically determined
legitimate boundary will help in determining how to draw boundaries in these other
controversies. The normativist, on the other hand, generally sees this as putting the cart
before the horse. The normativist stresses that it is the negotiation and pull of these
difficult social issues and boundaries that determine where the lines between legitimate
disease and illegitimate will be drawn.

The concept of disease is of interest to me because of the boundary issues,
especially the issue about whether or not the concept of disease can be used to draw a line
between using biomedical technology for therapy (treating disease) versus its use for
extra therapeutic reasons (i.e. enhancement). I am interested in arguments for and against
using biotechnology for enhancement. I am interested in how one discerns an illegitimate
disease claim. I am also interested in the relationship between science and medicine, and
in the nature of scientific knowledge generally. I am interested in the nature of psychiatry
and its relation to the rest of medicine. I am planning on practicing medicine as a career
and also hope to contribute to philosophical discussions involving medicine. I want to
continue clarifying my own concept of disease as I use it in medical practice, maintaining
a reflective equilibrium between theory and practice.

My perspective and project may bias my theory of what constitutes being a
disease. One of the fundamental divergences between normativism and naturalism is the
attitude taken towards science. I do have a positive attitude towards science, and believe
it is capable of producing useful knowledge. I believe that science can and sometimes
does offer descriptions of the world that are accurate and unbiased. Most of the
knowledge at the core of most of the fields involved in the biological sciences is as
objective and factual as human knowledge can be. In this regard, my position could be
conceived of as naturalist. Alternatively, my acknowledgement of the perspectival nature
of knowledge and my admission that scientific models are (in a non-pejorative sense)
social constructions, is more in line with normativist views. Chapter 2 offers a sketch my
position on science. My project is theoretical and not ethical. Disease is a theoretical and
descriptive term, not an evaluative one on my account. I believe that biologists and physicians have a coherent and wide concept of disease as pathology. This concept is essentially the same across different fields of biology, from plant and animal biology to human medicine. Historically, the notion of disease as biological abnormality has been among a cluster of notions determining what we designate as disease. Different aspects of these core notions may have been emphasized or marginalized across different historical and cultural contexts.\(^97\) As biological science has advanced, however, the causal notion of biological abnormality has displaced other notions at the core, especially those dealing with the experience and consequences of disease. Such displaced notions would include disability, suffering, pain, death, etc. As biomedical science has progressed, the concept of disease has become increasingly a descriptive and naturalistic one. The core of the modern concept of disease is *biological abnormality*—the malfunction or malformation of a biological system that could have operated normally otherwise. It would probably be appropriate to call what I am clarifying the concept of *pathology*. I believe that the narrower definitions of disease, wound, injury, trauma, infection, poising, etc. all have as part of their core meaning a departure from normal biological form or function. Therefore I feel it is appropriate to call the majority of those conditions discussed above (broken legs, burns, malnutrition, having six fingers, being a conjoined twin, suffering from drowning or poisoning, etc.) ‘diseases’ and ‘pathologies.’ They are pathologies just as much as having diabetes, cancer or cholera. The woman suffering a heart attack in the above example is slotted into an entirely new set of social networks and interactions, and has new rights and responsibilities that come with her new status as a sufferer of this particular kind of disease. Society often accepts these changes with astonishingly little skepticism or need for verification.\(^98\) This power to alter one’s place in the social network

\(^97\) The theoretical framework in which normal and abnormal function is understood may differ, and the technology to examine and manipulate things at levels of analysis lower than the grossly observable, are important differences across time and culture. However, as a core concept of disease, biological abnormality, is identifiable across cultures and historical periods. This is why we can recognize medicine as a profession in other times and places, and why we can to some extent evaluate the disease claims of these communities.

\(^98\) Part of the reason for this is the trust most people place in physicians. Part of the willingness to place such trust in physicians is a belief that diseases are important and
makes the term ‘disease’ and its pattern of application a natural target for sociological study. Although a valid project, it is not my project here to examine the notions of illness as understood by the general community, by patients, by the disabled, etc. I am not concerned with the concept as it is used to influence social practices and used to categorize people and acts in socially significant ways.

In this chapter I will review the naturalist and normativist positions. I do this by discussing the concepts of disease offered by Boorse and Engelhardt. I find these the most sophisticated and most frequently cited statements of the two opposing positions on the nature of disease. I shall highlight where I agree and disagree with their positions. I shall then give my own criteria for what makes a disease claim legitimate and distinguish this from illegitimate and putative claims. Finally, I review some of the resources my position provides in analyzing particular cases, and then discuss and analyze a number of interesting and controversial disease claims.

4.2 NATURALISM: BOORSE’S BIOSTATISTICAL THEORY

Naturalists tend to have a positive view of science and its products, tend to believe disease is a descriptive not evaluative concept, and tend to emphasize the importance of causal explanation instead of consequences. The fundamental problem facing naturalists is explaining how a non-evaluative and descriptive term can also express a sense of abnormality. How can abnormality be merely observed and not be the judgment of a human observer—a judgment necessarily made in reference to, and against a background of, personal and social values? To deal with this problem naturalists frequently appeal to statistics and evolution. Evolution offers the hope of explaining how it is that naturally occurring phenomena can appear as-if-designed without actually having been designed. As-if-designed biological creatures can then have something equivalent to the concept of normal operation and function that we apply to artifacts or machines. These authors usually equate (in some way) the natural and the normal. Statistics offers the hope of determining such normal function or operation without ‘real,’ and that physicians can distinguish legitimate claims from illegitimate. If people felt that the concept of disease was indeterminate or merely a social construction in some way, physicians would not retain the authority that they do.
appealing directly to value judgments or intentions of a designer. Christopher Boorse’s
theory of disease is the best argued version of the modern naturalist position.

Boorse wrote a series of articles in the 1970s developing his biostatistical theory. He argued for ‘objective’ notions of health and disease (Boorse, 1975, 1977, 1987, 1997). “It is a traditional axiom of medicine that health is the absence of disease. What is a disease? Anything that is inconsistent with health. If the axiom has any content, a better answer can be given. The most fundamental problem in the philosophy of medicine is, I think, to break the circle with a substantive analysis of either health or disease (Boorse, 1977, p. 542).” For Boorse health is the ‘normal’ operation of the body and disease is a departure from this normal operating state. These are not unusual understandings of health and disease, but Boorse thinks that they get primarily descriptive and therefore objective conceptual elaboration by the biological sciences, and physiology in particular. He says, “I suggest that the distinction between normal and pathological conditions is the basic theoretical concept of Western medicine. A bodily state or process is disease, disorder, injury, lesion, defect, sickness, or illness only if it is abnormal in the sense of pathological; in other words, these are all specific kinds of pathological conditions. (Boorse, 1987, p. 365)”

Disease is abnormal biological function. Health is normal biological function. Normal function is determined by looking at the dynamic operation of naturally occurring organisms. The biological sciences have approached and modeled the operation of parts of living systems in a manner analogous to analyzing the function of parts of a machine.

Theoretical health now turns out to be strictly analogous to the mechanical condition of an artifact. Despite appearances, ‘perfect mechanical condition’ in, say, a 1965 Volkswagen is a descriptive notion. Such an artifact is in perfect mechanical condition when it conforms in all respects to the designer’s detailed specifications. Normative interests play a crucial role, of course, in the initial choice of the design. But what the Volkswagen design actually is, is an empirical matter by the time production begins. Thence-forward a car may be in perfect condition regardless of whether the design is good or bad. If one replaces the stock carburetor with a high performance part, one may well produce a better car, but one does not produce a Volkswagen in better mechanical condition. Similarly, an automatic camera may function perfectly and take wretched pictures; guided missiles and instruments of torture in perfect mechanical condition may serve execrable ends. Perfect working order is a matter not of worth of the product but of conformity of the process to a fixed design. In the case of organisms, of course,
the ideal of health must be determined by empirical analysis of the species rather than by the intentions of a designer. But otherwise the parallel seems exact. A person who by mutation acquires a sixth sense, or an ability to regenerate severed limbs, is not thereby healthier than we are. Sixth senses and limb regeneration are not part of the human design which at any given time, for better or worse, just is what it is (Boorse, 1975, p. 83).

A strength of Boorse’s position is that it recognizes that modern biology has made impressive strides in explaining the normal operation of circumscribed biological systems. Something like natural design is approximated by evolution. The design is discovered by empirical investigation of the biological systems in question.

Boorse dismisses other candidates for the core notion of disease. First he dismisses the idea that diseases are basically undesirable biological conditions. “It is undesirable to be mildly below average in any valuable physical quality, e.g. height, strength, endurance, coordination, reflex speed, beauty, etc. It is undesirable to have such universal human weaknesses as a need for sleep or for regular access to food and water. These conditions are not diseases. Yet one could never distinguish them from diseases on grounds of disvalue alone (Boorse, 1977, p. 544).” The idea that disease is whatever physicians treat is also dismissed. Physicians intervene to control biological functions when no disease is present (e.g. reduction of fertility via hormonal birth control, circumcision, or purely cosmetic rhinoplasty). Physicians also consider many conditions to be diseases for which they have little or no ability to intervene (e.g. Charcot-Marie-Tooth disease).

Pain and suffering, for Boorse, are also not the central concepts involved with being a disease. He cites the common occurrence of diagnoses being made before any painful or disfiguring sign or symptoms of the problem have developed (e.g. tuberculosis detected on routine PPD testing in workplace or school physical or early detection of HIV infection). Conversely, many conditions are painful and cause suffering, but are not diseases or disease processes. Childbirth is one example. Boorse also dismisses the possibility that the central concept of disease involves disability. Again, many diseases

---

99 Many of the examples in this paragraph and the next are mine and not Boorse’s. Charcot-Marie-Tooth disease is a neurodegenerative disease that does not have a therapy at the moment.
are not disabling (e.g. ringworm infection or minor lacerations), while certain disabling conditions (e.g. pregnancy, certain cases of obesity, or normal deteriorations caused by aging) are not diseases. Like the other candidate concepts, disability picks out a class of conditions and problems different from those picked out by the concept disease.

Adaptation, or rather maladaptation, to one’s environment is also not the core notion that explains what conditions are legitimate diseases. Certain diseases might be very adaptive to certain environments—some genetic mutations might prevent us from acquiring HIV, or might help us avoid cholera or malaria. Depending on the environment, normal biological function or structure might be poor adaptations. Humans are not well designed to live on other planets in our solar system, but finding difficulty in doing so is not to acquire a set of diseases. Another possible candidate for the core concept of disease is homeostasis. Boorse points out that many physiological systems attempt to keep certain parameters within tight control, and have mechanisms to return these parameters to equilibrium if perturbed. However, many chronic disease processes are situations where, at least temporarily, new equilibria are achieved (e.g. congestive heart failure or chronic obstructive pulmonary disease). Also many biological functions, like movement and reproduction, have little to do with homeostasis. Boorse also considers simple statistical rarity in form or function as the means to distinguish disease from health, but quickly dismisses this for obvious problems. I shall discuss this dismissal below when I discuss how Boorse tries to modify and thus retain a statistical account of normality as typicality for his theory. The success of his biostatistical account of disease relies on making such a modification.

For Boorse, the core idea behind the medical concept of health is natural or normal biological function and behind disease is abnormal biological function. “…[O]ur

---

100 This raises an interesting question whether certain conditions such as caisson’s disease, acute mountain sickness, or even drowning, are really diseases, as these are similar to the situation of attempting to operate in environments not well suited for normal biological functioning. The crucial issue on my theory is how the models of normal biological function account for normal vs. abnormal environments. Models of normal function usually assume ideal external conditions. I discuss these cases in more detail below.

101 Again, the examples here are my own and are meant to clarify and strengthen Boorse’s arguments.
Evolution by natural selection has produced organisms with typical functions. These functions taken in sum can be seen as species typical design. This design can be examined and analyzed by the same reverse engineering techniques we might use to figure out how a never-before-seen foreign-made technology worked. With this picture in mind, Boorse tackles the central problem for a naturalistic theory of disease: How can normal function be determined without direct appeal to the preferences or value judgments of those creating or using the biological models? In answering the question Boorse must address two issues. He must address the issues concerning how it is that one knows which function among the many observable functions of a biological system is the proper, correct or normal function. This is the classic question about how we know that the proper function of the heart is to pump blood and not to produce certain noises, electrical discharges, or peptide hormones. The second issue concerns distinguishing, without appeal to values or preferences, normal variations from the significant variations—variations significant enough to be pathology. Boorse’s basic answer is that natural design and the natural level of efficiency and mode of function is determined by statistical analysis. But to utilize statistical analysis to distinguish pathology from non-disease, Boorse needs a statistical sense of normal function that both avoids any appeal to values or preferences and avoids the problem using simple statistical rarity as biological abnormality.

Boorse addresses the first issue with a theory of how proper function is determined. He relies on insights from several philosophers of biology concerning the fundamental difference between living organisms and non-living matter (even if very sophisticated in its chemical behavior). The fundamental difference is the fact that living organisms are goal-directed—namely, “they are disposed to adjust their behavior to environmental change in ways appropriate to a constant result, the goal. In fact, the structure of organisms shows a mean-end hierarchy with goal directedness at every level. Individual cells are goal-directed to manufacture certain compounds; by doing so they contribute to higher level goals like muscle contraction; these goals contribute to overt behavior like web-spinning, nest-building, or prey-catching; overt behaviors contribute to such goals as individual and species survival and reproduction (Boorse, 1977, p. 555).”
Thus the function of any part or process in the hierarchy is its “ultimate contribution to certain goals at the apex of the hierarchy.” The proper function of a sub-system (such as the beta-cells of the pancreas) are the contribution(s) it makes to the ultimate goals of the entire system. The function of the heart is thus to pump blood and not to produce heart sounds, because pumping blood is a contribution to the apical goals of the type of animal possessing such a heart, while making sounds is a by-product. The apical goals chosen by scientists may vary depending on the branch of biology or the species in question. For medicine and physiology, survival and reproduction are the ultimate goals. Boorse points out, however, that regardless of the apical goals specified, the matter of what contributions actually are being made by a particular subsystem to the specified ultimate goals is an empirical matter and not an evaluative one. Boorse also states that however the issue of determining standard or proper function is settled, as long as it can be done without merely appealing to human evaluation, it has little consequence on how most real-word cases might be handled by the biostatistical model of disease. Larry Wright’s (1973) analysis of function, Boorse thinks, could easily replace his own, without significant changes to the list of pathologies on his account.

Boorse realizes that simple statistical abnormality (i.e. rarity as abnormality) cannot, alone, serve as a means to distinguish disease from mere variation in form or function. He says:

…[T]here is a persistent intuition that the average person—or at least the average heart, lung, kidney, thyroid, etc.—must be normal, or we would have no way of telling what the normal person or organ should be like. I shall return to this intuition when we construct our functional account. Here we note only that statistical normality fails as a necessary or sufficient condition of health. It cannot be necessary because unusual conditions, e.g. type O blood or red hair, may be perfectly healthy. It cannot be sufficient because unhealthy conditions may be typical. No doubt the average person or organ is healthy in a particular sense of displaying no indications for treatment, but that is not the same as complete freedom from disease. Some of what medical tests consider disease processes are at work in virtually everyone below the level of clinical detection. There are also particular diseases—atherosclerosis, minor lung inflammation, perhaps tooth decay—that are nearly universal. In spite of these difficulties, we will give statistical normality an important role in our view, which shows that necessary and sufficient conditions are not the only possible components of an analysis (Boorse, 1977, p. 546).”
There are variations of form and function within species. Simple statistical norms would make type O+ blood normal and AB- abnormal. Brown irides would be typical while green would be rare and pathological. Boorse claims these sorts of polymorphisms can be (as they actually are) handled disjunctively as variants of normal. Another difficulty for statistical normality, also noted above, is pathologies that typically affect most species members, such as atherosclerosis. Simple statistical analysis would make atherosclerosis a normal biological process.

Boorse, citing inspiration from King (1945), thinks that a notion of statistically abstracted species-typical design can be saved:

The question therefore arises how the functional account avoids our earlier objections to statistical normality. King did explain how to dissolve one version of the paradox by saying that everyone is unhealthy. Clearly all members of a species can have some disease or other as long as they do not have the same disease. King somewhat grimly compares the job of extracting an empirical ideal of health from a set of defective specimens to the job of reconstructing the Norden bombsight from assorted aerial debris. But this answer does not touch universal diseases such as tooth decay. Although King nowhere considers this objection, the natural-design idea nevertheless suggests an answer that I suspect is correct. If what makes a condition a disease is its deviation from the natural functional organization of the species, then in calling tooth decay a disease we are saying that it is simply not in the nature of the species—and we say this because we think of it as mainly due to environmental causes. In general, deficiencies in the functional efficiency of the body are diseases when they are unnatural, and they may be unnatural either by being atypical or by being attributable mainly to the action of a hostile environment. If this explanation is accepted, then the functional account simultaneously avoids the pitfall of statistical normality and also frees the idea of theoretical health from all normative content (Boorse, 1975, p. 83).

Boorse sees that the sample over which one must average may include many defective individuals. Part of the problem is the ‘hostile environment’ which potentially effects all individuals that live in it. Once a particular sub-system X is isolated and a hypothesis function Y highlighted, how are we to determine what is the normal range of functional efficiency if all individuals potentially have diseased Xs at the time of observation? Similarly, perhaps X behaves differently in different environments, or at different times of the year, or even at different stages of the organism’s life. Taking account of the environmental effects is not the only factor that needs to be addressed. Variation that is
not necessarily pathological may be due to differences in age or sex of the individuals in a sample of conspecifics. Selecting the type of sample over which to average will also be important for these reasons. The sample should not be too large or completely random. Boorse suggests a reference class that shrinks the sample by taking into account age and sex.\(^\text{102}\)

The perfectly healthy individual or even the perfectly functioning organ or biological system is to some extent an abstraction or idealization. Boorse believes appreciating the effects of hostile environments and also carefully selecting the appropriate sample over which to generalize allows us to continue using a statistical notion of abnormal function. We avoid the problem of simply averaging over all individuals under all environmental conditions by selecting from a smaller sample in an idealized environment. Another modification allows inabilities or malfunctions even if the inability or malfunction is never actually manifest. Take the example of hemophiliacs that never experience the trauma necessary to demonstrate a dysfunction in clotting. Boorse replaces actual functioning with normal functional readiness or ability. These are the modifications Boorse makes to address some of the basic problem facing naturalistic statistical theories of disease.

Having made modifications, Boorse provides the following definitions:

1) The reference class is a natural class of organisms of uniform functional design; specifically, an age group of a sex of a species.

2) A normal function of a part or process within members of the reference class is a statistically typical contribution by it to their individual survival and reproduction.

3) A disease is a type of internal state which is either an impairment of normal functional ability, i.e. a reduction of one or more functional abilities below typical efficiency, or a limitation on functional ability caused by environmental agents.\(^\text{103}\)

\(^{102}\) The problem of the environment is even more difficult than the aspect addressed here by Boorse. How scientific models handle the environment, and particularly how they designate normal environments is important for disease claims. Conditions that appear to be disease or pathology may be normal (but undesirable) functions of a biological system placed within a harsh or unusual environment.

\(^{103}\) This last clause is one suggested by Boorse to handle common environmental damage to functional ability.
4) Health is the absence of disease (Boorse, 1997, p. 7).\(^{104}\)

These are Boorse’s naturalistic concepts of disease and health. Normal functioning is “performance by each internal part of all its statistically typical functions with at least statistically typical efficiency, i.e. at efficiency levels within or above some chosen central region of the population distribution (Boorse, 1977, p. 558).” Normal function is determined statistically over a sufficiently large enough sample to detect typicality but not so large or random as to miss it. Functioning below some determined minimum cutoff of efficiency is pathological and would constitute a disease of the part or process which is poorly contributing to the hierarchy of goals. Boorse says “abnormal functioning occurs when some function’s efficiency falls more than a certain distance below the population mean (Boorse, 1977, p. 559).”

For Boorse, biomedical researchers look at all of the members of some relevant group and note both the typical mode of function (based on statistically-typical goal-contribution) and the typical level or range of functioning (based on statistical analysis) of the system in question. Diseases are internal states of the organism. They are usually the result of proximal causes internal to the cutaneous boundary of the organism in question. However, there may be distal environmental contributions to malfunction or even direct proximal causation of malfunction (i.e. trauma) due to environmental factors. Careful attention to environmental effects is also therefore necessary. Biological sciences do carefully control environmental factors during laboratory experiments used in generating evidence for the models being produced.

Boorse does a good job distinguishing theoretical concepts concerned with health and disease from concepts referring to more practical, clinical concerns. “Contrary to some critics, I have never doubted that medical practice is permeated by values, nor that a good doctor must have more tools than a scientific knowledge of pathophysiology. I have not spoken to issues of clinical sensitivity for lack of anything original to say. But I did

\(^{104}\) Boorse offers this fuller definition of health in an earlier article: “Health in a member of the reference class is normal functional ability: the readiness of each internal part to perform all its normal functions on typical occasions with at least typical efficiency (Boorse, 1977, p. 562).”
insist on the value-ladenness of medical practice... [and continue to do so with] ...the distinction between disease and a richer family of practical concepts... (1997, p. 13).”

This is an effective distinction for Boorse and deflects many criticisms from the normativist camp based on concerns with practical problems beyond the concept of disease. He reminds the reader that being careful to keep the theoretical concepts of disease separate from the more practical concepts used in patient care, such as suffering or the stigma of being deformed, can prevent much confusion concerning the more evaluative aspects of medical practice. Boorse believes failing to separate the concept of pathology from these wider social and personal concerns causes much of the willingness for people to assume disease is an evaluative and subjective judgment.

When I first read Boorse I found so much to agree with that it took me years to discover how I subtly disagree with his theory. I still think it is an insightful account and a classic in the philosophy of medicine. We share admiration for the accomplishments of modern biological science, and physiology in particular. I agree with Boorse that the concept of disease as used currently and developed (over centuries) is not an evaluative one but a theoretical and descriptive one. Those practicing medicine or doing biological research currently are aiming at distinguishing normal from abnormal function when discussing disease or building explanatory models of disease. Usually such explanations offer details of unobservable parts and processes posited as the causes of the observable suffering or problems of the patient. The core concept is biological dysfunction—disruption of a physiological process or the malformation of an anatomical structure. I also agree with divorcing theoretical issues from the practical issues. Boorse’s discussion

---

105 Boorse has a very nice figure in his 1997 essay (Fig 2, p. 13) that lays out some of these other practical concepts. These are concepts that might be useful in issues of when and if disease can be detected by the tests available, whether it can or cannot be treated, whether the symptoms and problems brought to the physician are currently understood as disease or not, and issues with positive notions of health (i.e. those that go beyond merely the absence of disease and require other criteria). Theoretical health can also be distinguished from positive health and other practical health concepts. Theoretical (or negative) health for Boorse is the lack of any abnormal functioning parts or processes of the individual organism in question. Theoretical health is the absence of any disease. Boorse also discusses positive health as the idea of functioning above the minimal efficiency required to not be pathological. Likewise Boorse discusses diagnostic health, which is lack of any detectable or diagnosable subnormal efficiencies of functionality, and therapeutic health, which is lack of any treatable pathology.
of the interlocking hierarchy of functions produced by the piecemeal work of constructing biological models for circumscribed systems at different but related levels of analysis is insightful. I agree that it does not particularly matter for the concept of disease how functions are analyzed, and different model builders might have different ideas about the concept of function or even about what warrants that type of analysis of natural objects. But, like Boorse, I believe evolution explains why functional analysis is so fruitful.

Where I disagree most fundamentally with Boorse is his reliance on the statistical sense of normality. I understand the appeal. It appears as if statistical determination of normality and abnormality can produce this distinction in a way that is free of any value judgment. The fundamental problem is how to distinguish insignificant variation from significant variation purely by calculation and without a human agenda, preference or value. How is it determined that some variations are just unimportant small differences around the mean, while others are significant outliers? Human choice and perspective will be involved at some level. The details about how such human choice or perspective is involved and how it is controlled are important details. On my account it is crucial to understand how scientific models are created, and how they are improved, tested, refined and eventually gain the respect that comes with being judged as unbiased and accurate representations of the phenomena being explained. Chapter 2 suggests how philosophers and sociologists of science ought to continue to study that process. Ultimately appeal to the statistical sense of normality cannot by itself distinguish normal variation from pathology. I discuss this in Chapter 3. This was part of Amundson’s criticism of Boorse-like attempts to define the biologically normal.

Boorse’s position attempts to have judgments of abnormality made in reference to an ideal of normality. This ideal of normality is a statistical summation. Boorse says: “Clearly physiological function statements are about a trait’s standard contribution in some population or reference class, e.g. a species. A text may say that the function of the human lens is to focus light on the retina. This claim is not falsified by the existence of people with cataracts, or no lens at all. Similarly, one case of an animal’s life being saved by some characteristic would not be enough to make this effect a biological function. One squirrel might catch its tail in a crack en route to being run over by a car, but that would not make defense against cars a function of the squirrel tail. The statement about the human lens is true because it is overwhelmingly typical of members of the

---

106 Boorse says: “Clearly physiological function statements are about a trait’s standard contribution in some population or reference class, e.g. a species. A text may say that the function of the human lens is to focus light on the retina. This claim is not falsified by the existence of people with cataracts, or no lens at all. Similarly, one case of an animal’s life being saved by some characteristic would not be enough to make this effect a biological function. One squirrel might catch its tail in a crack en route to being run over by a car, but that would not make defense against cars a function of the squirrel tail. The statement about the human lens is true because it is overwhelmingly typical of members of the
abstraction away from any particular organism, but still is an appeal to the actual organisms. I agree with Boorse that appeal to an ideal is necessary for judging biological abnormality. However, the ideal cannot simply be some statistical average. As discussed in Chapter 3 when examining Amundson’s position, statistical averaging alone cannot distinguish significant from insignificant variation. My own position makes clear that judgments of disease must be made against an ideal of normality. The ideal, however, is the abstract and simplified scientific model in question—a creation of human imagination and investigation. The model is not merely a summation of observation. This is an important aspect of disagreement between Boorse and myself. The model (for a legitimate disease claim) is designed to distinguish what counts as normal variation and what counts as pathology. This leaves open the possibility that models may contain blatant biases and may contain elements of conventional judgments of normality. This feature of my account may be seen by some as a concession to normativism in allowing conventional value judgments to enter into disease claims.

I disagree with Boorse on his goal-directed analysis of functions. I have not included an argument for a particular analysis of functional ascriptions in biology because it is a topic too complex to briefly discuss, and because I agree that the fine details matter little for a theory of disease. There is no need to give a global understanding of function or its warrant. Globally, science is still struggling to define the goals or proper function for the higher order systems of the human body, and uncertainty population for their lens to contribute to their survival and reproduction in that way. In general, function statements describe species or population characteristics, not any individual plant or animal. As a result, the subject matter of comparative physiology is a series of ideal types of organisms: the frog, the hydra, the earthworm, the starfish, the crocodile, the shark, the rhesus monkey, and so on. The idealization is of course statistical, not moral or esthetic or normative in any other way. For each type a textbook provides a composite portrait of what I will call the species design, i.e. the typical hierarchy of interlocking functional systems that supports the life of the organism of that type. Each detail of this composite portrait is statistically normal within the species, though the portrait may not exactly resemble any species member. Possibly no frog is a perfect specimen of *rana pipiens*, since any frog is bound to be atypical in some respects and to have suffered the ravages of injury or disease. But the field naturalist abstracts from individual differences and from disease by averaging over a sufficiently large sample of the population. The species design that emerges is an empirical ideal which, I suggest, serves as the basis for health judgments in any species where we make such judgments (Boorse, 1977, p. 556).”
still exist for designations involving cognition and behavior. Physiology does quite well by allowing the fine details of function to be settled during the construction of models of isolated systems. I hold the view stated by Cummins (1975) and elaborated by Hardcastle (1999, 2002).

4.3 NORMATIVISM: ENGELHARDT ON CREATING MEDICAL REALITY

Normativism is the position that disease is an evaluative concept and not primarily a descriptive or theoretical concept. Normativist emphasize the consequences of a particular disease and human judgment about these consequences. Normativists tend to highlight the failings and biases of particular scientific research projects. Regarding the concept of normality in biology, normativism is often the position that there is only one sense—the conventional sense. When this is the case, legitimacy for normativists is a matter of gaining the willing acceptance of a disease claim by a majority of those in a community via some form of fair negotiation. Radical normativists may see no differences between legitimate and illegitimate claims of disease, except that some have gained acceptance over others.

H. Tristram Engelhardt is frequently cited as an exponent of the normativist position on disease. He synthesizes historical, philosophical and medical perspectives concerning human disease and produces a complex picture of the nature of disease claims. Drawing on his interpretations of Hegel and Peirce, he stresses the perspectival nature of all knowledge, as well as its being culturally and historically conditioned. Humans ‘see’ phenomena through a ‘lens’ of multiple assumptions and expectations that come from our particular perceptual and cognitive abilities, our cultural values, customs and projects, and our education, as well as from the theories and explanations we already hold to be true. He says:

The world in which we live is not furnished by uninterrupted facts. We see things around us in terms of social and theoretical expectations. We are taught early how to explain the occurrences of our world. Within the generally dominant scientific world view, we take for granted that a set of complex, etiologic forces directs the production of illness and disease. Individuals in other cultures, or our antecedents in our own culture, untutored by our current scientific world view, do not or did not see illness as a result of infectious agents, genetic flaws, or endocrinological abnormalities. We, however, do. Our world is structured by a special set of
assumptions about the rule-governed character of our experience. These scientific and metaphysical presuppositions fashion for us our everyday expectations. They give shape to our lifeworld. In addition, the particular character of our social intuitions invests occurrences with social significance… We see the world through our social, scientific, and value expectations. The medical facts with which bioethics deals are not timeless truths, but data given through the formative expectations of our history and culture. Recognizing a state of affairs as heart disease, cancer, depression, homosexuality, or tuberculosis is a rich and complex process. All knowledge is historically and culturally conditioned, and the influence of history and culture is often, as we shall see, particularly marked in medicine. This is not to say that investigators do not attempt to know, timelessly unconstrained by social and cultural forces. In endeavoring to know truly, one attempts to understand the world as it would be seen from God’s eye, from the viewpoint of dispassionate, scientific observers or investigators, so that the findings could be shared with other investigators, even those outside our culture—in principle, even with alien investigators on planets circling distant stars (Engelhardt, 1996, p. 190).

Therefore, despite efforts to present knowledge as purely objective and from no particular perspective, all scientific models will be perspectival and informed by cultural and historical period. Our own situation, even if we believe it to be more enlightened, offers no advantages, in these respects, over other knowers.

Medicine is not a purely theoretical science but an applied science. It offers interventions and interacts with humans at the level of their goals and plans, and at the level of their understanding of themselves and their role in the community. Because of these practical and social aspects, the art of medicine and its theory are even less capable of abstracting towards a ‘God’s eye view’ than purely theoretical sciences. Engelhardt says:

Portrayals of reality are cultural products. Although there are constraints placed upon us as knowers by the given character of the objects, objects appear to us through our concepts and in terms of the conditions of our experience. In the case of the unapplied sciences, where one’s choice among different construals of reality is dictated by the ideal of a fully intersubjective account free of idiosyncratic values and purposes, there is a commitment to avoid imposing the values and perceptions of one particular group of knowers. Within an unapplied science one attempts to know anonymously and impersonally. But even here portrayals of reality are fashioned through background commitments that involve choices among different epistemic values. The world of unapplied science is a compromise made in the face of controversies. In applied sciences where the role of non-epistemic values is more salient, the constructed character of reality is
easier to recognize. The goals that medicine can apply are manifestly varied and dependent on the visions of particular individuals and communities... There are choices to be made among alternative accounts of medical reality. Since these choices are not simply determined on epistemic grounds, we become accountable for ways in which we fashion that reality. How should one choose among competing descriptions of reality? The question is, Who gets to choose?...

Communities must begin with a recognition of the constructed character of medical reality. This recognition underscores our choices and indicates our responsibilities as individuals who not only know reality but also know it in order to manipulate it. One must also recognize that these manipulations tend to be communal. The issue of who decides is thus moved from the area of individual free and informed consent to a communal area of negotiations regarding construals of reality (Engelhardt, 1996, p. 226).

For Engelhardt, medical reality, including what is or not legitimately taken to be disease, is a social construction. This constructed reality is dependent on community and individual choices and we have a responsibility to participate in the creation (by negotiation) of this reality.

So biology as science may be partly concerned with the accurate, unbiased descriptions of biological processes. But medicine, as applied science, seeks explanatory models because it has as a central goal the control of biological processes so as to alleviate suffering. But, of the many problems in a community that can cause suffering, only some are going to be seen as medical issues. Participation in this determination of the goals and problems of medicine is how individuals can influence the creation of the medical reality that otherwise informs our view of such matters. Engelhardt says:

Each of the major social institutions identifies problems for its care in terms of sets of values that make problem situations stand out as inviting interventions, as failing to meet a standard, as a difficulty to be set aside. The institutions of education, religion, morality, law, and medicine in this fashion variously characterize circumstances as those of ignorance, sinfulness, blameworthiness, criminality, civil liability, or disease. To see circumstances as one of sinfulness, criminality, or disease is to place it within the province of one of the major social institutions with its peculiar models of explanation and with its own special directing goals. The facts available within the spheres of religion, law, morality, and medicine are seen as problems of a particular kind in terms of particular webs of values, descriptive conventions, explanatory models, and social roles. Religious accounts of reality involve supernatural causal models dependent on views regarding the final destiny of individuals and the universe. It is in this fashion that religions give ultimate meaning to life, suffering, and death. Legal
accounts will incorporate particular systems of evidence and proof, which
determine which findings can be assessed in what ways in order to achieve the
goals of particular practices of blaming and praising. One might think here of Hart
and Honore’s classic account of causation and the law. As they point out, when
flowers in a garden wither due to a gardener’s failure to tend them, the gardener is
held as having caused the flowers to die, though from a more neutral perspective,
the failure of a passerby to water the flowers and the absence of sufficient rainfall
are equally causes of the flowers dying. It is the social presumptions regarding the
duties of gardeners that make these other circumstances assume the role of
background conditions and highlight the gardener’s failure to water the flowers as
the cause of their dying. A set of social relevances is employed to establish a
particular account of causation, so as to identify certain causes because of their
role in a social practice. The result is that a set of social expectations and values
frames a context for experience and action (Engelhardt, 1996, p. 222).

Engelhardt highlights the complex evolution of social practices and values that create our
legal and religious expectations and even theories of causation and practices of
explanation. The boundary between legitimate and illegitimate disease claims is
determined in a manner importantly similar to how the boundaries between legitimate
and illegitimate crime and sin are determined.

Engelhardt explains how for any individual at a particular place and time, their
view of disease is conditioned by the ‘medical reality’ that exists for that community. He
discussed four broad “clusters of concern” that are important for shaping any particular
medical reality. These four complex, culturally relative sets of values, expectations, and
practices inform what problems will be recognized as diseases and which not. Engelhardt
discusses: 1) disease claims as influenced by the culture’s practices and expectations
concerning making evaluative judgments. He also discusses disease claims as relying on
the current conventions and practices of both 2) description and 3) explanation. Further,
disease claims are shaped by and help shape 4) the particular social roles involved with
being sick, being a patient, being a physician or healer, etc.\textsuperscript{107}

Disease claims are evaluative for Engelhardt. “Problems stand out as problems for
medicine because they are disvalued. They are seen as pathological. They are associated

\textsuperscript{107} He calls these complex clusters ‘languages’ such as in the ‘disease language as
evaluative,’ but I find it more instructive to continue speaking of disease claims.
Following this terminology alteration, Engelhardt would discuss ‘disease claims as
evaluative,’ etc.
with pathos or suffering, and suffering is judged, all else being equal, as a disvalue (Engelhardt, 1996, p. 203).”

To claim that some condition is a disease is to pass a value judgment, and the judgment is a comparison of the condition to an ideal conception of form, function, ability, etc. Such judgments are ultimately conventional, in that they appeal to the complex web of moral and nonmoral values concerning human form and function prevalent in our community. These values are learned from and shared by the community. “Abnormality is recognized as abnormality within a particular context of expectations. For example, if one steps away from moral traditions with strong notions of certain actions being improper, it is difficult to make sense of consensual acts being perverse or unnatural. Consider the examples of a brother and sister who are competent, sterile, and unmarried who decide at the age of fifty to have intercourse. Is such an act unnatural? Is it perverse? Is it wrong? (Engelhardt, 1996, p. 199).” Disease, like perversity cannot be perceived except through a set of assumptions, values and expectations about what is normal and abnormal. And these values and assumptions need to be shared by members of the community in order to be successfully appealed to. This is demonstrated when members of communities without shared values and expectations encounter each other and cannot arrive at the same judgments. In extreme cases, the two groups cannot even understand what the other is claiming. Such shared values, expectations, and social practices are required whether making claims about the perversity of incest or the pathology of diabetes. “One draws a line between innocent physiological or psychological findings and pathological findings because of particular human values in a particular circumstance, not because of the discovery of an essential distinction that exists outside of particular human expectations… The more nominalist or

---

108 He also says: “To see a phenomenon as a disease, or deformity, or disability is to see something wrong with it. Disease, illness, and disfigurements are experiences as failures to achieve an expected state, a state held to be proper to the person afflicted. This may be a failure to achieve an expected level of freedom from pain or anxiety. It may involve a failure to achieve an expected span of life. These genres of judgments characterize a circumstance as one of suffering, one of pathology, one of a problem to be solved. Such judgments may be made either by the individuals afflicted or by others regarding those individuals. In being characterized as perverted, diseased, or deformed, and adverse judgment is rendered. This is the case even with a disfigured nose, for which the bearer seeks cosmetic surgery (Engelhardt, 1996, p. 204).”
instrumentalist views of disease are better justified in that they eschew the ontological quest to discover the essence of disease (Engelhardt, 1996, p. 205).”

Engelhardt criticizes Boorse’s attempt to specify a value-free concept of disease by appeal to statistical typicality and evolutionary design. Engelhardt correctly perceives the central problem as revolving around the difficulty in distinguishing normal variation from pathology because evolution often produces a spectrum of similar designs each with slightly varied levels and modes of functionality. Evolution, notes Engelhardt, is also a historical and backwards looking theory. Without some normative viewpoint—some ideal of what is supposed to be normal—there is no way to separate out what range of functionality or variants of design are to count as typical or normal. At best, Engelhardt sees Boorse’s concept of species-typical design as something of interest only to zoologists and other theoretical scientists. It is far removed from the practical issues of medicine, and offers little help in explaining why certain variations are seen as disease while others are not.

Besides relying on our community’s expectations and values regarding what is biologically normal and abnormal, the legitimacy of a disease claim also relies on the practices, values, and expectations involved in describing and explaining a shared reality. “Descriptions require standardization of terms. Such standardizations will be fashioned through quasi-political or societal discussion and against background assumptions about what will be useful in achieving particular goals and purposes. Those assumptions are themselves structured by explanatory views (Engelhardt, 1996, p. 208).” Engelhardt discusses how certain signs and symptoms become describable in standard ways once we begin to see them as related to disease. Adopting a theoretical framework makes us see the world differently. The physician sees Koplik’s spots on the buccal mucosa of a child, not merely some small reddish dots inside her mouth. Engelhardt describes how at different historical periods theoretical frameworks within which physicians worked allowed for very different interpretations of evidence. It also allowed different ideas about what constituted a proper model of disease. For many centuries things we now consider just signs or symptoms of disease, such as fever or pain, were in themselves types of diseases. These disease claims had their own presumed causes and remedies. Engelhardt also reminds us that the very notion of causation—what counts as being a
cause—is neither unequivocal amongst professional philosophers, nor across history or culture.

Engelhard considers the issue that some cases of disease (such as heart attacks) are not as culturally relative as others. To the extent this is true, it reflects that certain things (like death) are disvalued and are counter to basic goals no matter what the cultural circumstances. Some conditions fit within almost any imaginable framework for defining problems that are involuntarily caused and disvalued states of the mind and body. Considering that disease claims are dependent on the evaluative and explanatory practices of the community that deploys them, Engelhardt suggest that we may want to dispense with the term disease and talk of clinical problems. “Given these concerns, one might even wish to eliminate or strongly qualify the term disease in situations where misunderstandings could arise. One might think of substituting the term clinical problems to identify those difficulties that stand out as conditions that ought to be addressed and solved by medicine. Such a term would more accurately indicate the ways in which clusters of value judgments make conditions stand out as problems to be treated (Engelhardt, 1996, p. 205).”

Rephrasing Engelhardt’s position as one regarding the legitimacy of specific disease claims is helpful. For Engelhardt, to be a legitimate disease claim, a condition must be judged to be harmful or otherwise disvalued by the community in question. It must also be seen as something not avoidable by means of sheer willpower or choice. Additionally, it must be recognized as fitting into the causal matrix of a physiological or psychological explanatory framework currently used by one’s community and culture. In other words, the claim is understood within a physiological, anatomical, or psychological causal matrix consistent with whatever the prevailing theories are concerning these subjects. To be seen as a disease a condition must be seen as abnormal against a background of shared community expectations and values concerning what is normal regarding human form, function, ability and behavior.

On Engelhardt’s account, controversy arises when smaller groups within a large community have different sets of values and expectations about what is abnormal regarding human form, function, and behavior. Take for example the current controversy over whether obesity is a disease or merely a series of poor lifestyle choices. Groups may
also differ over expectations, values and practices concerning explanation or what constitutes legitimate evidence. The current controversies over autism’s link to vaccines is an example. What counts as evidence for scientists and physicians differs greatly from what counts as evidence to observant mothers of autistic children. Engelhardt speaks of ‘confronting a moral stranger’ to illustrate the problem of trying to discuss issues of disease claims with those holding differing basic assumptions, values and expectations. We experience the confrontation of moral strangers when we attempt to persuade or negotiate with those operating within a significantly different medical reality. Catholics, conservative Christian fundamentalist, Muslims, feminists, secular liberals, etc. may at times confront each other concerning a controversial disease claim. These groups have different sets of assumptions concerning normality, the human good, and what constitutes appropriate evidence and explanation. This problem occurs within our modern secular societies—large communities without unifying religious or metaphysical principles—and it is often a problem encountered when trying to understand disease claims from distant cultures or historical periods.  

For Engelhardt, disputes over disease claims will have to be settled by negotiation, unless one set of values and practices comes to dominate by fiat or force. It is likely larger communities will have to accept multiple schemes of disease categorization. Such differences in understanding regarding what it means to know truly and decide fairly are often at the root of conflicts regarding the proper characterization of medical facts, or regarding whether a problem should be understood as a medical, legal, or moral problem… The choice of how to view a circumstance is not simply an epistemic or knowledge-based determination. It is also a

109 Engelhardt says: “Particular scientific communities at particular times embrace particular facts, findings, and rules of evidence and inference that we later find to be idiosyncratic, just as particular moral communities involve the acceptance of particular moral rules and views of the good life. Scientific controversies can occur between individuals united in a general commitment to science as an endeavor of intersubjectivity establishing claims regarding the nature of the world, but divided due to different particular understandings of rules of evidence and inference. When the debates involve scientific issues with heavy ethical and political overlays, the conflicts become, as one would expect, complex. The individuals involved in such controversies will be participants in different moral and scientific communities and, as a result, will be in conflict with regard to different understandings of knowing truly and deciding fairly (Engelhardt, 1996, p. 225).”
determination based on a set of value considerations. The very characterization of reality can thus become a moral issue. The very naming and characterization of a problem can raise questions of what values ought to be invoked, and of how they ought to be ordered. It will raise as well the question of who should participate in framing the classifications. And whose hierarchies of costs and benefits should have precedence. This issue must be answered in part, as has been indicated, through the practice of free and informed consent. It must also be answered through public acknowledgment of the ways in which problems are classified as medical, legal, religious, and so on. Since medical characterizations of problems depend not just on knowing reality truly, but on deciding in a fair manner among various ways of classifying reality, problems of individual rights, democratic prerogatives, and rights of privacy become salient. If classifications are natural, no one is at fault for describing reality as it is. One may not like the way reality is, but the scientist has not taken away anyone’s rights by describing things the way they are. However, if one is fashioning classifications in order to pursue particular goals, the choice of the particular goal is open to negotiation among those involved (Engelhardt, 1996, p. 225).

…different constructions of medical reality can be embedded in alternative health care systems, which carry with them quite different understandings of what should count as a disease to be treated and what treatment expenses should be sustained by the community. Within different communal systems, there may very well be different notions of the sick role and the role of the physician versus that of the priest. Since there are numerous understandings of medical reality, those who so wish should be at liberty to act on their own moral and metaphysical visions in the company of consenting collaborators… There is no canonical content-full secular vision of medical reality, of illness and disease, of health and proper health care… Given the limited secular moral authority of the state, and given the diversity of the moral and metaphysical visions of medical morality, there should be, to paraphrase Mao Tse-tung, space for a thousand views of health care to develop and for a hundred different systems of health care delivery to contend (Engelhardt, 1996, p. 227).

Essentially people can negotiate over the legitimacy of a disease claim or agree to disagree. Conventional senses of normality and abnormality must be negotiated. Appeals to nature cannot distinguish what problems are to count as medical problems. The illusion of science delivering a morally and politically neutral boundary between normal and abnormal that would help settle these issues should not tempt us, for the same reasons we should not allow force to simply impose a medical reality.

Engelhardt’s is just one normativist account, but it captures the strengths of the position and explicitly confronts (and accepts) what I believe are the counter-intuitive results of a strong version. Normativism about disease suffers from the classic
philosophical conundrum of either saying something accurate but uninteresting, or very interesting, counter-intuitive, but highly inaccurate. If all the normativist wants is to discredit a picture of science and medicine as infallible producers of purely descriptive, unbiased knowledge then I would agree.110 However, the normativist position implies much more than this. These more radical implications are the ones that make the position interesting and counter-intuitive.

Part of this more radical position is the claim that science and its products share significant similarities and few dissimilarities with other socially constructed institutions and their products--institutions such as religion, law or politics. This is an attempt to discredit or downgrade the special (and potentially dangerous) authority that science has garnered in Western secular societies. I would agree that this authority does need to be challenged in some circumstance. However, globally discrediting all scientific explanations or undermining scientific authority regarding empirical matters is a foolish project.111 The counter-intuitive result of viewing science this way is that all issues of legitimacy become political and ethical issues. It follows that all disputes about the legitimacy of disease claims should therefore be handled by fair negotiation, not expert authority or further empirical investigation. Legitimacy becomes an issue of the moral and political acceptability of the details of the explanatory model. Even if one avoids taking the activist stance, and takes a sociological stance, the legitimacy issue concerns whether the sense of abnormality claimed ‘fits’ with the social expectations, values, and practices already in place (i.e. can the current claim be understood as legitimate through the lens of our community’s medical reality). Normativists discount or ignore the possibility of issues about the ‘fit’ between the models explanation of biological abnormality and the evidence gathered. The accuracy of representation is ignored as a factor in legitimacy. It is then easy for normativists to hold that all judgments or claims about normality and abnormality involve the conventional sense of normality.

110 See Chapter 2.
111 One reason normativists and other social construction activists think they should push such a radical position is because they believe scientific authority is so ingrained and unquestioned that only a radical, counter-intuitive statement of their case will awaken people from their mesmerized homage to science and its dictates.
The central problem with normativism develops from over-emphasizing certain examples and drawing counter-intuitive (but interesting) sweeping generalizations. The expectations and values held by the normativist influence their selective choice of examples of disease, which in turn, once analyzed, reinforce their position. Homosexuality, drapetomania, masturbation, and hysteria are all frequently discussed.\textsuperscript{112} The problem for the normativist becomes how to explain the less controversial and less bizarre claims of disease—those that do not seem culturally relative like type 1 diabetes, Grave’s disease, cholera, or spina bifida. It is difficult to overcome intuitions that conditions like these, especially when coupled with detailed, reliable explanatory models, are biological malfunctions independent of any cultural or personal evaluations or perspectives. It is hard to imagine these conditions might not have been considered biological abnormalities had our culture developed differently or our values and expectations been different. Normativists explain the difference between legitimate and illegitimate disease claims as resting on complex cultural developments. I find such normativist accounts unsatisfying.

Engelhardt acknowledges that some disease claims are less culturally relative. He gives the example of a heart attack at one end of a continuum of cultural relativity. “Thus, on the one hand, crushing substernal pain radiating down the left arm accompanied by weakness, collapse, and a feeling of impending death is likely to make myocardial infarction a disvalued circumstance across cultures, even if there are appreciable differences in cultural values regarding human function. On the other hand color blindness, the inability to roll up the sides of one’s tongue, or the incapacity to taste phenylthiocarbamide may or may not count as genetic diseases or defects, depending on the environment in which the person lives and the goals they and their culture support (Engelhardt, 1996, p. 204).” It is important to carefully distinguish the outward signs and symptoms of what we may suspect is a myocardial infarction, from the actual disease itself. First, these signs and symptoms (i.e. chest pain, weakness, sweating and fearing impending death) may be considered signs and symptoms of a divine calling in some cultures—signs of a supernatural force taking hold of a person. I see no reason why the

\textsuperscript{112} I doubt anyone would know of drapetomania as a disease claim if not for the normativist literature in the philosophy of medicine.
interpretation of these symptoms is not as culturally relative as the interpretations of color blindness. Second, within a Western medical framework these symptoms are not necessarily associated with myocardial infarction, they might be caused by other pathologies, some leading more directly to death or harm than others. It is the cause—seeing them as associated with a certain hidden biological malfunction—that allows signs and symptoms to be part of a legitimate disease claim and allow us to pick the correct therapy and avoid the wrong ones. Heart attacks (i.e. MIs) and cholera are less controversial and less culturally relative because the explanatory models upon which the claims rest are unbiased, and accurate. We feel comfortable that these problems are caused by biological malfunctions and we find the explanations accurate and unbiased. These claims also involve appeal to the sorts of processes and biological functions that are not open to direct observation and thus not easily open to evaluation by the average member of the community. The average community member may judge the consequences of a heart attack as (conventionally) undesirable and bad. However, the sense of abnormality involved in its being a disease is not conventional. Even assuming Engelhardt is correct that some symptoms are disruptive enough to be disvalued in all societies, this does not explain why such societies interpret these as biological phenomena. Being disvalued does not guarantee that a recurrent kind of problem will be seen as a biological malfunction instead of a spiritual or criminal matter.

---

113 The symptoms could be from a pulmonary embolism, an aortic dissection, a panic attack, a pheochromocytoma, esophageal spasm, flash pulmonary edema, etc. Engelhardt adds certain details to the symptoms and history that make MI a likely first guess, but on just these details alone the diagnosis cannot be made with enough confidence to intervene only for MI and not test or watch for the other diseases.

114 Engelhardt provides other factors needed to constitute a disease claim beyond being disvalued. I think this is a hedge due to the fact that there can be cases where the perception of value is not correlated with the theoretical assessment of biological abnormality. This is particularly true if we look at cases of diseases in plants and animals, where theoretical biological normality and abnormality are much less well correlated with human goals, values and expectations. Humans often manipulate animal and plant abnormality to achieve valuable goals—there are many examples from pest disposal. The basic point is that assessing biological abnormal function and assessing the perception of a particular community of humans about whether the consequences of this are harmful, valuable, good, bad or indifferent are two different assessments. Such assessments can and do diverge.
I disagree with the normativist emphasis on the consequences of diseases and biological abnormality at the expense of issues of causation. I believe that discerning the causes of problematic clinical signs and symptoms in order to control them has always been at the heart of medical practice. Disease has always implied an unseen or esoteric abnormality that causes the perceived or known problems. Discerning the true or hidden cause is the art of diagnosis. Over time the theoretical framework within which causes are explained has changed, as well as the technology and procedures to investigate increasingly remote aspects of the causal processes. I agree with Engelhardt that the practices and assumptions involved with explanation and even causation may have changed, however the notion of biological abnormality has always been part of the core conception of disease.115

Engelhardt dismisses Boorse for focusing on a purely theoretical concept and trying to separate this from practical or clinical issues. He dismisses Boorse’s concept as of interest only to the zoologist or plant biologist. Engelhardt stresses that medicine is an applied science, even suggesting we might stop using the term disease and instead use the term ‘clinical problem.’ Boorse, however, was correct to separate theoretical and practical issues. First, an applied science does not necessarily have trouble with such a separation of concerns. Civil engineering is an applied science, and there is no conceptual difficulty separating the descriptive or theoretical details that explain why a bridge failed and collapsed, from the social interpretation or consequences of the event. Natural disasters, like earthquakes, can also be explained in descriptive and theoretical terms regarding causation, regardless of whether they do or do not have catastrophic effects on humans. Secondly, the issue about separating practical issues from theoretical is related to the failure to distinguish signs and symptoms from causal explanations. Signs and symptoms are clinical problems. Dealing with them results in all the complex practical issues Engelhardt notes. These symptoms must be evaluated as harmful by the patient

115 I have not done explicit historical research. However, I would argue that across culture and time the concept of disease is a ‘cluster concept’ and can involve varying core elements. Over the past 200 or so years or so, the notion of biological abnormality has displace other core elements in the concept of disease. Being painful, disabling, or otherwise stigmatizing are no longer essential parts of the understanding of disease in modern western medicine and biology.
(and community) to be the sort of problem that leads one to seek medical intervention. Many abnormalities of biological function that do get people’s attention are ones that cause conditions perceived to be harmful, dangerous or undesirable and lead to people seeing their doctors. However, not all clinical problems (or symptoms) are caused by disease. There is not reliable correlation between symptoms and clinical problems that are brought to doctors and signs and symptoms that are caused by disease. Current American medical practice often chooses to ignore some diseases, while, on the other hand, chooses to treat some clinical problems considered theoretically to be normal variation. The medical community often chooses not to ‘treat’ albinism, but does intervene to treat noses that are too big or breasts that are not the desired size or shape. I shall discuss more cases in the next section. There is no practical or theoretical reason why we cannot (or should not) continue to separate theoretical issues from practical issues in medicine.

Once we separate the clinical and social issues of dealing with human patients from the theoretical issue of biological function, many of the points that Engelhardt makes are worthy of note. Many of his points apply to ‘clinical problems’ and with treating symptoms. It is difficult to understand the full nature of the problems that an individual patient has, and many aspects of discovering and alleviating these problems involve deploying evaluative and prescriptive judgments. The way that members of our community perceive people who have particular medical problems has a huge impact on individual lives and on the community. The roles those labeled as diseased are expected to fulfill, etc. are complex but important topics of study. I do not mean to imply that they should not be pursued. However, I do believe that the theoretical issues of what constitutes a disease for pathologists, physicians, and the biosciences can be separated from these issues of clinical care and the creation of a particular community’s medical reality. The distinction here is one often painfully recognized by third year medical students, freshly arrived to the clinic and ward, as they discover that understanding and diagnosing pathology (or even intervening) is only a small part of what patients expect from their doctors.

I do not agree that negotiation is the appropriate means to distinguishing legitimate from illegitimate disease claims. In negotiations over a controversial disease
claim, with people who do not share our moral values and social conventions, we can in certain circumstances appeal to scientific models, and we can offer to gather more evidence and either re-confirm the explanatory model in question or revise it if needed. The case of parents insistent that scheduled childhood vaccine panels cause autism is one example of offering more evidence to convince those with different assumptions that a particular model of biological abnormality was inaccurate (Institute of Medicine, 2004).

In the case of autism and the MMR vaccine, the scientific boundaries need to guide political policy. In certain circumstances there may only be hypothetical and speculative models. But knowing that is the situation, and that therefore only political and ethical negotiation may occur until more evidence is available, is significant. Ultimately this returns to issues about one’s faith or trust in contemporary science and its ability to provide accurate and reliable explanations. Are the scientific explanations supporting legitimate disease claims better than those behind illegitimate or putative claims? Is there more to being a disease than the perception by the community that the condition is undesirable, not voluntarily controllable, but potentially controllable via medical intervention? Ultimately the strong normativist is beholden to actual community practice and whatever consensus can be reached—there are not matters of more or less accurate explanations of etiology. There are just different explanations that happen, for the time being, to fit with the expectations, values and practical needs of the community. On this radical account there is also nothing like an actual cure for a disease—what constitutes a cure is also relative to the community and a matter only of perception. Regarding the disputed legitimacy of a disease claim, my position gives priority to the quality of the scientific explanatory models involved. If these models exist, and are unbiased and accurate, then the boundary between normal and abnormal biological function might help settle other practical or ethical boundary disputes, such as those mentioned at the outset of this chapter. For the normativist, my position gets the order of priority wrong. The normativist position holds that by settling the complex political and ethical issues involved in many of these boundary disputes, we clarify the expectations and values needed to distinguish legitimate and illegitimate disease claims.

I sketched my view of science and what it produces in Chapter 2 and discussed the different conceptions of normality in Chapter 3. I agree with many of the subtle points
Engelhardt raises when discussing the factors that go into the creation of knowledge by a community of people with shared goals regarding understanding and controlling a phenomenon. In creating and refining scientific models there is a complex interplay of perspectives and practices and there is opportunity for people to ‘talk past’ each other if they operate from positions and world-views that rest on importantly different assumptions and values. I would agree that scientific models are social creations in a strict sense and are the products of humans with perspectives and agendas, and who reside within a culture and in communities. Such creatures cannot step outside of their culturally conditioned viewpoints. However, I also have reasonable faith in the self-correcting nature of the social phenomena and institutionalized practices that have come to be called the scientific ‘process’ or ‘ethos.’ I believe that models can increasingly be improved and refined to gain more fit with the actual phenomena being represented and also decrease the amount of bias and conventional evaluative judgments involved. It is possible to accurately represent salient features of phenomena in simplified models. I agree that scientific communities and individual scientists have passed off (and might try again to pass off) value judgments as if they these were natural facts. However, this does not implicate all scientific models as biased or as incapable of offering usefully objective and reasonably unbiased empirical explanations of biological phenomena. There are important differences in the quality of scientific models and explanations that serve as the foundation of different disease claims.

I agree with Engelhardt that even a modified statistical sense of normality (such as the one used by Boorse) is insufficient to address the problem of distinguishing insignificant from important variations in biological form or function. I agree with Engelhardt that judgments of abnormality are made in reference to an ideal. For Engelhardt, this ideal of normality must be a product of expectations or value judgments about human form or function.116 I do not agree, and believe that some judgments of abnormality, particularly many having to do with disease, are made in reference to the ideal of normal function specified in a biological model. There are important differences in the sense of normality offered in accurate biological models compared with the sense

116 This can either be explicitly held and perpetuated by individuals in a society or implicitly part of the community’s social practices and institutions.
used in the evaluative judgments we make about observable consequences of disease. Frequently, many of the people making the claim of abnormality, as in those labeling someone a diabetic, have little or no knowledge of the actual details of the model—no knowledge of the details of normal function. However, those making claims can recognize that the claim is not one made against a social convention or socially negotiated value, but rather in reference to a theoretical sense of normal biological function. When a lay-person calls their friend diabetic and therefore diseased, he knows that there is an explanation of theoretical normality available from experts and in textbooks, even while he might admit he does not know the details.

In contrast to his analysis of disease, I do believe Engelhardt may have a valuable point about the *positive* conception of health. This may require more than a piecemeal set of biological models of normally functioning systems. It may require a content-full vision of the capacities and abilities necessary for human flourishing.

4.4 MY POSITION: DISEASE AS THEORETICAL BIOLOGICAL ABNORMALITY

I consider Boorse and Engelhardt to be good representatives of the naturalist and normativist positions on disease. I have presented their positions above and highlighted what I think insightful and helpful, as well as highlighting where I think their positions go wrong. Naturalism correctly sees disease claims as descriptive and not primarily evaluative. Naturalists, however, struggle with explaining how scientific theory can produce a non-evaluative sense of normality, and usually appeal to statistical analysis or evolution. Normativism starts from the perspective that all judgments of abnormality, including disease claims, are made in reference to an ideal or norm, and that the only source for such ideals are social practices and personal values. Basically, all judgments of normality are conventional. Normativists, however, tend to undervalue the knowledge created by successful scientific explanation of natural phenomena, and are mistaken that all claims of normality are essentially the same.

In this section I will offer my own theory of disease—a theory that incorporates some insights of normativism and naturalism. My theory offers a reasonable reconstruction of what physicians and biomedical scientists mean when they use the term disease. It appeals to a realistic conception of how science operates, what it produces, and
the nature of those products. My account makes clear the distinction that is of most interest to policy makers and applied ethicists—those interested in the boundary issues mentioned at the outset—the distinction between legitimate disease claims and illegitimate claims of disease.

Before specifying what I take a legitimate disease claim to be, I want to reiterate the main problem faced by a philosophical examination of the concept of disease, as well as reiterating some key points to recall for understanding my explication. An analysis needs to specify how to distinguish insignificant variations in form and function from significant variation. This will be influenced by the assumptions implicit in the analysis about the nature of science and its products. An analysis must explain how science can create a sense of normal and abnormal that are not primarily evaluative, and do not use as their ideal of reference social and personal judgments about what is normal or proper. As noted, naturalistic theories often utilize a statistical notion of normality. Most normativists basically believe there is only one sense of normality, the conventional sense. My position holds that there are at least three largely distinguishable senses of normality, and that some successful scientific models utilize a sense distinguishable from statistical or conventional normality, namely the theoretical sense.

Some points to keep in mind include that one’s willingness to accept my position depends on one’s willingness to see science as creating accurate and useful models of biological systems. Science does produce new and significant knowledge about the mechanistic functioning of biological systems. This progress is piecemeal and often slow.

---

117 See Chapter 2.
118 While I am trying to provide a definition of disease as currently used by physicians and biomedical researchers, I nonetheless realize that there is some amount of vagueness and imprecision in the concept. By calling my project an explication I admit that what I am doing is also subtly reconstructing the concept. My term may be viewed as an improvement on older less precise historical variants. Would I go further and insist that my explicatum is superior, or that it replace the vaguer explicandum? I see no problem with allowing variant uses of the word in situations that do require precision. However, when boundary issues necessitate knowing legitimate claims about some clinical problem being a disease, more precision is needed. Also, to be precise, what I am providing in the next few pages are the criteria needed to be a legitimate disease claim. However, the explication of ‘disease’ as compared to a syndrome, clinical problem, lifestyle choice, behavioral problem, personality quirk, etc should be easily extrapolated from what I have said throughout this essay.
Larger and larger circumscribed systems are modeled, and the composite models become more and more detailed with regards to the chemical processes involved. One such example is the model of normal and abnormal function and structure of the thyroid. Of note is the hierarchical structure of interrelated models that emerges, and the ability to talk of ‘levels of analysis.’ For instance one can discuss the function and structure of the thyroid as a whole organ, or ascend to a ‘higher’ level and discuss the role of the thyroid in calcium metabolism, or its role in regulating energy metabolism, or even discuss the role of the thyroid in the larger endocrine system. One could ascend to a discussion of the gross anatomy and physical appearance of the normal and abnormally functioning thyroid gland, such as various types of goiter. One could descend ‘levels’ and discuss the structures and functions of the group of cells that produce calcitonin, parathyroid hormone, or thyroid hormone. One could examine the structure and function of individual cells, their receptors and organelles. One could descend to the level of describing enzymes and structural molecules, or genes, or move lower and describe how these entities utilize certain atomic-level chemical properties to accomplish there functions and maintain there structures.

In the process of creating these models and in testing and refining them many conceptions of normal function emerge at many levels of analysis. These conceptions of normal function emerge from the process of trying to understand and accurately describe how the thyroid gland works, and the sense of normality created is neither statistical nor conventional. It refers neither to an analytical sum nor a social norm, but rather back to the developing idealized model itself. These models also explain various types of malfunction or malformation. Possessing models of malfunctions and malformations of a specific biological system warrants legitimate disease claims. The core concept involved in being a disease or pathology is being the malfunction or malformation of a biological system which otherwise could have functioned normally. Without accurate

The distinction between malfunction and malformation is often just a matter of the level of analysis. Lower level issues of malfunction (whether current or prior malfunctions) can lead to higher level issues of malformation. The diseases of collagen metabolism are an example. Similarly, lower level issues of malformation (such as a gene point mutation) can lead to higher level malfunctions (in enzymes or channel proteins).
models of biological systems that describe both normal and abnormal function and structure there cannot be a theoretical sense of normal and abnormal.

Of crucial importance, however, and often overlooked by proponents of the success of scientific model building, is the fact that the models offered at any one time vary regarding their virtues and limitations. They are works in progress, always open to refutation or refinement. And, most troubling for policy makers who want to rely on them to settle boundary disputes, they can greatly vary as to their reliability, their fit with the phenomena being described, or vary regarding the amount of bias or misleading aspects they contain. The need to assess models for such issues becomes particularly acute in newer models or those that operate at higher levels of analysis (i.e. those that involve aspects of grossly observable human form and behavior). Bias and the blending of conventional senses of abnormality with theoretical senses are possible. One must be aware of such blending and potential bias especially when the designation of something as a disease (a biological malfunction) will potentially result in new monetary or social assistance for some group, or the potential discrimination of a group. In such cases the values and biases of our culture, and individual and group preferences, easily mix with theoretical senses of what is and is not normal.

Any scientific model must be individually examined and evaluated. Such ability to evaluate models can be acquired, but even for the non-specialist there are many clues that can more or less be relied on to estimate their relative virtues and problems. Biological and medical models are perspectival. This does not detract from their usefulness and value. They represent some of the most useful, objective, and explanatorily powerful scientific models that humans have ever produced. They are in many senses the best examples that we have as far as empirical knowledge of natural processes. Dismissing science generally or entire fields of inquiry, instead of judging models individually, is a gross error. The normativist position, to the extent that it globally dismisses scientific models as merely social constructions that inform us more about our social values than about natural phenomena, is misguided.

120 Acquiring the background and ability to make such evaluation is part of what makes someone a specialist in a medical or scientific field.
My own position is that a disease claim is legitimate if it is based on and includes reference to:

1. a specification of a circumscribed biological system
2. a specification of the normal structure and normal function for this system (and/or its parts)
3. a specification of a particular malfunction or malformation in the structure or function of the system (and/or its parts)
4. these specifications are part of a scientific model (i.e. generated by those in the community recognizable as the authorities on the details of the biological system in question)
5. the scientific models involved are assumed to be or judged to provide (at least by the appropriate scientific authorities) an accurate unbiased representation of how human biology normally functions and how it can in this particular way malfunction or be malformed.

Several paradigm examples of legitimate disease have been used throughout the essay, such as type 1 diabetes mellitus or Graves’ disease of the thyroid. Several examples of illegitimate disease claims have also been discussed, such as the diseases particular to antebellum slaves proposed by Samuel Cartwright. Illegitimate claims rest on problematic models of biological dysfunction or malformation. The problems are usually issues with poor fit between the model and the phenomena being modeled, and/or unacceptable bias in the model. Even contemporaries of Cartwright could probably see the political and moral agenda behind his proposals. Illegitimate claims usually involve the recognition that conditions 4 and 5 cannot be met.

Putative disease claims are those that do not meet the criteria for legitimacy, but which are not clearly false or so biased as to obviously be veiled value judgments. This may be a large area of disease claims. Potentially there can be problems with any or all of the conditions above. A putative claim could be a hypothesis that a cause exists for a set of symptoms or signs, without clear designation of a circumscribed system. It might lack clear specification of any normal functions. Usually a putative claim proposes some mechanism that is speculative in that it awaits more evidence or better alignment with
less speculative models of biological function. Many ‘syndromes’ in medicine start out as putative disease claims. These are sets or patterns of signs and symptoms that recur or hang together in such a way as to generate suspicion about a common lower level causal mechanism. Putative disease claims assume an underlying biological malfunction even when the details are not yet clear. This presumed common cause (an etiology) is in distinction to an individual sign or symptom, such as red patches and fibers growing out of the skin, or eliciting pain on palpating certain areas. These sign and symptoms might be observed or produced on examination, and might be caused by disease, or be used in order to diagnose a disease, but are not themselves disease, putative or otherwise. They might however be clinical problems in and of themselves, and be reasons to see a physician and be treated.

My account makes it clear that in designating a biological system as abnormal, the normal ideal against which this is contrasted is the scientific model. This is in contrast to Boorse and Engelhardt. For Boorse, the ideal against which abnormality is contrast is a statistical abstraction of the typical goal-directed function of the human part in question. For Engelhardt the ideal being contrast with abnormality is a complex of social and personal conventions.

Before closing this section I will review a few of the resources available on my account for examining controversial or interesting cases regarding disease claims. It is a strength for any account to be able to explain why some disease claims are controversial and whether they are legitimate, putative, or illegitimate. In the final section of this chapter I will deploy the resources of my account and analyze some controversial cases.

One obvious resource is the ability to somewhat untangle the three senses of normality from each other and avoid equivocation in that regard. With these different senses of normality in mind, it becomes easier to determine if what is being claimed is that a biological system or an individual is abnormal. For legitimate disease claims the individual has a malfunctioning biological system. This system is functioning or has been formed in a manner different than the ideal or normal. The ideal (the normal function and structure of that system) is specified in a scientific model, and most likely the details are available in a definitive textbook. The ideal is not necessarily an average or statistical abstraction, nor the prevailing evaluative opinion of the community. Claiming that an
individual is abnormal or handicapped is often a judgment made in reference to social, conventional norms and ideals. Keeping the ideal against which abnormality is judged clear allows us to recognize illegitimate and putative claims more easily.

Another resource for evaluating controversial disease claims is the idea of ‘levels of analysis.’ The example of ascending or descending levels with regard to the thyroid gland was discussed above. The closer one gets to gross observable human anatomy and form, or to human behavior, the harder it becomes to keep the conventional sense of normality out of explanatory models. In the other direction, the closer one gets to physical chemistry and physics, the more prevalent statistical senses of normal become and the less frequently is functional analysis used. The models for the behavior of an ideal gas or for protein folding, if they appeal to any sense of normal at all, appeal to a statistical sense.

My account offers a satisfying distinction between signs and symptoms of the disease at one level of analysis and mechanistic causes at lower levels of analysis. This can help in situations where a biological function may be ambiguous between being abnormal or being just a normal variant. Take hair loss as an example. Hair loss may be part of the causal story of a disease (e.g. polycystic ovarian syndrome) due to a much lower level malfunction. In this case it is a symptom of disease. Considered by itself it may be ambiguous regarding whether it is normal biology or abnormal. Some cases of middle aged balding seem to be just normal variations of development and aging. In some cases hair loss might be difficult to classify as a case of abnormal function in and of itself, but if it is explained by or linked to a lower level abnormality it can be seen as a sign or symptom of disease. These sorts of considerations also explain why some historical disease claims, such as fever, are currently regarded as just signs or symptoms of disease but not diseases in their own right. As science develops more elaborate and sophisticated tools and techniques, as well as a greater stockpile of reliable models, it can delve to increasingly lower levels of analysis. Sometimes what looked like abnormal biological function is now reconceived as a normal response to a lower level abnormality.

As a practical issue of building disease models, the model is felt to be complete if the lowest level of analysis at which there is a biological malfunction is reached, and below that level there are only mechanistic explanations of the normal functionality allowing the
malfunction to be instantiated. Reaching this point might be finding a gene mutation, and below the level of the mutation might be only analysis that explains the normal biochemical phenomena that allow genes to have their form and function. This leads to an interesting issue concerning the theory of human disease: should a disease classification be carried out only at the lowest levels of abnormal function, and are all diseases potentially traceable to just one level of analysis? It might be that all disease is genetic in origin and explanation, or perhaps all disease is ultimately environmental.

My account offers a realistic, albeit simplified, sketch of science and its products. My sketch is pragmatic and discusses the creation of various kinds of models of natural phenomena. The human processes of generating hypothesis and evidence for them, and the community aspects of testing, communicating, verifying, criticizing and refining these models is sketched simplistically. Discussing ‘science’ grossly and making any accurate general observations while doing so is difficult. The point here is that both for illegitimate and legitimate claims of disease, what is determinate are the virtues of the model being referred to, especially the fit between a model and the phenomena it describes and explains. This is also what is missing in putative claims. The basic idea is that the better the scientific model (i.e. regarding these virtues) upon which a disease claim is based, the more legitimate the claim, and the easier it will be to demarcate the kind of boundaries discussed in the opening section (e.g. between therapy and enhancement). Figure 4.1 below represents this crude idea. One could select the quality of the scientific model being used in a disease claim (assuming the virtues of the model could be summed and different models compared on a spectrum). From that point on the top spectrum, a roughly perpendicular line could be drawn down through the other spectra (assuming these qualities could also be arranged on a spectrum).

My analysis explicitly distinguishes and explains putative disease claims. It explains why these are so problematic from a policy standpoint. Putative claims prevent the clear settlement of boundary issues while better evidence or while expert consensus is not available. Until these claims better approximate legitimate claims their use in policy or bioethical debates is more problematic than helpful. However, such putative disease claims, especially when a biotechnology potentially allows a medical intervention, are usually the cases that put pressure on policy makers. Boundary disputes that rest on
putative disease claims remain indeterminate as long as the needed uncontroversial model of theoretical normality vs. abnormality is lacking. Until it’s available the policy decisions and line drawing will have to be made based on negotiation, legislation, or fiat. The process of acquiring uncontroversial new evidence and better models may take a significant amount of time. Decisions in such contexts will often involve appeal to community values and conventional senses of what is normal and acceptable, and to what we as individuals and as communities might prefer. In such circumstances our guard against discrimination and personal agendas should be fully alert. Being aware of the nature of situations involving putative claims is valuable. In this way my account is messier than most naturalistic accounts, but more realistic.

It is possible on my explication to extrapolate an account of how most disease claims evolve from speculative and putative, to those based on less tentative experimental models, to finally evolving into claims based on definitive models of biological systems. My account captures one of the basic idea of disease across historical periods. Even if the explanatory models were of a different nature, a core idea behind human disease is a malfunction of those systems that otherwise operate normally to sustain life and provide ability.

One last point to highlight concerns the definition of health. Health (negatively defined) is the absence of pathology. As science slowly builds up a more complete picture of human biological form and function, and a much more complete catalogue of all possible malfunctions, such a negative assessment of health will be possible. But this is not a positive definition of health. It is not a positive definition of the ideal (i.e. ideally normal) human. For this, Engelhardt may be correct that we need an evaluative theory of human nature—a theory of what the good is for grossly observable human form and function. This may be particularly true if human cognition, behavior and appearance are part of the complete theory of the ideal. But even if there are such requirements for a positive definition of human health, this does not present a problem for most run-of-the-mill disease claims. Disease claims only require is well-respected, verified models of simplified normal and abnormal function of isolated and circumscribed biological systems.
FIGURE 4.1.

ACCURATE

• Well tested/verified
• Entrenched
• Robust
• Accepted by relevant experts
• Has appropriate fit or relation to real phenomena being modeled

HYPOTHESIS

• Highly speculative theories
• Untested/new
• From poorly grounded discipline
• Controversial
• More room for bias to slip in

Science

Theoretical Normal Human Functioning

• Unproblematic notions of normal biological function
• Problematic claims of theoretical or objective normal function

Diseases and Dysfunctions

• Unproblematic claims of disease
• Problematic claims of disease
• Potential for medicalization

Medical Interventions

• Clear distinctions possible between therapy and enhancement
• Difficult, if not impossible, to distinguish therapy from enhancement
4.5 ANALYSIS OF CONTROVERSIAL AND INTERESTING CASES

In various places in this essay, I have discussed examples that are unproblematic and straightforward regarding being legitimate disease claims. Type I diabetes and thyroid adenoma have served as examples. I will not discuss further these sorts of uncontroversial examples. Excellent textbook explanations offer the normal models of function for the pancreas and thyroid glands, and for their parts, and the cells that make up these parts, and the sub-cellular organelles and molecules whose functions combine to allow the cells to operate normally, etc. Medical textbooks offer models of abnormal functions as explanations of particular diseases in these organs. In this section, I shall discuss challenging or interesting cases from the point of view of my philosophical explication of disease.

My goal is to provide a realistic way to distinguish legitimate disease claims from illegitimate. Many of the cases that raise the legitimacy issue, however, are going to be putative disease claims. Many putative claims revolve around a clinical problem (i.e. a problem or issue that someone brings to or wants to bring to a physician or medical specialist). Potential medicalization, as discussed in Chapter 3, is part of the controversy surrounding many of these examples. The grouping of the examples is somewhat arbitrary. Many of them can be approached from different perspectives—that is, they are interesting on multiple dimensions.

4.5.1 Pathological Biological Functions That Are Not Harmful

For normativists disease is culturally relative because the criteria for being a disease, namely certain kinds of perceptions and value judgments, can be different in different communities or cultures. Most normativists hold that a biological condition needs to be perceived as harmful, or inconvenient, or disabling, to be perceived as disease. The examples in this section are challenges to such forms of normativism (i.e. pathology without harm or disability). From my perspective such examples can exist because diseases are biological abnormalities but not necessarily harmful. In fact, on my account certain biological conditions could be considered beneficial or desirable by a community but still be a theoretical biological abnormalities. There can be complex relations between community perceptions and values and the theoretical models that
explain biological function, however there is not a direct or necessary connection between the two—they are separable. For a given biological abnormality, the community’s perception of whether or not it is undesirable, harmful, etc is malleable in ways that scientific theory is not.

Offering examples of such situations is difficult. One problem is that, while there need not be any connection, many biological abnormalities do cause symptoms that are judged harmful. Another problem is the potential to argue over details. These can be either the details about the theoretical models that present some biological process as abnormality or the details about whether the community in question perceives the condition as harmful or inconvenient. Because of these problems science fiction examples are often used so that such details can be controlled. However, this does not entirely solve the problem because the details of what constitutes normal biological function and what does not are not easily imagined—they are the result of empirical investigation. The verified, accurate models that explain normal and abnormal function do so in ways that are often unpredictable and sometimes counter-intuitive. When a scientific model specifies a biological abnormality but the community does not perceive the effects of this abnormality as negative the question is: Is this a disease?

Diseases not found harmful raise many interesting practical issues. The most interesting scenario would be within a community where there was divergence regarding the communities judgment of the value of the condition or symptoms. If some in the community found the condition harmful and undesirable while others thought just the opposite, would we consider treating patients with the condition? Would we treat only the ones who found it undesirable? Would we try to prevent people who desired the condition from getting the condition? Might physicians even be called on to help some people acquire this disease? The possibilities and issues raised by certain science-fiction scenarios of ‘beneficial diseases’ are numerous.

---

121 Part of the difficulty here is the fact that in the early days of hypothesizing symptoms that are judged harmful and undesirable are the ones we search for explanations of their being abnormalities in biological function.
4.5.1.1 Genius as Abnormality

Consider the case of a genetic mutation that causes one to be perceived as a genius by our current community. This is, to my knowledge, a science-fiction example, but potentially similar, real-world examples will be considered below. Is this a disease? The problems mentioned above make the case difficult to analyze. There are not currently enough accurate models explaining normal and abnormal cognitive function of circumscribed neurobiological systems. Furthermore, we have no uncontroversial notion of human intelligence, nor a clear notion of what makes for genius. Statistical attempts to measure intelligence are inadequate for reasons similar to those I have claimed make statistical measures inadequate for determining biological normality.\footnote{The statistical measures alone cannot tell us what is normal variation from significant variation.} The details of the biological (or psychological) malfunction that allowed enhanced cognitive abilities would be important. To be a disease claim, the biological or psychological models developed of those systems implicated in causing the symptoms, would have to specify how the system was malfunctioning—how it was deviating from normal function. Importantly, it would also have to make clear how this was not just a variant of normal function. If such models existed, described a biological malfunction giving rise to desirable cognitive abilities, then, I would have to bite the bullet and admit this was a case of a desirable disease. The abnormalities would also need not give rise to any undesirable symptoms. Again, to the best of my knowledge, there are not currently any cases like this.

Compare this to the situation of certain cases of severe mental retardation. The cases of Down’s syndrome and Fragile X syndrome will serve as examples. In these cases we do have increasingly good models of abnormal biological function and structure leading to changes at the cellular and metabolic level. These changes purportedly lead to the observable findings of the syndrome including the particular intellectual disabilities. However, there are gaps in the explanation, potentially as one gets to higher levels of analysis. The signs and symptoms of these diseases were observed together and constituted a syndrome for which explanatory models are now being built and refined.

For Fragile X syndrome, certain features are caused by problems in elastin
production, including the findings of prominent ears, hyperextensible finger joints, soft skin and flat feet. Related physical findings can include pectus excavatum, mitral valve prolapse, strabismus, an unusually long face, prominent chin and enlargement of testicular size. A significant number, but not all, children with Fragile X show behavioral symptoms similar to autism, including stereotyped movements, and decreased interest in social interaction. These affected children show developmental delay and an IQ in the range of mental retardation. Mathematical skills, executive function, visual memory, visuospatial-processing abilities, visual-motor coordination, auditory short-term memory, processing of sequential information, sustained attention, and working memory are all areas that can typically be affected. The theoretical model of Fragile X describes a region on the X chromosome composed of repeating trinucleotide sequences which as it expands in size (to somewhere between 30 to 200 copies), eventually causes a loss in the expression of mRNA and thus the failure of synthesize a protein. This protein, FRMP (fragile X mental retardation protein), normally has a multitude of regulatory effects on the expression of other genes whose products are important in synaptic signaling and the establishment of synaptic structure and on other metabolic functions. Malfunction due to loss of this and other proteins and nucleic acids causes the syndrome. More detail and an excellent pictorial summary of part of the model of the normal cellular signaling mechanisms dependent on FRMP is available in a review article by Chonchaiya (2009).

Down’s syndrome relies on a similar putative model of normal functions and of the abnormal dysfunction that gives rise to the clinical findings, including the various features of lower intellectual abilities. I would judge these both to be legitimate disease claims. At worst they are putative disease claims that are extremely close to meeting criteria 5, namely that the models of normal and abnormal function are close to being taken as being accurate, having good fit, and are unbiased. If anything is missing, it is some of the details about how lower level defects in proteins and metabolism cause defects at the cellular level that eventually sum to create the cognitive deficits. Our science fiction example of a genius mutation, in order to be a legitimate disease would require such accurate models to back the claim. It would also need to have only desirable consequences.

Other science fiction examples might be explored, such as a genetic mutation that
makes one less sensitive to pain (but not so much that frequent injury becomes a problem), or mutations that make one age slower. These would be legitimate disease claims if respectable scientific model existed to explain abnormality against a backdrop of otherwise normal function. If the models specified these things as abnormal biological function, then regardless of whether or not the community found the resultant effects to be beneficial, they would be legitimate disease claims. Again, there would be room to argue about whether these were really biologically innocuous conditions, regardless of the perception of the public. The issue faced by researchers would be determining normal variation in function from abnormal function. On my account the process of creating, testing and refining the model would ultimately produce these distinctions. The normativist would challenge that it is the conventional judgment of the community, or of the researchers involved, that determines if these variations in biological function are normal or abnormal. My position concedes that initially conventional values can and do enter into hypothesis generation, but this is not the end of the story—scientific research and model building engage with the phenomena and increase the epistemic virtues of a model. Researchers seek to reduce the bias in the model. Many subtle issues go into the determination of whether the activity and the variations in activity of some circumscribed microscopic biological system are considered abnormal or normal. Even if initially influenced by statistical and conventional senses of normal, as the model develops and gains ‘fit,’ it develops a different sense of normality and abnormality. The sense of abnormality generated in the models for fragile X syndrome is not the same as the sense used to judge the ears or testes of those perceived as abnormal. For genius to be a symptom of disease some biological model would have to specify the abnormality that gives rise to what we judge as genius. In this case my account would make it a disease. Boorse has the option of falling back on ‘natural design’ as a distinction between normal function and abnormally beneficial functionality. He defines disease only for functional efficiency below a cut-off. Extremes of functionality are unnatural for him, but not necessarily disease.

See Cox, et. al., 2006 and Cox, et. al., 2010 for case reports of a rare genetic mutation that does cause congenital inability to feel pain. However, as predicted, affected children seem to get injured frequently.
Many intellectually sophisticated fiction genres have considered scenarios of superpowers as disease. Often the superpowered become either heroes or villains and usually must keep their abilities anonymous and their identities hidden due to the possibility of discrimination and persecution due to their being different. Perhaps in a future where such things are possible (perhaps made possible by bioengineering) we might need a new term besides ‘disease’ to describe biological abnormalities granting superhuman ability. Our scientific models will likely become complex enough to make these new distinctions, but until then, biological abnormality satisfactorily modeled and explained is legitimate disease regardless of whether or not we perceive the effects as harmful, beneficial, or inconsequential.

4.5.1.2 Asperger’s Syndrome

This may be an actual example of increased intellectual abilities as part of a pathology. Autism is usually understood to manifest along a spectrum of clinical severity with regard to the symptoms of cognitive and linguistic impairment and developmental delay. At the less severely effected end of this spectrum is Asperger’s syndrome. Diagnosis is made in the face of symptoms such as impaired social interaction skills, repetitive stereotyped movements, and a restricted pattern of interests, while lacking the typical language and developmental delays of more severe autism syndromes. Intense pre-occupation with certain narrow subjects or abilities, and physical clumsiness are typical. Technically, Asperger’s is a pathology but to make the diagnosis in the United States in accord with the latest DSM-IV-TR, the patient must experience significant impairment in day-to-day functioning.\textsuperscript{124} So the case is not one that neatly separates perceived undesirability from theoretical abnormality. Furthermore the biological models behind autism are less well worked out than those for Fragile X syndrome. Autism and the autism spectrum diagnoses are putative diseases, awaiting better mechanistic models. The spectrum of severity may breakdown into importantly different disease entities distinguished by different causal mechanisms but similar observable symptoms.

However, assume that further along the spectrum of autism there are people with

\textsuperscript{124} Interestingly, this requirement of interference with daily life, is a frequent part of psychiatry diagnosis. More on this below in the subsection on psychiatric disease.
poor but not debilitating social skills and intense interest and talent regarding certain kinds of applied mathematics, such as engineering, economics, and accountancy. One study found that in a sample of 919 families with a child with autism, 28.4% had either a father or grandfather who was an engineer, vs. only 15% of control group families (Baron-Cohen, et. al. 1997). Perhaps these engineers are very high functioning autistics and have special abilities without significant loss of social skills. The crucial issue is whether we can imagine a disease or pathology that is not undesirable. This example is problematic because it takes a case of what intuitively is clearly a disease and waters it down until what remains is minimal harm and the prospect of benefit. On my account some of these engineers, accountants, or mathematicians may have a putative disease, and one that helps them with their profession. I have also heard similar speculation about a spectrum of psychopathy (i.e. an abnormality resulting in lack of normal empathy). Perhaps there are people lacking empathy such that they thrive at certain professions but not so much as to produce trouble with the law or with socializing. There can, in my opinion, be diseases (i.e. theoretical biological or psychological abnormalities) that are not necessarily harmful, even if the actual cases of this are rare and difficult to find. Asperger’s syndrome is a putative disease claim, but a problematic one, due to the existence of mostly hypothetical and still evolving models of normal and abnormal function of a biological or psychological system involved with the observable symptoms. Criteria 1 and 2 on my account of legitimate disease claims are not met. Such models of psychiatric disease face special difficulties as we will discuss below.

4.5.1.3 Paganini

It is hypothesized that the virtuoso violinist Paganini suffered from a connective tissue disorder, perhaps Marfan’s syndrome (Schoenfeld, 1978) or Ehlers-Danlos syndrome. This example is very much like the Asperger’s example, except that we have a physiological disease with more respectable and complete biological models. Again, science fiction examples could be generated off of this theme, and one could imagine athletes or contortionists having significantly increased ability due to connective tissue mutations. Presumably any potential harms, like weaker artery walls in Marfan’s would not be manifest in these science fiction examples. On my position, Marfan’s syndrome is
a legitimate disease claim and some of the science fiction examples might also be legitimate diseases, even if they produced no harms and only benefits for those affected. An interesting wrinkle would be such diseases that affected people to different degrees along a spectrum. Then we would have a physiological disease that in some cases caused what was perceived as harm and in some cases caused what was perceived to be benefit. Regardless, if our best biological models of the systems involved describe the effects as caused by a biological malfunction, then all the effects or symptoms would be legitimate disease claims.

4.5.1.4 Immunity to HIV-1 Infection from Mutated CCR5 Allele

Perhaps the best real-world example involves a genetic variation that may give substantial immunity to infection by HIV (the virus that causes AIDS). The genetic variation is in a chemokine receptor (CCR5) on white blood cells. These receptors are used by the HIV-1 virus to gain entry to these cells so that the virus can highjack the cells DNA and RNA transcription functions to reproduce itself. Certain mutations (delta 32) when present in all the copies of the genome (making the person a homozygote regarding that allele) prevent certain prevalent strains of HIV-1 from entering the cells (Samson, 1996). There has been a case of a patient with HIV-1 infection treated with a bone marrow transplant as therapy for development of myeloid leukemia. The transplant was with cells from a person homozygous for the delta 32 CCR5 genes, and thus after transplant, the new white blood cells of the HIV patient, now expressed only the mutated CCR5 chemokine receptors. This patient then achieved undetectable levels of the virus for 20 months—essentially the infection was controlled and possibly cured (Hutter, 2009).

So is this a case of acquiring a disease that has a benefit—acquiring one disease that alleviates another, much worse disease? Is this an example of the kind of disease many of us may not mind having? Let us assume that the variant CCR5 protein has no other biological effects. The normativist position would be that the mutation is not a disease because it causes no harm, and in fact, in certain circumstances is a benefit. My position would be that the scientific model will (at least eventually) decide whether this gene allele is the cause of a disease, namely causes a biological malfunction, or is just a
normal variant, like variations in blood type. On my position regardless of the perceived benefits or harms, if there are respectable biological models describing the causal effects of the allele as malfunction it is part of a disease explanation. So far, at least, having the allele (even being homozygous for it) is considered having a normal variant receptor.

The crucial issues here, for a theory of disease, are the distinction between normal variation and significant, pathological variation, and how this distinction is achieved. Is it conventional judgments of what is normal or abnormal? My position is that to be a disease there must be a theoretical model specifying that a variation in function or structure is not merely a normal variant, but an abnormal structure or function. Statistical data are often important elements in such models, but typicality or rarity in themselves cannot settle the issue. The function and structure of cell receptors and the role they play in larger biological functions is not the type of issue about which conventional notions of normal and abnormal are formed. In the current example, the CCR5 cytokine is one of many different proteins on the surface of cells that instantiate the normal immune functions of the body. The possession of a mutant CCR5 genotype is a variant of normal and not a disease. Among its many functions, its expression allows T helper lymphocytes to migrate to the correct area when an immune response is being mounted. Certain groups of chemokines expressed on the endothelia that line blood cells in these areas of localized immune response adhere to CCR5 and other molecules on the surface of the T lymphocytes and help facilitate migration from the blood to the area in which they are needed (Janeway, et. al, 2007). There appears to be some redundancy in the immune system so that complete loss of the CCR5 receptor as in the homozygotes in this example does not lead to any detectable malfunction.\textsuperscript{125} There is also interesting speculation as to what selective pressures are responsible for the increased prevalence of this genetic allele in white northern-Europeans. Some have speculated that immunity from other bacteria and viruses may have been part of the selective pressure.\textsuperscript{126}

\textsuperscript{125} It may be that the allele causes greater susceptibility to the West Nile virus. See Glass, et. al., 2006.  
\textsuperscript{126} CCR5 mutations may have been selected for as part of increased immunity from \textit{Yersinia pestis} and/or the smallpox virus. \textit{Yersinia pestis} is the bacteria that caused the Black Plague in Europe (see Galvani, 2005).
4.5.1.5 Sickle Cell Trait and Cystic Fibrosis Carriers as Examples of Heterozygote Advantage

Sickle cell anemia is a disease caused by the inheritance of mutated alleles, specifically alleles that encode for the amino acid valine instead of glutamic acid at the 6th position of beta globin. The beta globin protein is a component of hemoglobin which functions to carry oxygen, effectively picking it up in the lungs and releasing it in capillaries near oxygen requiring tissues. The mutated hemoglobin that results in sickle cell disease is prone to form gelatinous polymers in low oxygen conditions, and these polymers cause distortion of the roughly torus shaped red blood cells. These distorted cells can become stuck in smaller capillaries and cause infarction of these vessels, leading to the familiar clinical sequelae (Hauser, 2005). People who carry only one mutated allele are said to have sickle cell trait, and only rarely have some mild complications at very high altitudes or after extreme exertion or in other oxygen deprived states. The frequency of these alleles is quite high in certain populations, and in particular where malaria is endemic. It is theorized that being a heterozygote with this mutation (i.e. having sickle cell trait) offers some survival advantage in an environment were malaria frequently kills. Similar hypotheses have been made about a survival advantage explaining the high rate of certain alleles responsible for cystic fibrosis. These include survival advantages in environments were cholera or tuberculosis infections frequently killed (Alfonso-Sánchez, et al., 2010). Sickle cell disease, sickle trait, cystic fibrosis, and being a carrier of cystic fibrosis, are all legitimate disease claims on my view, even in the cases where some carriers have no perceived harmful effects, and may even have an advantage avoiding malaria or tuberculosis infections.

4.5.1.6 Dyschornic Spirochaetosis

This is a disease caused by the bacteria Treponema carateum, a relative of those bacteria that cause syphilis and yaws. This sub-species of bacteria is limited to the Americas. Infection of a human occurs by skin-to-skin contact and unlike syphilis, skin is the only organ affected. Like syphilis the manifestations of the disease occur in stages, with the initial infection being an isolated cutaneous lesion, but progressing to diffuse lesions which become both hyper- and hypopigmented. The skin of the affected patient
can become mottled with variously pigmented lesions, including shades of blue, brown, red, purple and white (Hook, 2005). The disease is also called Pinta for painted or colored spot in Spanish. The infective agent was determined in 1938 when the bacteria was isolated from a Cuban patient (Editorial in JAMA, 1944). This example is sometimes sighted in the literature on disease claims due to the fact anthropologists have observed a tribe in the remote Amazon for whom the disease was endemic and who believed that the resulting skin lesions were (conventionally) normal. Furthermore, this tribe believed that those who did not have the lesions were diseased, and even forbid young women from marrying men lacking such ‘normal’ skin pigmentation (Dubos, 1965).

Pinta clearly is a biological abnormality and legitimate disease claim on my account. However, the condition is considered to be normal and desirable by some cultures and undesirable and abnormal on other perspectives. Interestingly even from a Western cultural viewpoint the disease might be perceived as having consequences no more serious than a sort of natural tattooing or body art. For the normativist this is an excellent example of the relativity of the perception of disease. It is important to separate the issues of theoretical biological abnormality from any conventional judgments about normality and abnormality. Regardless of perception, the cause of the condition is a biological abnormality. Similarly, it would not be any less of a theoretical pathology (in reference to our best models of intestinal parasitosis) to be infected with a large tapeworm, even in a society that valued and pursued this condition for its weight loss and allergy alleviating benefits.

The creation of increasingly detailed and accurate models of biological phenomena simultaneously create the theoretical sense of biological abnormality, and this sense, once the models are well worked out and verified, often has little to do with how the pathology might be judged beneficial or harmful. The issues involved with accurate theoretical explanation of what causes biological abnormality are not issues of desirability or harm.

---

127 Being infected by certain parasites can cause weight-loss and can modulate the immune system such that certain allergies no longer manifest. Although, malnutrition and other undesirable effects may manifest.
4.5.2 Harmful or Disvalued Conditions That Are Not Pathological

Now consider painful or otherwise generally undesirable conditions that are not theoretical abnormalities. Just as some states of having a disease may not be harmful, some normal biological functions and activities are harmful, painful and anything but desirable. The normativist explains these conditions as somehow not meeting our expectations of disease or abnormality regardless of being harmful or disvalued biological conditions. For the naturalist these examples show the lack of necessary connection between being an inconvenience or harm and being a disease. The most obvious example of this is pregnancy. One of the more challenging is the biological process of inflammation. But here too, the more one looks at interesting cases the more one realizes that at the edges the distinction between what is normal variation and what pathology is sometimes difficult to establish. On my account it is best to allow carefully refined but fallible theory to determine such differences. Biological theory can become as detailed, complex and nuanced as needed. An area that requires such nuance will be certain biological processes that may be pathological conditions in some contexts but not in others.

4.5.2.1 Pregnancy, PMS and Fertility

Pregnancy is described in modern medical theory as a normal biological process and there are models that detail its normal stages and development, and the processes that produce these. The result of many of these processes is inconvenience for the mother. The state is one of relative immune compromise, and has potentially harmful complications. It is necessary to clearly distinguish the details of normal pregnancy from the potential complications, which are biological abnormalities and legitimate disease claims. Such complications could include pre-eclampsia, failure of labor to progress normally, placental abruption, traumatic tearing of the vaginal wall and perineum, etc.

PMS (pre-menstrual syndrome) is another putative disease claim. The example is interesting because it is a case of distinguishing normal variations in biological processes from extremes. The putative theoretical model of PMS as a disease holds that it is an endocrinological disregulation along with significantly increased inflammation that leads
to the physical and mental symptoms posited to compose the syndrome. The hormonal system that controls the menstrual cycle, and the effects of such surges and dips in hormone levels on cognitive and metabolic functions, is very complex and still not well understood. The question is how to distinguish abnormalities from the normal effects of the normal changes in hormones as the body goes through the normal processes of ovulation and menses. The solution to the question about whether this is a legitimate disease claim will have to await much better scientific models, including those that specify the normal interaction of the endocrinological system with the purportedly effected neural and psychological systems. Of note, the current DSM-IV-TR has a putative disease entity called premenstrual dysmorphic disorder that is a variety of mood disorder that occurs consistently in the 2 weeks before menses. The diagnostic criteria highlight the mental symptoms and the fact that the condition is a severe interference with social or professional functioning (APA, 2000). The practical solution in this case for demarcating a clinical problem versus a problem not extreme enough to treat is to include the requirement of disruption of daily functioning. This is a subjective criterion that allows patients and physicians to negotiate whether or not to utilize biotechnology to address problems without needing a full explanation of causation by a biological abnormal process.

Fertility is another normal biological function. However, under certain circumstances it might be perceived as a harm or benefit. People may seek treatment to restore it or enhance it when deficient, or to prevent or decrease it (vasectomy and tubal ligation, oral contraceptives, etc.). On my account, fertility would be an illegitimate disease claim it itself, but infertility may be a sign or symptom of a biological abnormality. Interestingly, on my account the reversal of a vasectomy may be the treatment of a disease—the reversal of an iatrogenic pathological trauma. I shall discuss this further in the section on trauma and environmental causes of disease.

4.5.2.2 Apoptosis, Inflammation, Septic Shock, Autoimmunity, and Allergy

These examples are extremely challenging as they mostly represent normal processes that can be harmful, even deadly. On my account they are not legitimate disease claims in and of themselves. But, as we shall see, these normal processes may be
Apoptosis refers to a complex but well modeled set of pathways for the programmed and orderly death of a nucleated cell. The importance and biological normality of such processes is stressed in the following passage from *The Molecular Basis of Cancer*:

Cell death is one of the fastest growing fields in cancer research. It is now well recognized that a fundamental characteristic of multicellular organisms is that some cells must die for proper development to occur and to maintain homeostasis and health. This propensity to die for the greater good of the organism has evolved so that cells are systematically dismantled through a hard-wired response termed ‘programmed cell death’ (PCD). The number of cells in an organism is tightly controlled by an exquisite balance between proper cell proliferation, differentiation, and cell death. Indeed, in mammals billions of epithelial and blood cells die every day. On the surface, the enormity of cell death in multicellular organisms seems incredibly wasteful, yet these processes play essential roles in maintaining the homeostasis that ensures that individual tissues maintain their correct size and proper function. All eukaryotic cells can undergo the cell death response, which can be triggered by internal or external stimuli. Important examples of this phenomenon are seen in vertebrate development during the sculpting of fingers where the cells between digits are cleared through cell death and in the selective removal of autoreactive lymphocytes. Similarly, cell death plays an important role in regulating blood cell numbers. Blood cell progenitors are continuously made in excess in the bone marrow, yet these progenitors, and their progeny are cleared by cell death, which prevents overproduction and disease states such as leukemia, lymphoma, and/or lympho-, myelo-, or erythrocytosis. In the case of erythrocytes, this excess again seems incredibly inefficient, yet plays an important role in keeping the organism prepared for times of hypoxia induced by rapid blood loss due to injury or following exposure to agents that provoke anemia. Here erythrocyte progenitors can be quickly rescued from the cell death program by increases in the hormone erythropoietin, which inhibits cell death and promotes the differentiation of these progenitors into erythrocytes. These examples underscore the importance of balancing cell proliferation, differentiation, and cell death. Indeed, when this balance goes away, disease ensues. (Dorsey, et. al., 2008, p. 205).

Apoptosis is the orderly and programmed, energy-dependent, destruction and death of a cell via specifically activated genes and the resultant encoded proteins. Death by apoptosis prevents the inflammation that would occur from less orderly other forms of cell demise. In apoptosis the cell implodes and neatly packages the waste materials for easy, orderly disposal by the body. The models of the molecular pathways involved
describe the normal process and also highlight various ways in which the process may malfunction. Some types of malfunction in apoptosis play a role in the causal mechanism and models of specific kinds of cancer.

The discovery and characterization of these biological functions could have been guided by the perception of early investigators that they were good, bad or indifferent. Initially the process was named when cells undergoing the process were perceived to be shrinking and wasting away like leaves off a tree in fall. The point is that as the models were built up and the processes were understood in relation to other biological functions, apoptosis was increasingly understood as a necessary and normal process. Even if initially perceived as harmful—it is basically mechanized suicide—it is ultimately determined to be theoretically normal. Theoretical normality and being perceived as beneficial are not necessarily coordinated. The loss of this ability to self-destruct at appropriate times is an important kind of abnormality. It is difficult to imagine how conventional value judgments could have determined that eventually apoptosis would be considered normal.

Aging is perceived as harmful. Aging may also be considered as a type of self-destruct mechanism. Some research supports the hypothesis that mammalian cells are programmed to have a certain lifespan and then to cease to reproduce, causing the breakdown and eventual ceasing of function of tissues. In contrast cancer cells that lose some of the normal biological regulatory processes via genetic mutation can theoretically reproduce and survive as immortal cell lines. Perhaps the requirements of building complex multicellular organism with the ability to grow, heal and accomplish very complex higher level functions, are always balanced by the need to reduce cancer-causing mutations during cell division and in the face of environmental damage. The resulting compromise of this balance is aging. Aging, on such a picture, would be a normal biological process and there could be various malfunctions in the systems that regulate normal aging. But the reliable and detailed models explaining these processes are still works in progress. On my view it might someday be the case that aging and death are biologically normal and not associated with disease, in contrast to the normativist view in which aging and death are, in themselves, diseases. Perhaps on my account there would be diseases (i.e. biological abnormalities) that cause longer life. It may, however,
always remain difficult to separate the conventional value judgments about death from any theoretical sense of its normality.

Inflammation is another interesting biological mechanism. It is harmful and often the most proximal explanation of pain and discomfort in many clinical situations. It was once considered as disease in its own right, but is now seen as a general mechanism involved in many immune responses to infection, cancer, or other triggering stimuli. Inflammation is a normal biological process, necessary to remove toxins, necrotic tissue, or invading pathogens. Like apoptosis, abnormalities in the pathway of the normal inflammatory response lead to specific diseases. It is also known that inflammation is a necessary stimulation for the healing of damaged tissues, and blocking inflammation will delay or prevent the initiation of the general pathways for healing at the cellular and molecular levels. In the normal functioning of the mammalian body inflammation is necessary but must be very carefully and complexly regulated (see Kumar, 2004).

Inflammation is a part of the causal mechanism that instantiates many diseases. It is an example of normal biological processes occurring in the wrong context or occurring as the normal response to signaling initiated by a biological malfunction. It is worth noting that in many cases of disease a malfunction at a lower level of analysis will initiate otherwise normal processes to ensue at higher levels of analysis. Inflammation is involved in autoimmune diseases and is part of the mechanism creating the clinical problem of shock.

Septic shock is an important clinical problem. It is the leading cause of death in intensive care units in the United States, and has an overall mortality of 28.6% (Angus, et. al., 2001). Shock is a state of global hypoperfusion—a failure to adequately perfuse all the tissues and organs of the body with blood. It can have several causes, including broad categories like cardiogenic shock (pump failure), hypovolemic shock (caused by acute loss of blood volume), and distributive shock (caused by inappropriate activity of the vasculature causing low resistance and pressure). Operationally it is defined as a low systolic blood pressure, despite at least one hour of adequate IV fluid resuscitation, that continues to be less than 90 mm of mercury (or a mean arterial pressure less than 60 mm of mercury).

Sepsis means literally a rotting of tissue. When applied to a wound it refers to a
pus producing infection. When applied to an individual, as in septic shock, it refers to an infection that has spread to the blood. The patient who is ‘septic’ is one who presents with a set of signs consistent with a systemic infection causing a response. In this syndrome, even tissues remote from the original source can display the signs of inflammation, including dilation and increased permeability of the vasculature. The diagnosis of sepsis (by definition) requires two components: evidence of this systemic inflammatory response and a known source of infection. The systemic inflammatory response syndrome (SIRS) is a host response to infection (although it can be caused by non-infectious causes like acute pancreatitis, and not all cases of localized infections elicit a SIRS). Models of SIRS provide details of the types of molecular signals (e.g. certain pieces of bacterial cell wall) that elicit the response and how the cells of the immune system mobilize specific receptors and initiate chemical cascades to signal the various other cellular and tissue components that instantiate the response. Operationally SIRS is defined as the presence of at least 2 of the following 4 conditions: 1) temperature greater than 38 C or less than 36 C; 2) heart rate greater than 90 beats per minute; 3) respiratory rate greater than 30 breaths per minute; and 4) a white blood cell count greater than 12 million per liter, or less than 4 million per liter.

Thus septic shock is a strong systemic inflammatory response caused by an identifiable infection that is severe enough to lead to hypoperfusion of vital organs even in the face of adequate attempts to boost blood pressure with IV fluids. Septic shock is a type of distributive shock—the main malfunction is that arteries are failing to properly maintain pressure so that normal perfusion occurs. A classic hypothetical model of the general malfunction involved in distributive shock is presented by Landry and Oliver (2001). Although inflammation is a normal host response, many current models of robust systemic inflammation and septic shock hypothesize that it is an abnormal response, with dysregulation of the normal control of inflammation creating a chain of events that leads to shock. Research has investigated different genetic variations that allow differential levels of response, and the details are being worked out concerning which differences are normal variation versus variations significant enough to be malfunctions (see Kumar, 2005).

It is important to note that SIRS and shock are clinical problems diagnosable
based on vital signs and the sorts of rapid tests (CBC, chest X-ray, etc.) that might be done at the bedside in a typical emergency medicine department. They are not in themselves disease claims, but clinical conditions caused by one of several possible disease processes. Septic shock moves closer to being the diagnosis of a disease as it requires that the unstable condition of hypoperfusion be caused by an infection, but it is still just a general clinical syndrome (i.e. set of signs and symptoms) that are presumably caused by an underlying disease process. The issue of abnormality here is difficult. 

Distributive shock may itself be a type of biological malfunction. It may be that certain genetic predispositions allow a hyper-active immune response compared to a more normal response. There are models of distributive shock that hypothesize that it is a condition of global over-response by the immune system and interventions aimed at damping down the immune response have been developed. There are also models that suggest that a crucial part of the abnormality in septic shock is an exaggerated response by the body aimed at down-regulating the immune system once it is strongly stimulated and globally activated. These later models would explain the extremely high mortality of a malfunction that turns off the immune system in the setting of a serious infection.

Determining what is normal and abnormal about the body’s response to certain kinds of serious infection is an area still being worked out. Sepsis and septic shock are clinical problems and syndromes, and some research projects posit them as putative diseases. It is important to remember that pathology at one level can give rise to normal processes at higher levels. Such normal processes, like inflammation, might be the most proximate cause of the clinical signs and symptoms that are harmful. This is another example of the recurring problem of distinguishing what constitutes extremes or otherwise significant variation. As I have repeatedly stated, the best way to settle this issue is to allow scientific research and models to increasingly develop theoretical senses of what is normal variation and what pathology. Biomedical research does this and will continue to do so. These examples also reiterate the lack of necessary coordination between a biological process being perceived as undesirable and what biological science describes as abnormal.

Another interesting area is diseases of autoimmunity. Generally, these are a group of diseases that involve the immune system causing inflammation, destruction, or other
types of dysregulation of the normal tissues of the body. The field of immunology continues to work on a theory of the normal operation of the immune system, and part of this theory includes models of how the immune system recognizes ‘self’ from ‘other’ (Janeway, et. al., 2001). Some autoimmune diseases are failures of the normal process of determining what is considered ‘self.’ It also contains models detailing how the immune system has checks and balances to ensure that it does not get out of control. Such models describe the circumstances when the system is appropriately switched on and when off, as well as the initiation of the process of repair and regeneration. Autoimmune diseases are cases of otherwise normal processes and mechanisms occurring at abnormal sites, or due to abnormal regulation.

Many of the paradigm examples of legitimate disease claims in this essay have involved autoimmune aspects in their models. Type 1 diabetes and Grave’s disease are examples. As I have argued, models may, as needed, become very complicated as far as distinguishing theoretical senses of normal and abnormal. Consider graft-versus-host disease that occurs in the setting of certain organ transplants. Basically the white blood cells of the organ donor are, by a normal process, ‘educated’ in the donor’s body to recognize that donor body as ‘self.’ The white blood cells of the organ recipient are normally ‘educated’ in the recipient’s body to recognize its tissues as ‘self.’ When a liver or bone marrow from the donor is transplanted and contains donor lymphocytes, these cells recognize the new recipient tissues as foreign and via normal immune processes can elicit inflammation and destruction of normal recipient tissues. Is such graft-vs.-host disease a legitimate disease claim? It is caused entirely by normal biological processes active in a certain specific context. The therapy is aimed at preventing normal immune responses. Perhaps some would argue that it is a type of infection—invasion by a type of cell that causes tissue damage. Consider also the various kinds of immune mediated rejection of transplanted organs by the normal behavior of the recipient’s immune system. Is it a disease to reject these transplanted organs? On my account, I would argue these processes are all normal. It is human desires and agendas that perceive such rejections or immune responses as unacceptable or abnormal. Organ transplant is a

---

128 This process involves the appropriate (and normal) death of certain immune system mediating white blood cells, and the survival of others.
therapy for legitimate disease claims, but the normal biological processes that make it challenging are not in themselves causing disease.

4.5.2.3 Mountain Sickness and Decompression Sickness

Continuing on the theme of undesirable states that may not be diseases, consider the conditions of mountain sickness and decompression sickness. These problematic conditions occur when humans move from one set of environmental conditions to another. In mountain sickness, humans acclimated to living in more typical elevations nearer sea-level ascend too rapidly to mountainous elevations. Decompression sickness, also sometimes called ‘the bends’,\(^{129}\) occurs when humans place themselves in conditions of high ambient pressure and move too quickly to a lower ambient pressure. Decompression sickness is a risk for those working in deep sea and space exploration. The challenge for disease claims is that these might just be normal physiological responses to unusual or extreme environmental factors. A simpler version of the basic problem would be a person suffocating when moving from a well-oxygenated environment to one without oxygen, such as an astronaut being exposed to space without any life support apparatus. Would such a death be due to a disease—to a biological abnormality? The same issue seems to occur for drowning. These cases highlight that what is considered theoretically biologically normal may be relative to simplifications and idealizations of the environment in which the models are assumed to operate. The models of normal biological function assume environments with certain atmospheric pressures and gas contents. If there is a spectrum of responses as we subtly change environmental aspects, how do we determine where normal physiological responses end and abnormal or pathological responses begin? How similar are some of these situations to trauma from hostile elements of the environment—such as being burned by the sun?\(^{130}\) How are some of these similar or different from being poisoned by the bite of a venomous snake or by the treachery of an angry spouse? Perhaps they are importantly

\(^{129}\) It is also called caisson’s disease, and all these different names are considered alternate designations for the same disease because the model of underlying biological process involved in each case are the same.

\(^{130}\) Getting a little red is a natural reaction, although still trauma. A blistering, cracking, bleeding sun burn is severe trauma.
similar to cases where the environment fails to provide the necessary nutrients for certain biological functions—such as cases of endemic goiter and cretinism due to iodine deficiency.

As far as whether mountain sickness is a legitimate disease claim, I again allow the models of the biological phenomena involved to distinguish normal from abnormal processes. Acute high altitude illness is a clinical syndrome that affects some people at high altitudes (esp. those higher than 8000 ft.). The ‘sickness’ is caused by the thinning of air—less molecules per square feet, measured as a drop in barometric pressure. The partial pressure of inspired oxygen drops and thus the driving force behind oxygen movement in and out of the blood in the lungs is decreased, and tissue hypoxia can result. Symptoms such as headache, fatigue, and malaise can develop. The symptoms are caused by relative tissue hypoxia and from resulting changes in the vasculature. Certain acclimatization processes begin to occur and take minutes to days to adjust. Potentially fatal complications may rarely result due to pulmonary or cerebral edema, most likely caused by extreme forms of vascular permeability (Allemann and Scherrer, 2010). There are clearly normal variations in the efficiency, speed and reserve capacity of the acclimatization functions. The models of the biological systems involved do designate the more extreme effects, such as pulmonary or cerebral edema, as putative disease claims. These rare complications are malfunctions compared to the normal functioning of these systems, even when these systems are stressed by ascent to higher altitude. The more common less extreme symptoms due to overly rapid ascent do not appear to be legitimate disease claims but variations in normal function.

Decompression sickness basically occurs when gasses in the body that were dissolved into solution at one pressure ‘boil’ out of solution as pressure changes. This happens when lower ambient pressures are entered and when the normal pulmonary and circulatory processes that deal with such out-gassing are overwhelmed. Here the models involved designate normal processes that are elicited merely by humans moving between extreme environmental conditions. I believe that decompression illness is not a legitimate
Decompression sickness seems only to involve normal biological responses to extreme environmental changes. However, it may be disease if what constitutes a normal response assumes only idealized environments. With such assumptions it would be a kind of trauma like being poisoned or stabbed or hit by lightning. The case of suffocation due to lack of oxygen, some would argue, is different only in the rapidity and extremity of the consequences when compared to decompression sickness or to disease caused by lack of nutrients from the environment. Suffocation, I believe, is a trauma and pathology caused by insult from the environment. Theoretical models describe the sustained lack of oxygen on most cells in the body as eliciting abnormal metabolic processes. Interestingly, whether the water was hypertonic, hypotonic, or isotonic would change what kind of trauma and biological abnormality was occurring.

4.5.3 Conditions Perceived as Pathological but Which Are Theoretically Normal

These examples involve conditions that are normal in reference to biological theory, but are potentially considered to be disease-like or abnormal in a conventional sense. For the normativist these would be diseases. Many examples here will involve the normal flora found in the intestinal tract and on the skin. Modeling of the human body’s normal anatomy and physiology has revealed that the gut is normally the home to trillions of bacteria, with viruses, protozoa and fungi as well. These organisms in the gut, unlike those on the skin, are not mere colonizers, but function in symbiosis with the host. The gut flora metabolize certain products secreted into the gut and synthesize other products that are absorbed into the body. Complex immune system functions monitor traffic across the gut wall, and monitor the make-up of the flora present in the gut lumen. An excellent review article (Guarner and Malagelada 2003) details some of the scientific models explaining the normal functioning of the gut flora and the role it plays in several possible biological malfunctions. The abstract of that article states:

The human gut is the natural habitat for a large and dynamic bacterial community,

---

131 Interestingly, most medical insurance policies do not cover treatment of decompression illness caused by scuba diving, due to it being expensive and because the activity is a high-risk and voluntary activity that occurs in extreme environments.
but a substantial part of these bacterial populations are still to be described. However, the relevance and effect of resident bacteria on a host’s physiology and pathology has been well documented. Major functions of the gut microflora include metabolic activities that result in salvage of energy and absorbable nutrients, important trophic effects on intestinal epithelia and on immune structure and function, and protection of the colonized host against invasion by alien microbes. Gut flora might also be an essential factor in certain pathological disorders, including multisystem organ failure, colon cancer, and inflammatory bowel diseases. Nevertheless, bacteria are also useful in promotion of human health. Probiotics and prebiotics are known to have a role in prevention or treatment of some diseases (Guarner and Malagelada, 2003, p. 512).

Loss of this normal flora by sterilization would be an abnormality. Such losses of normal flora are an important component mechanisms in the disease of *Clostridium difficile* overgrowth. The overgrowth can result in subsequent destruction of the colon wall and is often the resulting side-effect of using anti-biotics that inadvertently kill off significant portions of the normal flora. It also turns out that many of the species found normally in the gut and that are involved in symbiotic normal functions, in one context, can be pathogens in other contexts. Many varieties of E. Coli are found normally in the gut and feces, but if these species enter the urethra (normally sterile) then a urinary tract infection can result. Certain bacteria or worms are always pathogenic if they infect the gut. Other bacteria not known to be part of the normal flora can *colonize* without infecting the gut or skin, meaning that they are present but not at that time eliciting an immune response, but potentially could. The presence of bacteria in the large intestine is not a legitimate disease claim, and has necessitated the development of concepts such as normal flora, colonization, commensal and symbiotic relationships, all in contrast to infection. These concepts and the notions of normality and abnormality here are generated by the development of the models that describe these phenomena. These senses of normal and abnormal are independent from conventional judgments about the desirability or normality determined by our community’s perceptions about bacteria (or ‘germs’) inhabiting our bodies.

Pregnancy is also a type of symbiotic relationship and involves the growth of a biologically foreign entity inside the body. It resembles cancer in some aspects and infection in others. The immune systems response to pregnancy is complex. However, pregnancy and the changes of the immune system and other physiological changes during
pregnancy are not diseases—they are designated as normal functions in the models developed to explain these phenomena. An interesting example of biological malfunction during pregnancy is hemolytic disease of the fetus and newborn. Basically this is a condition where the immune system of the mother has come into contact with certain proteins on the surface of fetal blood cells—proteins different from those on the mother’s red blood cells. The mother’s immune system produces antibodies to these proteins. In future pregnancies if the new fetus has a blood type with those particular surface proteins, the mother will produce antibodies that can cross the placenta and destroy fetal red blood cells and cause anemia in utero (Janeway, et. al., 2001). This is a legitimate disease for the newborn, and represents an abnormal attack on the fetal red blood cells by a toxic antibody produced by the maternal immune system. For the mother the production of antibodies to the fetal RBCs is a normal process. Interesting models of how different blood groups emerged in human populations, how these are normal variants, and how such variation is involved in resisting infectious disease and causing transfusion related sickness are reviewed in Antsee (2010).

4.5.3.1 Spermatorrhea

This is, on my account, an illegitimate disease claim. It is basically the medicalization of a normal biological function or process due to cultural notions of abnormality. It is a supposed pathology of sperm production whose most reliable outward symptom was involuntary emission of male ejaculate. This was a putative disease claim for Western European medical theory in the nineteenth century. In Victorian England it was taken quite seriously and respectable medical thinkers and journals considered theoretical hypothesis about its nature, causes and potential cures. Its relationship to the ‘disease’ of masturbation and the curative effects of circumcision were seriously discussed (Darby, 2005). Interestingly the disease entity (yijing) was one of great concern within the framework of traditional Chinese medicine during the same period. Yijing had been a disease in Chinese medicine for at least two thousand years and continues to be today (Shapiro, 1998).

Here we have an example of a putative disease claim that is rejected as illegitimate within one framework due to biological and sociological research concluding
that the symptoms were theoretically normal and not the result of any underlying abnormality, while another explanatory framework continues to see it as a theoretical abnormality. These theoretical notions of normality stand regardless of the conventional judgments of harm or deviancy that might be held by particular communities. Within traditional Chinese medicine yijing is a legitimate disease (or at least a well established putative claim), while in Western medicine it is now an illegitimate disease claim.

4.5.4 Normal Variations as Opposed to Pathology

The problem of distinguishing normal variation from pathology was discussed in Chapter 3 and included examples such as blood groups and eye color. The sinister conditions of left-handedness and hirsutism are also interesting cases. With hirsutism there is a continuum regarding the amount of body hair that people have, and describing what is normal is challenging, just as determining when baldness is or is not biologically abnormal. As discussed in Chapter 3, the basic problem exposes the weakness of trying to establish disease by strictly statistical senses of abnormality (i.e. atypicality). The problem is one that the normativist believes can only be solved with the use of conventional senses of abnormality, and more specifically by appealing to the perception of the biological function or structure as disvalued, harmful or inconvenient to those afflicted. My position is that scientific models in the process of increasing the accuracy and detail with which they describe the biological phenomena in question, create a theoretical sense of normal and abnormal function that is importantly different than the purely conventional or statistical notions of normality. The sense of normality is not established globally. Science settles the variation issue locally, creating the sense of normality appropriate for each model and the explanatory task at hand. The disease claims for left-handedness, green eye-color, and type A blood are all illegitimate. Hirsutism in itself is not a legitimate disease claim, but might be a sign of an underlying pathology, and might form one amongst other signs of a legitimate disease claim, such as ovarian cancer. It is important to keep straight the level of analysis involved with normality judgments. At the observable level admixtures of conventional and theoretical abnormality are hard to prevent.
A very controversial example involving normal variation is homosexuality. This is currently not a legitimate disease claim. However, there are research projects looking at the neuro-endocrinological and genetic aspects of sexual preference and behavior. These projects are working on explanations that not only distinguish biological processes causing sexual preference but differences between male and female as well. Suppose that science develops models explaining sexual preference for the same gender as caused by a certain combination of genetic factors. How would the question of whether this is a normal variant (like eye color) or a mutation leading to disease (like sickle cell anemia) be settled? One issue would be developing models of a circumscribed neural system that has a normal function. Models would also have to distinguish normal variation in function and structure from abnormalities. In such a scenario same-gender-preference might be described as a result of an abnormal neural structure, and this would be a putative disease claim. Legitimacy would require the models to survive criticism and further testing, be refined, and eventually accepted as unbiased and accurate. This scenario seems unlikely given what is known currently, and I assume that such research will eventually explain the neural differences between sexual preference (as well as any between males and females) are variants of normal. But my opinions are surely influenced by conventional senses of what is normal and abnormal. Suppose that genetic differences in neural structure were discovered that explained the preferences of pedophiles. Would we expect this to be explained as just a normal variation in neural structure? Explanations of diseases that involve human behavior or gross human appearance or functionality will always present challenges as far as untangling the conventional senses of abnormality from the theoretical. However, even if conventional senses help initiate the research, the eventual resulting theoretical notions of normality and abnormality cannot be predicted and depend increasingly on the details of the phenomena as described in the model. No one would have guessed when work on intestinal physiology and infections began that it would have been normal for so much biologically necessary bacteria to reside there. Furthermore, regardless of whether

132 Recently a research group in China sent a brief communication to Nature (a prestigious journal) about the correlation between normal serotonin signaling in mice brains and gender preferences in mating behaviors (Liu, et. al. 2011).
differences in sexual preferences are theoretical variations of normal or some are abnormalities, how we decide to handle such information or individual differences in preference or behavior in our community is something that can be negotiated.

4.5.5 Statistically Pervasive Diseases (i.e., Widespread Pathology)

The naturalist that wishes to rely on statistical notions of normality faces the problem of what to do with prevalent disease. Examples here include diseases such as atherosclerosis, hypertension, tooth decay, etc. If most or many species members have a disease, then it becomes hard to distinguish diseased functionality from normal merely by observing and measuring functional mode and efficiencies. Theoretical models can start with tentative hypotheses until further details allow more accurate determinations. Theoretical models can also abstract away from the actual circumstances that organisms operate under in their natural environments. In fact, most laboratory experimentation with biological function is done under exquisitely controlled and simplified conditions. In this way theoretical models are simplifications and idealizations away from any actual organism or environment.

Many of the examples along these lines have complex multifactorial models of causation. Causation results emergently from the interplay of many complex components. Many of the subsystems involved in causation, if examined in isolation, might be considered as operating normally or as variants of normal, but in combination with other variant systems produce emergent abnormal function. Issues with multiple realizability occur. These models also involve complex mixtures of genetic predisposition and environmental factors. These diseases tend to increase in prevalence with age. As with any disease that is pervasive and increases in prevalence with age, the issue of what constitutes normal from pathologic aging becomes a difficult issue. Atherosclerosis and essential hypertension are legitimate disease claims (or at least putative disease claims very close to legitimacy).

4.5.6 Ontogeny and Development

There is hope that science may be able to develop sets of reliable scientific models for the normal biology of aging, because it has made great strides in doing so for
ontogeny and development. The field of embryology and pediatric medicine provide impressive epidemiological (i.e. statistical) and mechanistic (i.e. theoretical) models for normal development. These fields have also developed models of abnormalities that explain human diseases that effect development and growth. Disease claims such as De Georges syndrome, Down’s syndrome, thalidomide limb aplasia, and many more are legitimate.

4.5.7 Aging as Disease

For our contemporary society, aging and death are seen as undesirable aspects of biology. Many aspects of our culture deal with the fear of aging and death, and many facets of our economy deal with easing or avoiding these inevitabilities. There are many research projects whose goals are to slow or prevent the effects of aging. Some might even claim that medicine itself is essentially the technological attempt to stave off aging and death. So is aging a disease?

For the normativist it is disease. However, on my account we must wait until science has much better models of the mechanisms of aging, and how these relate to the necessities of multicellular organisms and cancer. As best I can tell, based on the research now available, there is a normal programmed life span for humans. There is a normal aging process, even if it has larger variations than those found in growth and development from zygote to adult. The burden of proof is on those claiming aging is a pathology. All the familiar issues will plague the development of such models. It will involve grossly observable changes in ability, structure and behavior. It will be hard to separate conventional value judgments from theoretical notions. Idealizing normal function to that of the young male adult has been and continues to be an issue in biological research. Conventional norms influence theoretical models, but part of seriously addressing issues of aging will require that these sorts of biases are minimized as much as possible. It will also be difficult at first for researchers to distinguish normal variation from pathology.

The ultimate difficulties will not revolve around whether or not aging in general, or any particular aspects of aging, are designated as legitimate disease claims. The ultimate difficulty will be what to do with the knowledge gained by developing the
models. Even now the economic burden of sustaining the chronically ill is a problem. Such knowledge will likely allow intervention and such intervention will be desired (at least based on our current culture). Issues of who has access to such intervention and the impact such widespread intervention would have on society will be difficult regardless of how the disease issues are settled. The settling of the issue of disease will help prevent the creep of medicalization and may help focus people on the real types of negotiation and discussion needed to make decisions on what we should do about slowing or preventing aging.

Consider osteoarthritis. This is basically a condition caused by wear and tear on the joints. However, there is variation in how early and how severely people are affected by this condition. Is it a legitimate disease claim or just part of normal aging? To the best of my knowledge concerning the models, run-of-the-mill osteoarthritis, although a clinical problem, is not a legitimate disease. If it were a putative or legitimate claim it would have to be so in comparison to the joint function and structure of the young adult joint which has not been exposed to much vigorous use. Here the model of normal function and structure would be significantly idealized. Skin wrinkling with age, on my analysis, is another case of an illegitimate disease claim. The weakening of the immune system with age is also probably part of normal biological development. We treat this clinically with vaccinations. Another age-related disease claim involves senility—loss of memory and cognitive speed with age. Is some amount of senility normal? What about extremes? This disease claim faces many of the general problems just reviewed. It is at best a putative claim, and I believe garden-variety senility will prove to be an illegitimate disease claim. But compare this to the case of certain dementias, such as Alzheimer’s or Huntington’s. For these we have increasingly good models about what is causing the loss of cognitive ability. Huntington’s is a legitimate disease claim, and Alzheimer’s is a syndrome and putative claim that looks promising as a legitimate disease. Alzheimer’s has complex multifactorial causation and like essential hypertension and atherosclerosis the models of causation are complex and involve

---

133 However, it may occur at an extremely young age in a person not unduly exposed to the sun, and may be abnormal biologically.
elements of aging. Distinguishing normal variation from significant variation is challenging and takes time.

4.5.8 Environmental Insults and Injuries

Several conditions claimed to be diseases are due to less than hospitable environments. Such examples highlight the issue concerning the idealization of the environment in models of normal biological function. It is also interesting to consider whether all hostile effects on an organism result in pathology, or only some. Perhaps some effects of a hostile environment elicit a normal biological response. A normal response might even be one that is perceived as harmful to the organism (see the discussion of suffocation and decompression sickness above). It is also important to note that my account of ‘disease’ is wide enough to encompass injuries and traumas, such as being stabbed by a knife.

Infectious diseases seem to be paradigmatic examples of disease. Such invasions by hostile organisms from outside the sterile core of the body are legitimate disease claims. Sun burns of the skin can come in a spectrum of severity, but are also legitimate destructions of normal structures and disruption of normal function. Nutritional diseases, such as beri-beri, pellagra, vitamin A deficiency, and scurvy are also classic examples of disease, and have models of the increasing dysfunction of metabolism as the body is deprived of necessary nutrients. Reliable and verified models explain that certain vitamins and minerals are required by certain species and that the body cannot normally synthesize these and must acquire them by ingestion. Other substances ingested can cause metabolic malfunctions and are diseases of poisoning or overdose. Above we considered decompression sickness and I stated that it was not a legitimate disease claim, but rather the normal biological response to unusual or extreme changes in the environment, but that suffocation and drowning were legitimate claims on my account. Ultimately it is the presence of reliable models contrasting normal and abnormal function of a circumscribed system to environment effects that determine legitimate disease claims. Traumatic injuries also count as legitimate claims if there are legitimate models of the structural and functional damage caused. Hanging, gun shot wounds, burns, etc are all legitimate disease claims.
4.5.9 Old Injuries, Old Traumas, and Malformations

The examples in this section are not active processes, but the result of previous trauma or malfunction. These lingering malformations or residual malfunctions are part of the current stable structure and functioning of the organism. Normativists are eager to point out that labeling these differences as abnormal invites discrimination (Amundson, 2000). They also point out that in some cases concentrating on ‘fixing’ abnormalities instead of accommodating differences perpetuates discrimination (Dreger, 2004). But these decisions about what to do in the face of disability, differences, or disease are different decisions than whether or not the condition results from a biological abnormality. These issues were discussed at greater length in Chapter 3.

There are many examples of legitimate disease claims such as the residual weakness, aphasia or dysarthria that remain years after a stroke, due to the neural connections lost at the time of injury that could not be regained or compensated for. Paralysis from car accidents, or from spina bifida, are also legitimate claims to pathology. A person who has a vasectomy or tubal ligation can be understood to have acquired an iatrogenic disease. The existence of a previous biological malfunction or trauma to explain the current condition is important in the legitimacy of such disease claims. There are also many legitimate and putative birth defects and congenital syndromes that result from malfunctions in embryological development.

4.5.10 Macroscopically Observable and Cosmetic Abnormalities

Some diseases affect appearance and have very good causal explanatory models. These types of abnormalities are observable and can be considered and judged without any specialized knowledge. Any theoretical model of abnormality will have to co-exist or compete with conventional judgments. Certain cases of ichthyosis (i.e. fish-scale-like skin), psoriasis, and alopecia areata are increasingly understood at the molecular and genetic levels of analysis. However, when explanatory models are not available and are only suspected, conventional senses of abnormality fill the void, and medicalization often occurs, especially if those having the condition believe biotechnology can help them. But
in these cases where the theoretical models are not yet available, regardless of whether medical intervention is or should be offered, these are not yet legitimate cases of disease.

Consider baldness again. To normativists this will be a disease that is relative to cultural views at any given time or place. The normativist focuses on consequences. Our current culture mostly values youthful appearance and will find baldness undesirable and abnormal. However, some cultures may find baldness a sign of experience and survival and value it. It may not be considered abnormal in those contexts. In such societies men might even fake baldness, the way that certain celebrities wear fake glasses to appear more sophisticated. The naturalist will tend to consider baldness a part of normal aging, but this will depend on how normal is determined. If it is determined statistically, then the sample selected will matter. There is the potential for biasing by idealizing what is normal to the young members of a species. My position will stress mechanistic function and the cause of the external appearance in question, rather than its typicality. Was the cause a biological malfunction at a lower level of analysis? If so it is a disease.

Alopecia areata\textsuperscript{134} is a legitimate disease claim. Baldness, secondary to trauma, like a burn, would also be pathological. Androgenic alopecia in a middle aged man is a putative disease, but may turn out just to be a normal variation in older men. Currently we lack a well verified and uncontroversial scientific theory of normal aging or normal appearance for humans.\textsuperscript{135} Wrinkles are in a similar position to baldness regarding being a sign of disease.

Obesity and shortness as disease claims are interesting. They are macroscopically observable and in our current culture judged as less desirable aspects of body habitus. These qualities come on a spectrum and are relative. Even to define them operationally requires a judgment about where to make a cut-off between normal variation and an extreme. I discussed these problems in Chapter 3. These observable signs can sometimes be part of a syndrome with a biological abnormality as the explanation. There are models of disease caused by mutations in the genes for growth hormone receptors that result in extreme short stature. See Kamboj (2005) for an excellent review of those diseases.

\textsuperscript{134} This is an immunological attack on hair follicles causing loss of hair in smooth round patches, and in severe cases all hair on the body.

\textsuperscript{135} Science does have much better worked out models for the normal appearance and normal changes in appearance for species other than humans.
associated with short stature and discussion of the many clinical problems for which recombinant exogenous growth hormone might be used as intervention. There are also genetic defects that are implicated in extreme obesity due to disruption in the biological systems that regulate energy storage at the cellular level and involved in signaling the brain concerning hunger and satiation (i.e. the system involving leptin, grehlin, orerin, insulin, etc.). See Grimm and Steinle (2011) for a concise review. Some legitimate disease claims involve obesity or shortness as grossly observable symptoms. These models explain why a person is extremely short or obese, but do not necessarily make obesity or shortness in and of themselves diseases. It is conceivable that eventually all cases of extreme shortness (or obesity) might be explained as caused by one or another known biological abnormality. This situation would perhaps allow calling extreme shortness (or obesity) a disease. These comments apply equally to gigantism or cachexia.

Norman Daniels has argued that treating shortness (a symptom) only in those patients who can be diagnosed with a lower level biological abnormality is unfair. Daniels argues that the stigma and disadvantage is the same for those with diagnosable causes and those without (Daniels, et. al., 1992). I agree that if one is intervening to prevent the social stigmata then fairness requires treating all those with the symptoms. However, only some cases of shortness on my account are legitimate disease claims while others are putative. The scenario where all cases of extreme shortness are considered signs of disease raises an issue about what it is that we are classifying—clinical problems or underlying causal mechanisms. I have resisted putting into my explication of legitimate disease claim that it refer to the lowest level cause of theoretical abnormality—below which there is only normal biological function. This specification would allow us to dismiss some high-level-of-analysis abnormalities (one’s with difficult admixtures of theoretical and conventional senses of abnormality). Shortness itself, even if all extreme cases had good lower level explanations, like receptor mutations, would then not be a disease claim, but rather the disease would be the lower level abnormalities giving rise to the observable feature. This might greatly increase the number of diseases, as several distinct lower level models might give rise to the same observable signs and symptoms. I have chosen not to decide on this nosological issue in this essay, and leave it an open question. I believe that biomedical science is actually in the process of dealing
with this issue, particularly as more and more models of disease are bolstered by the information coming in the wake of the human genome project.

Acne is also a grossly observable clinical problem and an interesting case regarding the claim of disease. In a certain age group it is prevalent and mild. Is it just a normal aspect of development through puberty? Is it only extreme cases that are legitimate disease? In the Amazonian society that judged the rash of dyschromic spherocochiasis to be desirable, perhaps acne would also not be a clinical problem, nor a disease. In our own culture may be a case of medicalization. Patients who wish not to be afflicted with the problem and those with potential therapies conspire to cast the problem as a biological abnormality. Models of the mechanisms causing the development of acne (inflammation, sebum production, the cycle of epithelial cell turn-over, the colonization of follicles with Propionibacterium acnes, etc.) and those factors that exacerbate and alleviate its severity are also continuously under research (Wolff, et. al., 2005). The extremes of acne can cause severe scarring and involve extreme and abnormal amounts of inflammation, and represent strong putative claims for biological abnormality, if not legitimate status. The less severe cases of acne are only putative disease claims, and controversial ones as they may turn out to be just normal parts of development. Furthermore, tentative models tend to idealize the normal as the current conventional notions of youth, beauty, and desirable appearance.

4.5.11 Implication for Discredited Disease Claims

There are disease claims that, in retrospect, we recognize as illegitimate. In hindsight they are speculative, sometimes intriguing, claims about biological abnormality. However, they are sometimes reminders of the terrible injustice and suffering that certain groups of people can inflict on others in the name of medicine and normality. Normativists and those with projects more activist than theoretical highlight these sorts of examples. On my account such cases are illegitimate disease claims because we now recognize that criteria 5 cannot be met; the theoretical models proposed were unacceptably biased and had very poor fit with the phenomena being described. In many of the most egregious cases the evidence available, even to contemporaries of those proposing the models, should have been enough to make these claims illegitimate. Of
note, though, is that even these poorly veiled attempts at passing off personal and social agendas as theoretical abnormality did posit causal explanations in order to medicalize the condition. These examples usually involve aspects of human behavior and gross physical anatomy and thus mixtures of conventional and theoretical senses of abnormality. Some illegitimate claims were due, in part, to an inability to explore causes below a certain level of analysis. Biomedical theorists had to make due with what could be observed, and with the types of evidence that could be gathered to test hypotheses.\textsuperscript{136}

Masturbation and homosexuality have been discussed as illegitimate disease claims, as have drapetomaia and dysaesthesia aethiopsia. These are all attempts to pass off value judgments as theoretical abnormality and thus employ medicine to enforce social and political agendas. There is no shortage of such attempts to medicalize certain conditions today so as to shift the nature of condemnation from the political and religious spheres and into the medical sphere, where purportedly nature and science can emphasize where the boundary between normal and abnormal resides. These controversies are similar to the abortion debate, where science is unfortunately asked to determine the boundary between human personhood and mere living cells. The theoretical model offered by Rush to explain what we now recognize as vitiligo was a poor model and has extremely poor fit with the actual phenomena being modeled. It is an illegitimate disease claim even if we want to believe that Rush’s was a more honest attempt at a theoretical and descriptive model than Cartwright. Cartwright clearly was repackaging his own personal politics and values. Normativists often do not highlight the history of the refutation of Rush’s disease claim and its replacement with models for the abnormality actually involved with vitiligo. The story involves more than shifting social practices and perspectives.

\textsuperscript{136} As mentioned, looking forward we might expand the number of diseases by deciding that claims must be made in reference to the lowest level of analysis for which a model posits theoretical abnormality, we might also look backwards and see different ways of categorizing based on issues with levels of analysis. In the past there were diseases that we now recognize as merely being signs or symptoms, or even recognize as normal processes that can cause harm. There are also examples of illegitimate claims that have been split up into combinations of illegitimate and legitimate disease claims as more evidence and models of lower level phenomena became available. Perhaps the cases of hysteria and melancholia are examples.
We should not conclude that all judgments of biological abnormality are equally conventional and ultimately based on widespread social practice. On my position, what needs to be done is careful analysis of each putative disease claim. Each claim needs an assessment of the qualities of the science being appealed to, and a close examination of the senses of normality involved.

4.5.12 Some Current Controversial or Interesting Putative Disease Claims

There are many interesting and controversial disease claims in contemporary American society. These are usually cases where a clinically identifiable problem is postulated to be due to an underlying biological abnormality, and thus deserving of medical diagnosis and intervention. This often involves issues about the designation of a disease claim, as legitimate, opening access to medical insurance and disability coverage. The case of obesity as a putative disease is interesting when compared and contrasted to alcoholism or other substance abuse syndromes. Some of these controversial putative diseases involve very specific claims about causation. An example would be the tremendous grass roots movement to validate the claim that autism is linked to childhood vaccinations. Regardless of good evidence and expert consensus, those believing this connection exists have been very resilient (see Institute of Medicine, 2004). This controversy has even resulted in the forgery of evidence, its publication and then its retraction from a prestigious medical journal (Specter, 2009). Evaluating and settling controversies about putative disease claims involves difficult issues. Who bears the burden of proof for producing the evidence, and who should foot the bill? How fast should we expect the evidence? Effectively dealing with such issues requires some combination of improved basic scientific literacy by the public and a willingness to recognize and trust expert consensus. It would also help if the media were not so obviously commercialized and determined to sell entertainment at the expense of fact checking and reasonable skepticism. The media continually reports on the latest ‘big discovery’ as if it were a reliable and settled view, when in reality it is merely an interesting and poorly understood new piece of evidence or a very speculative new model. The behavior of the pharmaceutical industry, as well as the organized profession
of physicians, could also be criticized at length. I shall examine just a few examples of current controversial putative disease claims.

Fibromyalgia is a putative disease claim. It is part of a family of putative chronic pain disorders. Each of these is identifiable as a clinical syndrome. This family includes fibromyalgia, irritable bowel syndrome, irritable bladder syndrome, and temporomandibular disorder. Fibromyalgia is characterized by chronic and diffusely distributed pain, particularly in response to tactile pressure on the surface of the body, along with increased fatigue, and possibly cognitive deficits. There are screening criteria to identify those likely to have fibromyalgia. The criteria consist of a history of the symptoms for at least 3 months with pain affecting all 4 body quadrants, both lateral sides, and above and below the waist. In addition, characteristic pain must be elicited at least 11 of 18 classic ‘tender points’ when a standard amount of force is placed on the skin over these body areas. An excellent review of the history of the clinical problem, current models of the biological systems involved, their normal function and the abnormalities that purportedly cause fibromyalgia, and current interventions, is available in Clauw (2009).

Irritable bowel syndrome presents as otherwise unexplainable abdominal pain, often with intermittent changes in bowel habits, including bouts of diarrhea or constipation, and complaints of bloating, bowel urgency and tenesmus. It is a diagnosis of exclusion, meaning that the responsible physician rules out all other causes of such symptoms before considering irritable bowel as a diagnosis. It is also sometimes called a ‘functional disorder’ which basically means that no underlying theoretical abnormality can be associated with the clinical syndrome. At the current time the consensus on this disorder is that it involves a psychosomatic component, and this is why designations such as ‘functional disorder’ and ‘diagnosis of exclusion’ are quickly mentioned by professionals reviewing the disorder. Khan and Chang (2010) present an excellent review of the clinical syndrome, how to diagnose and categorize it, assess its severity, and a review of possible treatments, along with speculations for some mechanisms at work.

Chronic fatigue syndrome (CFS) is a name given to the clinical problem of unremitting fatigue resistant to rest and unrelated to exertion. The symptoms are vague, and it is a controversial diagnosis, and certainly considered a diagnosis of exclusion by
responsible physicians. A review of hypothetical etiologies is given in Craig and Kakumanu (2002). Many of those searching for an explanatory model have focused on viral etiologies. Recently, associations between a family of murine leukemia viruses and chronic fatigue syndrome have increased hope of those advocating for this putative disease claim. However, the controversy has merely increased and recent editorials in the journals *The Lancet* and *Science* make clear that this disease claim is putative and still very controversial:

There is a general consensus that CFS is a heterogeneous family of disorders, and it seems most likely that these disorders arise from a constellation of pathophysiological causes. The results in the *Archives of Paediatrics and Adolescent Medicine* [providing some evidence of a link between CSF with a specific virus] received great media attention. But they do not prove that CFS is a physical disease. CFS is still far from being a well-defined entity. When the totality of available evidence is considered, the uncertainty around our understanding of the physical–psychological interaction taking place in patients with CFS only strengthens the case for giving research into chronic fatigue the high priority it deserves (Editorial *The Lancet*, 2010, p. 930).

The stormy debate over a potential cause of chronic fatigue syndrome (CFS) is nearing hurricane force. Last month, it prompted headlines suggesting that researchers have reached a dead end, scores of blog posts from disappointed patients, and accusations that scientists had gone beyond their data. The 14-month-old row intensified when four papers appeared in *Retrovirology* suggesting that reports linking the virus XMRV to CFS were based on false positives. The debate began in 2009 with a report in *Science* that XMRV, a retrovirus recently reported to have been found in prostate tumors, had been detected in 67% of a set of CFS patients but in only 4% of controls (*Science*, 9 October 2009, p. 215). Since then, one other group has found XMRV like viruses in CFS patients’ blood. But several teams have failed to detect the virus in CFS or cancer patients or in healthy people. Researchers have struggled to explain the discrepancies (*Science*, 17 September 2010, p. 1454). The potential link to CFS has had important consequences: Some CFS patients have begun taking antiviral drugs, which can have side effects. Last month, after being briefed on the original XMRV studies, advisers to the U.S. Food and Drug Administration recommended that CFS patients be barred from donating blood (Kaiser, 2011, p. 17).

The federal government has funded a large $1.3 million experiment with standardized criteria and techniques to try and settle some of these controversies with chronic fatigue syndrome.

Another interesting putative disease claim is that of Morgellons disease. In 2001
Mary Leitao reportedly observed a small fiber emerging from the facial rash of her two year old son, and then observed more emerging from different parts of his body. These fibers returned at intervals and she began a crusade to discover the cause and gain legitimacy for what she believes is a poorly understood disease. She has largely been dismissed by the established medical community and even accused of having a psychological disorder and of using her son as a means of attention. Mrs. Leitao has helped develop an international non-for-profit patient advocacy group. The organization has exerted enough pressure to cause the United States Centers for Disease Control to establish an epidemiological investigation into these claims of a new disease. Details can be found [www.cdc.gov/unexplaineddermopathy](http://www.cdc.gov/unexplaineddermopathy). Further characterization of the disease, including symptoms, pictures of representative fibers taken from patients, and case reports can be found at the advocacy web-site: [www.morgellons.org](http://www.morgellons.org). This patient advocacy web site says of this condition:

Morgellons disease is a poorly understood condition which a growing number of physicians believe to be a chronic infectious disease. The disease can be both disabling and disfiguring. The symptoms include itching, biting and crawling sensations, “filaments” or fibers which emerge from the skin, skin lesions which range from minor to disfiguring, joint pain, debilitating fatigue, changes in cognition, memory loss, mood disturbance and serious neurological manifestations. Although the Centers for Disease Control and Prevention (CDC) is currently investigating the disease, it is not yet fully recognized by the medical community. At this time, the cause of Morgellons disease is unknown and there is no known cure. Morgellons Disease is best explained in detail by our Case Definition ([from www.morgellons.org/faq-home.htm](http://www.morgellons.org/faq-home.htm) taken 3/16/11).

In contrast conservative physicians believe that the symptoms and signs of this disease are actually due to a persistent delusion on the part of the patients (and their families). They prefer to diagnose patients having these symptoms with *delusions of parasitosis* (see Accordino, et. al., 2008). An excellent review of the story of Morgellons and a sociological analysis of the interplay between the patient advocacy group and the established medical field are offered in Fair (2010).

For these putative disease claims, the legitimacy issue concerns the quality of (or

---

137 Several physicians who examined Mrs. Leitao’s son have speculated she suffers from Munchausen’s by proxy. Which is itself a putative psychological disease.
even existence of) any scientific models explaining the normal and abnormal functioning of a circumscribed biological system implicated in causing the symptoms. From this perspective the four disease claims above are putative, but have importantly different assessments as to how close to legitimacy they may be. For example, fibromyalgia is the closest to being a legitimate disease claim. The models are more advanced and more likely to be seriously considered not only by experts in the appropriate fields (neurology, psychiatry and rheumatology), but also more likely to be accepted by established practitioners of clinical medicine. Such acceptance is not fool-proof, but is frequently a reliable proxy usable by the non-expert regarding the quality of the biological models involved. Morgellons is the worst off regarding legitimacy. It is still considered an illegitimate disease claim by experts and the established medical community, and there is little in the way of models of abnormal versus normal function. What biological system has gone haywire and is producing those colorful fibers emerging from irritated skin? What is its otherwise normal function? Somewhere between these two, regarding distance from legitimacy, are chronic fatigue syndrome and irritable bowel disease—both diagnoses of exclusion, and both still controversial topics amongst the specialized fields that deal with these clinical problems (gastroenterology and neurology).

I have discussed the putative claim of spermatorrhea above and raise it again only to point out that there are potentially many similar examples. Alternative scientific and ontological frameworks might provide many putative disease claims. For instance, various types of witchcraft practiced around the world might have syndromes and putative diseases for which they offer intervention. From the modern Western medical perspective these are illegitimate disease claims. Homosexuality was also discussed above as being an illegitimate claim, but one that might re-emerge as a putative claim if certain research projects progressed or if certain personal or political agendas pushed for a biological explanation. Controversy over the extent to which sexual preference is a choice versus a genetic predisposition continues. This putative disease claim would prove to be a difficult analysis due to the controversial issues involved with describing any human behavior as abnormal. Next I will examine some issues with psychiatric disease claims.
4.5.13 Implications of My Position for Psychiatric Disease Claims

I shall briefly sketch a few implications of my account for disease claims in psychiatry. This topic is one much discussed in the philosophy of medicine. Many of the recurring issues for putative disease claims mentioned so far become particularly noticeable when human behaviors are being classified as abnormal. Such behaviors are always the subject of personal and community judgments as far as conventional normality and abnormality. Our perspective on what is and is not acceptable can be difficult to put aside when thinking theoretically about gross anatomy and behavior. Furthermore, explaining human cognition, its normal operation, and the normal functions that allow it, are daunting tasks. As discussed in Chapter 2, psychology and psychiatry tend to be more prone to paradigm shifts and conflicts over what constitutes the core techniques and models defining these disciplines. For these reasons psychiatry has developed an approach to diagnosis that avoids (as much as possible) unnecessary theoretical and etiological requirements for diagnosis. The diagnoses of the Diagnostic and Statistical Manual (DSM-IV-TR; APA, 2000) for the most part focus on signs and symptoms. Essentially the diagnostic criteria consist of features that hang together as a syndrome or features that together constitute what I have been calling a clinical problem. This allows diagnosis to be based on clinical history and observable patterns of behavior, regardless of underlying biological (or psychological) causation. Psychiatric diagnosis also relies heavily on criteria requiring that the behavioral or cognitive difficulties being considered interfere with normal day to day functionality. Normal here is conventional and relative to the lifestyle and community of the patient. Consider the criteria for diagnosing pathological gambling from the current DSM:

A.) Persistent and recurrent maladaptive gambling behavior as indicated by five (or more) of the following:

1. is preoccupied with gambling (e.g., preoccupied with reliving past gambling experiences, handicapping or planning the next venture, or thinking of ways to get money with which to gamble)

2. needs to gamble with increasing amounts of money in order to achieve the desired excitement

3. has repeated unsuccessful efforts to control, cut back, or stop gambling
4. is restless or irritable when attempting to cut down or stop gambling

5. gambles as a way of escaping from problems or of relieving a dysphoric mood (e.g., feelings of helplessness, guilt, anxiety, depression)

6. after losing money gambling, often returns another day to get even ("chasing" one's losses)

7. lies to family members, therapist, or others to conceal the extent of involvement with gambling

8. has committed illegal acts such as forgery, fraud, theft, or embezzlement to finance gambling

9. has jeopardized or lost a significant relationship, job, or educational or career opportunity because of gambling

10. relies on others to provide money to relieve a desperate financial situation caused by gambling

B.) The gambling behavior is not better accounted for by a Manic Episode. (APA, 2000, p. 671)

Psychiatry does actively seek models of the theoretical biological causation of observed behavioral syndromes. The degree to which these models are accepted as speculative or hypothetical versus accurate and unlikely to be significantly modified varies between disease claims. It is also currently fashionable in psychiatry to appeal to biological models at lower levels of analysis than mental mechanisms. The models describing the neural pathways involved in schizophrenia are increasingly felt to have good fit with the phenomena and are beginning to offer a lower order explanation of the observable behavior and psychological malfunction, and explanations of why certain pharmaceutical interventions alter these behaviors and experiences. Other examples of more putative disease claims for which biological models are being developed include alcohol addiction, pathological gambling, autism, the attention deficit disorders, the dyslexias, bipolar syndromes, and many more. These are all putative disease claims and differ in the degree to which their models are considered to have good fit and lack bias. I shall leave it to the reader to investigate the speculative models of abnormal biological or
cognitive function backing these putative claims. Most psychiatric diseases on my account are putative, with some closer to legitimacy than others. Those that completely lack or have almost no theoretical biological or psychological models of causation, such as some personality disorders, are far from legitimacy. Disease claims like schizophrenia are closer.

I do not see any fundamental reason that psychiatry or psychology should be distinguished from the rest of medicine and biology. These fields are making honest attempts to understand the phenomena in question with an eye on alleviating the suffering of patients. As long as the disorders that their patients are suffering are hypothesized to be caused by underlying malfunctions whether biological or psychological, and as long as models of such dysfunction are posited as verifiable through empirical investigation, then no distinction is needed. The fact that most of the disease claims of psychiatry remain putative is evidence of how difficult it is to study and explain behavior and phenomenal experience, and how difficult it is to put aside conventional judgments of what counts as normal and abnormal regarding such behavior and conscious experience.

4.5.14 Notions of Health on My Theory

On my account, the negative notion of health as the absence of detectable legitimate disease seems unproblematic. This would just be the absence of any known biological pathology. However, a positive notion of health is not so simple. This would be a completed theory of normal human biological form and function. This requires a completed theory of human aging and also a theory of normal human cognition. This may also require a theory of the ideal human environment, perhaps including the ideal social and community environments. A positive sense of complete human health may require more than just theoretical models of biological functions. It would probably require what in philosophy is sometimes called a theory of ‘human nature.’

4.4.15 Implication for Boundary Disputes

Similar to my brief comments on psychiatry, I cannot do justice to the difficulty of many of the boundary issues influenced by taking a position on how disease claims differ regarding legitimacy. My position does have implications for discussion of the
many boundary issues I highlighted in the introduction to this chapter. It is important to remember that we can use other factors besides legitimate claims of disease to settle boundary issues or policy problems involving boundary issues. Boundaries between biologically normal functions and abnormal that arise from theoretical models can be ignored in determining other boundaries. Ethical and practical reasons can be seen as more important in helping demarcate a boundary. Unfortunately many people realize it is easier to appropriate the authority of theoretical science than argue on practical or moral grounds. Appropriating the authority of science allows policy makers to avoid accusations of poor practical or ethical insight—it removes chances for political criticism. For this reason, appeal to the boundary set by legitimate disease versus other kinds of clinical problems or other types of human ill is hard to resist when arguing about where to set other boundaries. Equally disturbing is the opposite trend to dismiss scientific consensus if it appears to help draw a boundary that is practically undesirable, such as those who remain dissidents concerning the science on global warming or childhood vaccination causing autism (see Specter, 2009).

However, even if we have a reliable theoretical boundary between normal and abnormal biological function, how this informs our ethical or policy debates is still a decision that needs to be made carefully. Just because theoretical science has developed in such a way as to explain the normal function of certain biological parts of certain species is no reason to accept such function as the best situation either ethically or practically. There is no simple equation here. Rape, enslavement, murder, racism, etc, might be naturally evolved or statistically prevalent behaviors, but discovering this would not fix their moral status. Similarly, in medicine, just because something is not a biological abnormality according to our best biological models, does not mean that we might not end suffering by intervening to change biology or anatomy. The difficult questions remain even if we get some input from a scientific model of biological functionality.

For legitimate and illegitimate disease claims the boundary between normal and abnormal biological function could aid in decisions to set boundaries between therapy and extra-therapeutic medical intervention. It could aid decisions about setting boundaries for the role of physicians in society, and for what we owe each member of
society as far as basic health care and restoration of normal functionality. My position, however, leaves open a large indeterminate area of putative disease claims. Furthermore the prospects that science will make significant progress in a short period of time is unlikely, especially in the time frame that policy decisions must be made. So even if we can determine that some disease claims are legitimate, many of the ones we are most interested in will be putative and again we will have to rely on ethical deliberation and political negotiation to decide as a community how to proceed.

4.6 CONCLUSION: NATURALIST OR NORMATIVIST?

In this large fourth chapter of my project I have provided an account of what makes for a legitimate disease claim. Diseases are biological abnormalities in structure or function. The sense of abnormality at work in disease claims is a theoretical sense and is made in reference to the simplified theoretical model of the biological system being implicated. The legitimacy of the claim depends on the virtues of the scientific models being appealed to in the claim. The sense of abnormality is not a type of conventional sense, and is not in reference to widespread community values and practices. Perhaps a naturalist would argue that my position on science discussed in the second chapter is a complex way of allowing the conventional judgments of the scientific community to determine abnormality. My response would be that the process of refining and testing models involves interacting with the natural world—there is input from sources other than human values or opinions. Human empirical knowledge is perspectival, but it is not the same thing as any individual human’s or any community’s simple opinion or evaluation of phenomena. Biological model building may start off with inspiration from conventional senses of what is normal and abnormal, but as theory develops and is challenged and refined, so as to gain fit and accuracy, and avoid bias and misrepresentation, it creates another sense of normality. My position makes me a kind of anti-normativist since I reject that all disease claims are ultimately conventional judgments of value. My position, however, may not be attractive to naturalists, especially those wanting abnormality to be statistical or warranted by evolution. Perhaps my position is a weak-naturalist position. However, my position accurately captures the concept as currently used by physicians and biomedical researchers.
CHAPTER 5
CONCLUSION

My main thesis is that the core notion involved in the modern concept of disease (taken broadly as pathology) is biological (or psychological) abnormality in form or function, for a circumscribed biological (or psychological) system, contrasted against a theoretical sense of biological normality. Based on this conception, criteria were provided for distinguishing legitimate disease claims from both putative and illegitimate claims. My account puts the burden of legitimacy, objectivity, and respectability for the particular claims of disease onto science and the models it creates. Some scientific models eventually emerge from the complex process by which science creates, evaluates, and refines its simplified explanations. Often these models include descriptions of the normal functions, as well as possible malfunctions, of the biological systems involved. These models (including accompanying details about normality and abnormality) can have varying degrees of several virtuous qualities, including accurate representational fit. I argue that this sense of normality/abnormality, which is simultaneously created along with the other descriptive details of the biological model to which it belongs, is an importantly distinct sense. It is as scientifically respectable as other parts of the description and explanation of the phenomena in question. This is how science determines what is normal and abnormal in a way that allows disease to be as naturalistic and as objective as other scientific categorization concepts like species, virus, or sex.

In chapter 2, I gave a gross overview of science as well as a brief sketch of what it produces and how. Biological science produces simplified representational models of complex empirical phenomena. These models are originally hypothetical and go through a complex selective process that eventually rejects, modifies, or increasingly validates the model. I discussed some of the virtues and vices such human-created representational models can and do have, and suggested that specialists, as well as educated adults, can and do assess these virtues and vices. In particular, models should have good fit with the phenomena they represent and as little personal or social bias as possible, especially when they contain descriptions of normal and abnormal processes or structures that are grossly observable. However, many virtues and vices as well as (more and less reliable)
clues to these qualities are available for those evaluating particular scientific models. When science has trouble or cannot yet uncontroversially work out normal variation from pathology in certain descriptive models of biological process, then disease claims based on those models will be suspect, controversial or otherwise putative.

Chapter 3 argues that there are three importantly different senses of normality and, conversely, of abnormality. Judgments or ascriptions of normal and abnormal can appeal or refer to different sources for their authority or force. Theoretical senses of abnormality appeal to the simplified scientific models (particularly those in biology and psychology) that in the process of describing the phenomena in question also designate which processes and structures are normal and abnormal. Statistical senses of abnormality refer to actual measurements of parameters of the phenomena in question. Conventional senses appeal to personal or social norms or to judgments concerning what is proper. Distinguishing and trying to disentangle these different senses of abnormality when considering controversial disease claims is important. Clarity regarding possible equivocation across these senses provides insight regarding charges that certain disease claims are merely social constructions or are the inappropriate medicalization of a problem. In chapter 3, I also discuss Ron Amundson’s (2000) argument that, in biology, we should stop using the term normal. In arguing against Amundson, I highlighted the importance, for a naturalistic concept of disease, of being able to distinguish normal variation in form and function from pathological variation.

In chapter 4, I discussed naturalism and normativism about disease claims and provide my own account. I contrast my own project and perspective with other reasons for interest in the concept of disease. This chapter examined the relationship between controversies over the boundary between disease and non-disease and some other controversial boundary issues. Unfortunately, when a particular disease claim is putative or otherwise in question, the other distinctions or issues that one desires to resolve by appeal to the disease claim will not themselves be clear or uncontroversial. This chapter presented the main problem for normativism and naturalism. Normativists must explain, in a satisfying way, the uncontroversial cases of disease and how these are significantly different from the obviously illegitimate disease claims they often highlight. Naturalists must explain how science can provide a relatively objective distinction between normal
and abnormal that does not primarily rely on social norms or personal judgments. Similarly the naturalist must explain how to distinguish normal variation from pathology. Chapter 4 ends with my ideal criteria for a legitimate disease claim and with an examination of interesting cases designed to clarify and challenge my account.
BIBLIOGRAPHY


