So you say you are a revolution:
The genetic revolution in medical science

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This project is dedicated to everyone who ever challenged me to think differently.

I hope this essay will challenge its readers to do the same.

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Acknowledgments

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Part I – Introduction

> Scientific and technological developments these days invariably inspire a rhetoric of revolution, be it a computer revolution, a robotics revolution, or a biotechnology revolution. True to form, current research in genetics and biotechnology has been welcomed in similar terms. The veritable flood of books that have popularized, eulogized, and criticized this work all describe a scientific revolution unlike anything ever before attempted - a genetics revolution that will transform our relationship to nature … In particular, people have heard the Human Genome Project called the Book of Man, or the Quest for the holy Grail. And they have read media reports of amazing discoveries and miracle cures … Today, the revolution in biotechnology has become a "gene dream" - less an economic panacea than an environmental threat. And the optimistic promises of genetic therapies are countered by growing worries about the social and even eugenic abuses of genetic predictions.[1]

Through a variety of mechanisms, we have been convinced that we are in the middle of a genetic revolution that will radically change our whole biological universe. At least, these are the metaphors presented to us, and metaphors shape our reality.[2] Yet, the question remains – how well do these metaphors reflect the true potential of genetics? What is meant by the term genetic revolution? Or genomic revolution? Is it the translation of the Book of Man? Will it lead to amazing discoveries or miracle cures? Or will it be a threat to nature and an affront to humanity? How will our world change because of our investigation of the human genome? Specifically, how will medicine change because of our investigation of the human genome? This is the critical inquiry of this essay.

This essay is divided into five Parts. Part I is the introduction, where, in addition to laying out the format of the essay, I define a few critical terms essential to understanding the science in question.

[1] Nelkin, Dorothy, 1992, p. 38-9.
[2] Nelkin, Dorothy, 2001 and Van Dijck, Jose, 1998.

Part II is a characterization of revolutions in science. This Part is based largely on Thomas Kuhn's seminal essay The Structure of Scientific Revolutions. Kuhn investigates a number of historical examples of scientific revolutions and distills from them characteristics that are central to scientific revolutions. In doing so, Kuhn also defines the normal proceedings of science, i.e. when science is not in the midst of a revolution. Kuhn's criteria will be used to judge the validity of the phrases "genetic revolution" and "genomic revolution" in this essay.

Part III is my attempt to describe modern medicine. That task would be impossible without first describing the environment in which modern medicine operates – Western civilization in modernity – and the route that Western civilization took to arrive at modern medicine. These segments of Part III are largely informed by the works of Michel Foucault and Martin Heidegger. The essential task of modern medicine is to place illnesses within the framework of medical science by codifying them as diseases and to use medical science to find cures for those diseases. As such, the next segment of Part III portrays the process used by modern medicine to accomplish this task. The penultimate segment of Part III is an introduction to the ethical framework that guides the clinical encounter in modern medicine. That framework includes positions for the perspectives of both the physician and the patient. We broach the central inquiry of this essay in the last segment of Part III.

The central inquiry is addressed more directly in Part IV. The first segment investigates "genetic revolution." The second segment investigates "genomic revolution." To bring relevance these revolutions for the physician and the patient, the topic of ethics within the clinical encounter is reintroduced and discussed from the post-revolutionary perspective.

Part V is a conclusion that ponders the lessons of the "revolution." What does it all mean for us now?

Definitions

What exactly is a gene? The definition of "gene" has changed since the term was coined by Johannsen in 1909.

> [H]e wished this unit of heredity to be free of any hypotheses regarding its physical or chemical nature, i.e. the genes could be treated as calculating units…The gene was generally regarded as the unit of a genetic system, an indivisible entity in the processes of genetic recombination, self-reproduction, and mutation … The gene [can] now be regarded as a heritable unit of function, a segment of a chromosome, in most instances a segment of a Watson-Crick DNA molecule, which specified the structure of a single polypeptide chain and was made up of a large number of mutational sites, nucleotides by their material nature, biologically separable by recombination. The discoveries of gene technology beginning from the early 1970s … have proven that the nature of genes is even more dynamic than was thought...[3]

Contextually, both of these conceptions of the gene are used within biology. For example, the statement "We want to find the gene for diabetes" expresses the desire to find the indivisible unit of heredity that causes diabetes to be more common in the children of parents (and grandparents, great-grandparents, etc.) who themselves have diabetes. Conversely, the statement "The cells in this tumor express the gene Her-2/neu" uses the molecular definition of the term. In this case, the tumor cells have gene products that come from the DNA sequence labeled Her-2/neu. In some cases, such as monogenic diseases, the indivisible unit of heredity is the specific DNA sequence. In other cases, such as polygenic or multifactorial diseases, the indivisible unit of heredity is much more than a specific DNA sequence. The belief in medical science is that further research can establish links between the observed heredity of the condition and the molecular construction of the body for the condition. It is essential to keep these two definitions separate conceptually and understand which definition is being used in a given statement. It is very easy to conflate the two and assume that all observations of heredity have some single analog in DNA. This has yet to be proven to be true. For my part, where unspecified, the term "gene" is used in the molecular context.

In order to further understand the impact of this conflation, two more terms need to be defined: genotype and phenotype. The term "genotype" is defined as the complement of indivisible units of heredity. Modern biology holds that the genotype is the blueprint for making us who we are in a physical sense; it is the basis for defining and creating our constitution. A related term is "genome." This term refers to the complement of DNA in each of our cells. Modern biology conflates the two terms; genotype is genome and vice versa. Even so, research is showing that there are other elements in our cells that guide heritability; these are termed epigenetic factors. Since this is very cutting edge research and not completely defined, I will participate in modern biology's conflation, knowing that there is much more involved in the making of people than just DNA. My reason for doing this is that the literature, for now, speaks about the genetic revolution and the genomic revolution, not the genotype revolution.

The "phenotype" is the outward expression of the genotype. Diabetes is a phenotype, as is having two arms, two legs, ten fingers, ten toes, brown hair, etc. Each of our phenotypes is unique, just as each of our genotypes (epigenetic factors included) is unique. On a molecular level, the phenotype of a given gene is the range of possibilities

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[3] Portin, Petter, p. 274-5.

of products that could come from that gene if the sequence was somehow modified. An example is the range of products from the genes for hemoglobin. In people without sickle cell disease, the gene produces the protein known as Hemoglobin A. In people with sickle cell disease, the DNA sequence is changed at a specific point on chromosome 11 causing the gene to produce the protein known as Hemoglobin S. Thus, Hemoglobin A and Hemoglobin S are two of the phenotypes that come from the hemoglobin genes. Some argue that DNA sequences are phenotypes for the genotype, as defined as the complement of heritable units. However, this belief misses the point that the phenotype is the range of possibilities, not a single possibility. A given DNA sequence is a single possibility.[4] In a sense, both sides of this argument are correct. There are unique phenotypes that come from unique genomes; each of us is an example of such a unique phenotype. At the same time, there is a range of phenotypes that come from a range of genomes; the human species is an example of this extreme variability. For my part, I will be using the unique phenotype definition through most of this essay. I will be specific when I do not.

As previously implied, phenotype comes from genotype. Modern biology holds that the following is the process by which phenotype comes from genotype. A set of environmental cues trigger the activation of a transcription process for a gene. The DNA sequence in question is copied to yield a specific RNA sequence. Cellular machinery translates that RNA sequence into a protein strand. The protein strand is compelled to fold into a certain conformation and move to a certain spot in the cell by other elements in the cell. The protein then responds to the environmental condition that triggered the activation of the transcription process in the first place. The collection of cell responses

[4] Ibid, p. 275.

is what we observe as each of our unique phenotypes. This process is what is known as the central dogma of biology: DNA → RNA → Protein → Phenotype. As previously presented, a number of studies are challenging the central dogma by adding intervening elements, adding arrows in the opposite direction, adding new elements that require new arrows, etc. However, as it will be argued below, each of these challenges is an addition to the central dogma, not a full criticism of the paradigm. This is explored in further detail in Part II.

Finally, illness and disease have very specific meanings in the current medical anthropology literature. This convention stems from the anthropological investigation of western medicine as an ethnomedicine, like faith healing or ayurveda. The distinction is encapsulated in the following statement: "patients suffer 'illness' … [while] physicians treat 'disease'…"[5] Illness is the experience of the patient with a strange state of being. Disease is the experience as understood from a biomedical perspective.[6] This distinction and the relationship between the two concepts are explored in more detail in Part III. In this paper, the distinction will be followed. "Illness" refers to a personal experience whereas "disease" refers to a concept in medical science.

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[5] Hahn, Robert A. and Kleinman, Arthur, 1983, p. 312.
[6] Ibid. See also Hahn, Robert A., 1982.

Part II – Revolutions

> Look first at a particularly famous case of paradigm change, the emergence of Copernican astronomy [in which the Earth revolves around the Sun]. When its predecessor, the Ptolemaic system, [in which the Sun revolves around the Earth], was first developed during the last two centuries before Christ and the first two after, it was admirably successful in predicting the changing position of both stars and planets...But to be admirably successful is never, for a scientific theory, to be completely successful...In the sixteenth century, Copernicus' co-worker, Domenico da Novara, held that no system so cumbersome and inaccurate as the Ptolemaic had become could possibly be true of nature. And Copernicus himself wrote in the Preface to the *De Revolutionibus* that the astronomical tradition he inherited had finally created only a monster. By the early sixteenth century an increasing number of Europe's best astronomers were recognizing that the astronomical paradigm was failing in application to its own traditional problems. That recognition was prerequisite to Copernicus' rejection of the Ptolemaic paradigm and his search for a new one.[7]

The Copernican revolution, which was powerful enough to change the Earth's position in the cosmos, is widely cited as the first scientific revolution and serves as the model for transformations of science of this sort. In The Structure of Scientific Revolutions, Thomas Kuhn, a scientist and a historian of science, gathers this and other examples of scientific revolutions and extracts from them the essential characteristics of scientific revolutions. Part II of this paper is a full depiction of these attributes.

Kuhn

At the heart of Kuhn's argument is the well-informed belief that normal science operates by building upon itself and when elements worthy of scientific inquiry are proven to be incompatible with what is understood by normal science, there is an opportunity for revolution, the overthrow of normal science. Kuhn defines two interrelated terms to make his argument: paradigm and anomaly. A paradigm is that which is established by normal science; it is the overarching description of the way the

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[7] Kuhn, p. 68-9.

world works in the view of a given field of science. It is the given field's picture of the world. For example, prior to the Copernican revolution, the paradigm in astronomy was that the Sun, Moon, and the Planets revolved around the Earth. Successful paradigms are self-validating. The paradigm predicts the results of a given experiment, and, when those outcomes occur, the paradigm is proven to be true and applicable to a new realm, albeit a small realm, of nature.

> The success of a paradigm…is at the start largely a promise of success discoverable in selected and still incomplete examples. Normal science consists in the actualization of that purpose, an actualization achieved by extending the knowledge of those facts that the paradigm displays as particularly revealing, by increasing the extent of the match between those facts and the paradigm's predictions, and by further articulation of the paradigm itself…No part of the aim of normal science is to call forth new sorts of phenomena; indeed those that will not fit the box are often not seen at all. Nor do scientists normally aim to invent new theories…[8]

Paradigms define what science and, by extension, scientists are allowed to perceive. Paradigms define the problems that are fair game for a given field of science. Astronomers do not investigate the Krebs cycle in plants because it is outside of the paradigm of astronomy. By defining problems, paradigms point to the answers to the problems and insulate the field from issues that could break the paradigm.[9] Since paradigms define what scientists are allowed to see, it could be argued that paradigms are normative.[10] In addition to defining the way the world actually is, they define the way the world should be, assuming that the paradigm is true. To borrow an apt phrase from Part III, paradigms normalize the objects of the world. They fit the objects of the world into a world picture.

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[8] Ibid, p. 23-4.
[9] Ibid, p. 36-7.
[10] Ibid, p. 109.

Anomalies are objects in the world that do not fit the paradigm. If the world is a jigsaw puzzle, anomalies are pieces from a different box. With this analogy in mind, it is clear that anomalies can only exist with a mature paradigm in the background.[11] Otherwise, how would the researcher know that the puzzle piece does not fit the puzzle being investigated? If the background paradigm is immature, then the supposedly anomalous puzzle piece might belong to the puzzle being investigated. In that case, the paradigm is adjusted to accommodate the anomaly and, in doing so, the paradigm is validated and made applicable to a new segment of nature. It is just as possible, however, that the anomalous piece is indeed an anomalous piece and the investigator has been deluded into forcing it into the dominant paradigm. This is especially true if the paradigm is fragile to begin with. Any ill-fitting piece of datum can knock a paradigm into crisis,[12] and a moment of crisis opens the way for a new paradigm to be adopted,[13] for a scientific revolution to occur. However, in order to validate a new paradigm, the anomaly and the paradigm it calls into question must be significant.

> Anomaly appears only against the background provided by the paradigm. The more precise and far-reaching that paradigm is, the more sensitive an indicator it provides of anomaly and hence of occasion for paradigm change. In the normal mode of discovery, even resistance to change has a use that will be explored more fully in the next section. By ensuring that the paradigm will not be too easily surrendered, resistance guarantees that scientists will not be lightly distracted and that the anomalies that lead to paradigm change will penetrate existing knowledge to the core.[14]

Once penetrated to the core, as Ptolemaic astronomy was by Copernicus' argument that the Earth went around the Sun, the old paradigm becomes too feeble to

[11] Ibid, p. 52-3.
[12] Ibid, p. 79.
[13] Ibid, p. 67-8.
[14] Ibid, p. 65.

guide normal science. However, not every scientist in the field will convert to the new paradigm immediately. For those who do, the world is a completely different place.

> Examining the record of past research from the vantage of contemporary historiography, the historian of science may be tempted to exclaim that when paradigms change, the world itself changes with them. Led by a new paradigm, scientists adopt new instruments and look in new places. Even more important, during revolutions scientists see new and different things when looking with familiar instruments in places they have looked before. It is rather as if the professional community had been suddenly transported to another planet where familiar objects are seen in a different light and are joined by unfamiliar ones as well. Of course, nothing of quite that sort does occur: there is no geographical transplantation; outside the laboratory everyday affairs usually continue as before. Nevertheless, paradigm changes do cause scientists to see the world of their research-engagement differently. In so far as their only recourse to that world is through what they see and do, we may want to say that after a revolution scientists are responding to a different world.[15]

The old puzzle is traded for a new puzzle. Just as new puzzle pieces did not fit in the old puzzle, i.e. they were anomalous, the old puzzle pieces do not fit the new puzzle. An astronomer cannot believe in both Copernican and Ptolemaic astronomy at the same time; they are mutually exclusive. Trying to fit data from experiments in one paradigm into the theories of another paradigm is an exercise in futility. Two paradigms will not communicate with each other.[16] This also means that, although a scientist may alternate between two paradigms, that scientist must alternate as one would flip a light switch; it is either one paradigm or the other.

Those who do not switch to the new paradigm, those who would rather adhere to the old vision of the world, become adversaries of the new school. But, the dialectic between the two schools of thought is essential to filling out the new paradigm and completing the downfall of the old paradigm.

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[15] Ibid, p. 111.
[16] Ibid, p. 149.

> Lifelong resistance, particularly from those whose productive careers have committed them to an older tradition of normal science, is not a violation of scientific standards but an index to the nature of scientific research itself. The source of resistance is the assurance that the older paradigm will ultimately solve all its problems, that nature can be shoved into the box the paradigm provides. Inevitably, at times of revolution, that assurance seems stubborn and pigheaded as indeed it sometimes becomes. But it is also something more. That same assurance is what makes normal or puzzle-solving science possible. And it is only through normal science that the professional community of scientists succeeds, first, in exploiting the potential scope and precision of the older paradigm and, then, in isolating the difficulty through the study of which a new paradigm may emerge.[17]

Since the search for converts is a slow one, new paradigms are rarely established immediately following a crisis, even a pronounced crisis with a number of unexplainable anomalies against the old background. Copernicus was called a madman for years until a large body of evidence for his position was amassed by other astronomers.

> Copernicus' theory was not more accurate than Ptolemy's and did not lead directly to any improvement in the calendar...Copernicus' theory, for example, suggests that planets should be like the earth, that Venus should show phases, and that the universe must be vastly larger than had previously been supposed. As a result, when sixty years after his death the telescope suddenly displayed mountains on the moon, the phases of Venus, and an immense number of previously unsuspected stars, those observations brought the new theory a great many converts...[18]

This quotation points to another very important characteristic of paradigms and scientific revolutions. New paradigms are not valid and accepted until they are proven to be valid by research performed under the aegis of the paradigm being validated. This point was previously argued; at their start, paradigms only indicate a direction for research and the results expected from that research. It is not until that research is performed successfully, from the perspective of the paradigm, that the paradigm is (proven) true.

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[17] Ibid, p. 151-2.
[18] Ibid, p. 154-6.

Once the new paradigm is established, it becomes prone to the same process as the old paradigm. The new paradigm will have its own anomalies. And those anomalies may create crises penetrating enough to cause a new revolution, small or large, depending on the centrality of the assumption being questioned. The new paradigm that results will be fundamentally incompatible with the paradigm in question. Thus, the cycle continues. There is never any progress forward, since goals or norms are premised on the truths established by a paradigm and those truths are subject to questioning at times of revolution, nor is there any regress backwards. There is only the never-ending search for a perfect picture of the world. This is the way science proceeds.

Applicability to medical science

Kuhn is commonly criticized because The Structure of Scientific Revolutions focuses on revolutions in physical sciences, like chemistry and physics, and is therefore inapplicable to other, non-physical, fields of science. It is true that most of his examples come from disciplines within the physical sciences. However, the assertion that it is inapplicable to other fields of science is not true. First, Kuhn recognizes revolutions in biology, like Darwinian evolution.[19] Second, Kuhn acknowledges the lack of biological examples in his essay and attributes this situation to a wish not to overcomplicate his arguments with an excessive number of examples. He believes that there is evidence in the biological sciences to prove similar revolutionary processes.[20] Finally, modern biology is increasingly informed by the physical sciences. This operates as proof that

[19] Ibid, p. 171-2.
[20] Ibid, p. xi.

biology is indeed part of the attempt to find a perfect world picture though the scientific process.

Moreover, it can be argued that, even if Kuhn is applicable to the biological sciences, Kuhn is not applicable to medical science because medicine is a normative practice and not a true science. This is also not true. First, medical science is the closest thing we have to a biology of humans, in which case Kuhn is just as applicable to medical science as it is to biology. Second, the normative aspects of evidence based medical practice are only extensions of the normative aspects of the paradigms of medical science. Practitioners of evidence based medicine use medical science to show them the right way to treat their patients. It is my contention, further developed in Part III, that medical science concerns itself with preserving life and, as such, is primarily concerned with defining the characteristics of and the means of eliminating the pathological. Finally, there is strong evidence that the genetic revolution could satisfy Kuhnian criteria even if medical science, as a whole, does not. As written by Richard Strohman,

> Thus, all the usual suspects of a Kuhnian paradigm in its different phases of rise and fall are present. (1) A consensus is created in which "normal science" provides further insights into the molecular-genetic mechanisms of life. Alternative paradigms such as systemic or epigenetic biology languish. (2) A profound shift occurs in our perceptions of the world (from organisms to gene machines) in which we learn more and more about mechanisms and less and less about life. (3) There is a rapid recruitment of scientists to the work laid out by the paradigm. (4) There is rapid development and deployment of a vast new (bio)technology. (5) The new technology dominates the training of the next generation of scientists. (6) Finally, through the inexorable process of science, paradigmatic anomalies are discovered with increasing frequency. Ultimately, the original scientific paradigm falls of its own scientific weight, with one Kuhnian caveat: There must be a paradigm ready to replace what has fallen.[21]

Strohman is correct in asserting that there is a Kuhnian revolution occurring in the realm of genetics. However, the crisis, by his description, is not penetrating enough, as the paradigm that results from his crisis is rather similar to the existing paradigm in medical science. At best Strohman's revolution happens as part of the normal progression of medical science rather than as a genetic revolution. This is explored in more detail in Parts III and IV.

Kuhn's conception of scientific revolution is very compelling and is applicable to modern medical science. Kuhn's conception fits neatly with the definitions of modernity discussed in Part III of this essay.

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[21] Strohman, Richard, p. 194.

Part III – Modern Medical Science

> And if one considers that disease is at one and the same time disorder – the existence of a perilous otherness within the human body, at the very heart of life – and a natural phenomenon with its own constants, resemblances, and types, one can see what scope there would be for an archaeology of the medical point of view. From the limit-experience of the Other to the constituent forms of medical knowledge, and from the latter to the order of things and the conceptions of the Same, what is available to archaeological analysis is the whole of Classical knowledge, or rather the threshold that separates us from Classical thought and constitutes our modernity. It was upon this threshold that the strange figure of knowledge called man first appeared and revealed a space proper to the human sciences.[22]

These words appear at the end of the preface to Michel Foucault's The Order of Things, Foucault's effort to define the difference between the means of understanding the world in modern Western society and the means of understanding the world in Classical Western society. I find it very curious that Foucault chooses to introduce his work on modernity with a reference to medicine. Medicine is not unique to the modern era; the history of the physician extends back to the time of the ancient Greeks,[23] if not further. However, Foucault recognizes that there is something special about medicine, especially modern medicine, that traps the discipline in modernity. The practice of medicine invariably involves the effort to return an individual to a state of health through the interpretation of signs by an experienced practitioner of the craft. However, I believe there are notions in modern medicine that are distinctly un-modern in their origin. This Part is my attempt to prove that point. To do so, I will first address modernity as defined in The Order of Things and Martin Heidegger's "Age of the World Picture" and "The Question Concerning Technology." This will be followed by a discussion of modern medicine as defined in Foucault's The Birth of the Clinic. I conclude with a discussion

[22] Foucault, Michel, The Order of Things, p. xxiv.
[23] Canguilhem, Georges, 1989, p. 40.

of Georges Canguilhem's The Normal and the Pathological and the origin of the notion of human health.

Modernity

Foucault isolates the seventeenth century as a time of transition between the Classical era and the modern era. He notes that it is during this time that the belief in magic and superstition disappears and that rational order of science takes its place.[24] This transition changes our relationship with the objects around us in a very subtle manner. We have always identified the world around us through our senses; the items in the world have perceivable characteristics that make them identifiable. These are signs. Before the seventeenth century, Foucault argues, signs could exist without necessarily being perceived and understood by humans. After the appearance of science, signs only exist in the domain of human knowledge. Science, after all, is an organized and unified means of understanding the world. It is, by its very structure, a conservative enterprise, basing every new discovery on the discoveries of the past, attaching new knowledge to what is already known. Heidegger writes that, in modern science, "Explanation is always twofold. It accounts for an unknown by means of a known, and at the same time it verifies that known by means of that unknown. Explanation takes the place of investigation,"[25] the same way that, in Kuhnian terms, scientific experiments and discoveries are both guided by and used to validate a paradigm. This transition results in the banishment of magic and superstition, i.e., the elimination of a supernatural or divine connection for the items in the world.

[24] Foucault, Michel, The Order of Things, p. 55.
[25] Heidegger, Martin, "The Age of the World Picture", p. 121.

> From the seventeenth century onward, the whole domain of the sign is divided between the certain and the probable: that is to say, there can no longer be an unknown sign, a mute mark. This is not because men are in possession of all the possible signs, but because there can be no sign until there exists a *known* possibility of substitution between two *known* elements…It is here that knowledge breaks off its old kinship with *divinatio*. The latter always presupposed signs anterior to it: so that knowledge always resided entirely in the opening up of a discovered, affirmed, or secretly transmitted, sign. Its task was to uncover a language which God had previously distributed across the face of the earth; it is in this sense that it was the divination of an essential implication, and that the object of divination was *divine*.[26]

To be divine is to be sacred, to be beyond the capacity of questioning or reinterpretation. In the Classical era, signs were static and certain. They do not need a reason to be what they are; the observer simply understands the sign to mean what it means. In the modern era, signs are probable and are only held in knowledge by being linked to the pre-existing network of probable signs. The observer understands the sign to mean what it means because that is what the body of knowledge says is true. This reliance on a pre-existing body of knowledge grounds the modern sign and makes it less arbitrary in a way that is impossible for the Classical sign.

Heidegger ascribes five characteristics to the modern era: the appearance of science, the essence of technology being equated to modern metaphysics, art's transition into aesthetics, the appearance of culture as a unifying force in human activities, and finally the loss of the gods.[27] Two of these five characteristics are familiar from Foucault's characterization of the modern era. Foucault also writes about the appearance of science and the loss of the gods. By "loss of the gods," Heidegger, like Foucault, is not asserting that modern people do not have any place in their lives for the gods. Rather, God is not a critical explanatory mechanism for the wonders of the world. "[T]he loss of

[26] Foucault, Michel, The Order of Things, p. 59.
[27] Heidegger, Martin, "The Age of the World Picture", p. 116-117.

the gods is so far from excluding religiosity that rather only through that loss is the relation to the gods changed into mere 'religious experience'. When this occurs, then the gods have fled."[28] Art and culture are beyond the scope of this paper; Heidegger's understanding of technology, on the other hand, is central to my project.

In "The Question Concerning Technology," Heidegger defines the term technology, not as the tools created by science as it is commonly defined, but rather as the mentality that causes such items to be created. To avoid confusion with the standard parlance use of the term "technology," I will use the term "technicity" to denote the Heideggarian concept. Technicity is the impetus for people to try to learn more about nature by challenging its limits. And by challenging its limits, people learn how to mold nature into meeting an end defined by the system in which the persons in question operate. In this way, science and technicity are intimately related. Technicity drives science; science produces new tools with which to expand technicity. An example that Heidegger uses is the airplane on an airport runway. The airplane itself is a piece of technology, and it is a cog in the technicity of transportation. And the technicity of transportation allows us to be wherever we want whenever we want, and, more importantly, wherever and whenever it is demanded by the systems in which we live. The people in the system are not in control of the system itself; rather they are cogs within the technicity in which they participate. The hallmark of modern technicity is that the objects being considered by the technicity are shaped into a form that allows them to be flexible enough to be a useful resource in multiple circumstances but stable enough to be only that which is needed of it in that given circumstance; in the modern era, the being or essence of objects, including people, end up being placed in the standing-reserve, the

[28] Ibid, p. 117.

conceptual sphere in which elements can be flexible and stable in this way. In the case of people, it is often the people themselves that voluntarily make themselves cogs. They are offered incentives, which can be as simple as a sense of normalcy, to be part of the standing-reserve. These incentives are regularly offered with the most benevolent of intentions; the reduction of being into the standing reserve is rarely done as part of an active cognitive process. Rather, each and every one of us, those with the power to reduce and those without that power, are cogs in the system. The modern era gives people a great deal of freedom, but when people come into contact with a system, such as society, they become what the society wants them to be. And that is the power of society, the ability to deprive people of their freedom of being.[29]

Heidegger and Foucault argue that Western civilization moves within its eras (Classical, Modern, etc.) until a series of watershed events drive it into a new epoch. Kuhn's argument is that modern science itself proceeds within a paradigm until crises drive it into a revolution. Copernicus' astronomical proposition neatly fits into both of these arguments. As previously stated, Kuhn uses that discovery as an example of a paradigm-changing event. Heidegger and Foucault could use that event as one of the many that brought us from the Middle Ages into the modern era. The idea that the Sun revolved around the Earth was deeply colored by the Christian understanding that God placed humanity at the center of all things in nature.[30] Thus, it was not a great stretch to believe that Earth, the home of humanity, was at the center of the physical universe, that the Sun and other celestial bodies would have to pay homage to the Earth by moving while the Earth stood still. The Sun revolved around the earth because that is what God

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[29] Heidegger, Martin, "The Question Concerning Technology".
[30] Huff, Toby, p. 9.

willed. The Earth revolved around the Sun because that is what science showed to be true.

Foucault examines the appearance of the modern disciplines of economics, rhetoric, and biology to fully develop his argument about modernity. Since it is more relevant to my argument, I will limit my comments to the development of biology. Foucault argues that life is a distinctly modern concept. The Classical era was populated by living beings.[31] The Classical equivalent of biology was natural history. The responsibility of the natural historian was to catalog nature, "to establish the great compilation of documents and signs – of everything, throughout the world, that might form a mark, as it were."[32] Zoos, herbariums, gardens, and other collections were the purview of natural historians. It was here that nature was displayed "stripped of all commentary, of all enveloping language."[33] It was here that "creatures presented themselves one beside another, their surfaces visible, grouped according to their common features, and thus already virtually analyzed, and bearers of nothing but their own individual names."[34]

The comparison inherent in classification necessitated a uniform means of recognizing signs. Visible signs, i.e. elements of structure, were given priority because of the seeming constancy of observation of sight over the other senses.[35] The search for more visible signs led to the advent of dissection and the birth of anatomy, the isolation and comparison of the parts of individuals in a given species. This transition was "the beginning of what, by substituting anatomy for classification, organism for structure,

[31] Foucault, Michel, The Order of Things, p. 128.
[32] Ibid, p. 130.
[33] Ibid, p. 131.
[34] Ibid, p. 131.

internal subordination for visible character, the series for tabulation, was to make possible the precipitation in the old flat world of animals and plants, engraved in black on white, a whole profound mass of time to which men were to give the renewed name of history."[36]

Natural history was undeniably a search for signs - the marks that distinguish the different elements of the natural world. At this point, it was not relevant why a given animal is structured the way that it is. The answer to "Why?" was always some sort of return to the divine. This was even true of the early conceptions of evolution, which, after Darwin, is a cornerstone of modern biology. Foucault writes the following about the system of evolution theorized by Charles Bonnet. "He implies in the first place that the chain of being, stretching up through an innumerable series of links towards the perfection of God, does not at present attain to it; that the distance between God and the least defective of his creatures is still infinite; and that across this, perhaps unbridgeable, distance the whole uninterrupted fabric of beings is ceaselessly advancing towards a greater perfection."[37] Thus, although the qualities of different species are now variable, they still vary in proportion with each other. A given species can be no better, in an absolute sense, than it was in the beginning. God's plan is perpetually preserved. Darwin's "*Origin of Species* recognized no goal set either by God or nature" making Darwin a revolutionary figure.[38] Hence, with reference only to structure, natural historians were able to organize the world so that it was in line with God's plan. However, natural history was still just taxonomy. Natural history investigates and catalogs these living beings; it never explains.

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[35] Ibid, p. 132-3.
[36] Ibid, p. 137-8.
[37] Ibid, p. 151.
[38] Kuhn, Thomas, p. 171-2.

The generation of taxonomies forced the recognition that there are elements that are similar between species just as there are elements that distinguish one species from another. These similar elements formed the basis for investigation into function. Since they were so frequent, they must be essential for every species. Characteristics (signs) are linked to function; the answer to "Why?" is no longer because God deemed it so. Rather the answer to "Why?" becomes because it is essential for members of that species to live. Life becomes the organizing concept that unites the function of one characteristic of a species to the function of a characteristic that is not adjacent in the body. Life unites the hoof with the stomach with the lung with the heart.[39] Life unites the organism.[40]

Function also arrives as a new way to establish taxonomy. In a purely morphological taxonomy, two characteristics that are visibly dissimilar would be used to separate the owners of those characteristics. In a functional taxonomy, two visibly dissimilar characteristics can unite the relevant species if those characteristics are hypothetically available for the same function.[41] For example, gills and lungs are morphologically distinct. The former is a water filter, whereas the latter is a bellows. However, both organs serve the same function – gas exchange - so they could be used to place a fish next to a mammal. This establishes another layer of variability in the signs of a given species.

Life is the only concept that is able to define biology. In that sense, the appearance of function revolutionizes natural history.

> The importance of this upheaval can be appreciated; in the Classical period, natural beings formed a continuous totality because they were beings and because there was no reason for any interruption in their deployment...living beings,

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[39] Foucault, Michel, The Order of Things, p. 226-229.
[40] Ibid, p. 266.
[41] Ibid, p. 263-270.

> because they are alive, can no longer form a tissue of progressive and graduated differences; they must group themselves around nuclei of coherence which are totally distinct from one another, and which are like so many different plans for the maintenance of life…But since the discontinuities must be explained by the maintenance of life and its conditions, we see the emergence of an unexpected continuity – or at least a play of as yet unanalysed interactions – between the organism and that which enables it to live.[42]

Natural history, the enumeration of nature, included non-living items, like rocks, in its directories. Now that there is a concept that can distinguish between the living and the non-living, biology, the study of life, as a discipline can be separated out of natural history. Life is the organizing value in modern biology. Hence, all that diminishes life is implicitly anti-biological. Life provides the dividing lines that establish physiology. Life establishes the normal and the pathological.

Medical Science

Modern medicine owes its conceptual framework to the development of biology and the appearance of life on the intellectual scene. After all, how can a physician attempt to use science to preserve life if there is no concept of life, i.e. function, in science? When the organizing concept of life did not exist, physicians could only treat symptoms using personal experience, access to the divine, or some other organizing concept as a guide. This concept is explored in more detail below. Modern medicine developed at about the same time and along a similar path as modern biology. Just as with natural history, medical science is deeply concerned with classifying disease based on the signs of that disease. For example, some signs of left-sided heart failure, a disease in which the heart fails to pump a sufficient amount of oxygenated blood to the rest of the body, are diaphoresis, tachycardia, tachypnea, and pulmonary rales, whereas some signs

[42] Ibid, p. 272-274.

of right-sided heart failure, a disease in which the heart fails to pump a sufficient amount of de-oxygenated blood to the lungs, are jugular venous distention, hepatomegaly, and peripheral edema.[43] Though both diseases come under the category of heart failure, a functional classification, they are separate entities based on the observable differences in people who suffer from the disease.

Symptoms are signs as perceived by the patient. Tachycardia is perceived as a racing heart. Tachypnea is perceived as rapid breathing. Diaphoresis is perceived as excessive sweating. It is important to note that symptoms are always some sort of alteration of function. In their usual functioning, hearts do not race, lungs do not demand to be filled and emptied rapidly, and skin does not mandate being kept moist. Furthermore, symptoms are un-modern in the sense that they do not necessarily have a referent in the body of knowledge. When I am ill, I know that I am ill without having to apply some sort of scientific logic to fit my condition into my understanding of the world. I simply feel it to be true. My recognition that I am ill stands on its own. This is why pre-modern physicians were able to only treat symptoms prior to the development of a medical science; symptoms existed in the pre-modern era. As previously argued, the assumption that living beings function in the pursuit and preservation of life is the key to the development of that science. Until life appears as a concept, a fever is just a fever, and, since it is uncomfortable, it is worth trying to eliminate it. Medical signs, on the other hand, are defined in relation to the corpus of medical knowledge. Tachypnea, for example, is defined as more than twenty respirations per minute and tachycardia is defined as more than one hundred beats per minute. Symptoms are not necessarily

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[43] Frankel, Stephen K. and Fifer, Michael A., p. 208.

defined by God, even if some people may attribute illness to the work of a higher power. As such, symptoms cannot be confined to the Classical period; God, rather than the corpus of science, is the external referent for Classical signs, illness related or otherwise. It is my assumption that both modern and classical people experienced being ill in the same fashion and that is a fashion that is not necessarily Classical or modern. It doesn't matter what concept we use to explain the illness. The point is that a Classical person and a modern person both feel sick in the same way. This is why I have chosen the term "un-modern" to describe symptoms. Symptoms are dysfunctions, modern concepts, perceived in an un-modern manner.

Just as with the appearance of biology, the search for more precise and duplicatable signs for diseases led to the reliance on sight as the most precise sense and the use of dissection to develop medical science. According to Foucault, the clinical gaze arose as just another botanist's gaze in the seventeenth and eighteenth centuries. However, at the beginning of the nineteenth century, clinicians began to use sensibilities gleaned from chemistry to explain what they were seeing.[44] The clinical gaze sought out "not the remains of morbid consumption…but the species of the combustion."[45] The gaze explores the body, both inside and outside, in a thorough search to find the structural defect that explains the signs of the disease.

The gaze is also what allowed the development of all diagnostic tools. From the practice of percussion to the otoscope to the MRI machine, diagnostic tools exist only to help the physician to find the lesion with more precision by manipulating some rule of nature. MRI machines take advantage of the fact that hydrogen atoms vibrate when

[44] Foucault, Michel, The Birth of the Clinic, p. 107-120.
[45] Ibid, p. 120.

exposed to electromagnetic radiation at a certain frequency and that they vibrate differently depending on the atoms that surround them in the mass being examined. Otoscopes take advantage of the physics principles of optics to make small objects appear large when viewed through lenses. Percussion takes advantage of the fact that air-filled cavities will resonate when they are struck with the flat pad of the finger while more solid cavities will sound more dull when struck in the same manner. By challenging the body, modern physician-scientists can ascertain the essential characteristics of the body before them. Physicians are part of the technicity of medicine – the clinical gaze.

The gaze necessitated the dissection of human corpses, a revolution in medical science. The problem that prompted the revolution was the need to literally see what disease did to the body. The norm being criticized in this situation was an explicitly religious belief saying that the dead had no knowledge to offer the living.

> The day it was admitted that lesions explained symptoms, and that the clinic was founded on pathological anatomy, it became necessary to invoke a transfigured history, in which the opening up of corpses, at least in the name of scientific requirements, preceded a finally positive observation of patients; the need to know the dead must already have existed when the concern to understand the living appeared. So a dismal conjuration of dissection, an anatomical church militant and suffering, whose hidden spirit made the clinic possible before itself surfacing into the regular, authorized, diurnal practice of autopsy, was imagined out of nothing.[46]

The first pathologists questioned the pervading belief that the dead had nothing to tell us, a belief couched in a religious taboo against defiling the body once the soul had left it. The first pathologists proved that "knowledge spins where once larva formed."[47] Pathology presents itself as a new means of classifying the world of illness, a means that unites structure, the sign-lesion continuum, and function, the set of symptoms

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[46] Ibid, p. 126.
[47] Ibid, p. 125.

experienced by the patient. It presents itself as a pure science of disease, a modern means of understanding the illness. "If there is an axiom in medicine it is certainly the proposition that there is no disease without a seat."[48]

The gaze establishes a paradigm for medical science, a paradigm I will call medical constitutionalism. By inscribing the disease on the body of the person with the illness, the clinical gaze focuses the vision of medical science on the body and blinds it to other objects in the world. Disease always creates a constitutional change; this change is essential to the diagnosis of the disease. A second tenet of medical constitutionalism, the heart of the tension in the paradigm, is that people, and the diseases that are inscribed upon them, are similar enough to study as a group. This is implied by the process of determining the signs of a disease, a process that starts with a number of people experiencing the same symptoms and bringing those symptoms to the attention of the medical establishment; people with symptoms focus the clinical gaze on the illness allowing the development of a disease from the illnesses. This process is discussed elsewhere in this Part. However, in order to explain individual variations in the experience of illness, the paradigm needs to incorporate the idea that people are sufficiently different to explain why a given exposure will create disease, i.e. the signs of disease, in one person but will spare a different person from that fate, into this second tenet. This second tenet is discussed in further detail in Part IV.

Medicine is very much like the biology of humans. However, in one very important way, medicine varies from all other subsets of biology. Medicine is an explicitly normative enterprise. Whereas biology, like other pure sciences, grew out of a history of observing that which exists, medicine grew out of a history of trying to make

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[48] Ibid, p. 140.

the patient feel better, a history of resolving symptoms.[49] A biologist in the field would not dream of intentionally forcing the system being observed to conform to a preconceived notion of what that system should be; if this does occur, as a result of technicity as Heidegger would argue, it is an unintentional process. Forced conformation of the body into the technicity of health is the primary function of the physician-scientist. For example, assume the existence of a biologist who had been trained to believe that true nature should have no polar bears. Then assume that that biologist is released into the Arctic portion of North America, where polar bears are quite common. I would hope that that biologist would take his or her first encounter with a polar bear as a moment of curiosity worthy of further investigation, an anomaly that could prompt a revolution if it could not be explained under the extant paradigm, rather than take that encounter as some sort of affront to biology and use every weapon available to make the polar bear extinct. Some would argue that the disturbance of a natural system being observed need not be so blatant; the very act of observation could change the system. As previously implied, Heidegger would even argue that there is no such thing as a pure science because scientists in all disciplines are forced to understand the world through the filter of technicity. These objections are beside the point. My argument is that medicine, specifically evidence based medicine, is a much more impure science than any other endeavor of technicity. Physician-scientists are trained both to observe the patient and to obliterate the disease. Physician-scientists don't just study life; they preserve it, for better or for worse. This is what is meant by the statement medicine is explicitly normative. Each pathological entity has tied to it an explicit wish to be eliminated. Otherwise it

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[49] Foucault's The Birth of the Clinic makes it very clear that the healing arts of old gave way to the science of modern medicine.

would not be pathology. As Foucault writes in his introduction to Canguilhem's The Normal and the Pathological,

> One had been able to believe around the time of Bichat that between a physiology studying the phenomena of life and a pathology dedicated to the analysis of diseases, one was finally about to disentangle what had remained confused for a long time in the mind of those who were studying the human body in order to "cure" it; and that having thus been freed from every immediate care of practice and every value judgement as to the good and evil functioning of the organism, one was finally going to be able to develop a pure and rigorous "science of life." But it proved impossible to make up a science of the living being without having taken into account, as essential to its object, the possibility of disease, death, monstrosity, anomaly, error... Hence a paradoxical fact in the life sciences: it is that if the "scientificization" process is done by bringing to light physical and chemical mechanisms, by the constitution of domains such as the chemistry of cells and molecules or such as biophysics, by the utilization of mathematical models, etc., it has on the other hand, been able to develop only insofar as the problem of the specificity of life and of the threshold it marks among all natural beings was continually thrown back as a challenge[7]... [I]t has had and undoubtedly still has an essential role as an "indicator" in the history of biology. And this in two respects: as a theoretical indicator of problems to be solved...as a critical indicator of reductions to be avoided.[50]

Medical science specifically cannot be constituted as pure science. Physical health, i.e. good functioning, is at its heart – medical constitutionalism. To apply chemistry or physics to living beings is to lose sight of life itself, and yet these are the only tools available to try to answer the question of life. To lose sight of this concept is to delude oneself into believing that there is no normative value in life. By adhering to the value of life, physician-scientists trap themselves in the normative enterprise that is medicine.

Medicine has within it two norms of health – a personal norm and a population norm. The personal norm of health fits the scenario in which the patient feels that he or she is ill. The patient knows that something is different and he or she would rather that his or her body was returned to its previous state of being; that is all there is to know.

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[50] Foucault in Canguilhem, Georges, 1989, p. 17-18. The footnote number 7 in the quotation refers to: Canguilhem, Georges, 1968, p. 239.

That state of being is the abnormal state of being ill; it is defined without referent in some body of knowledge. It is only a feeling. The personal norm of health is the state of the patient when he or she is not ill. Normal is defined in strict opposition to the abnormal state of being diseased.[51] Well-being does not exist without the state of being ill. It should be noted that a number of definitions of health, including the World Health Organizations pronouncement that "health is a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity",[52] attempt to define health without first defining disease. These definitions are meaningless because they cannot effectively define a lower limit to the well-being sought absent a state of being ill. The definition of well-being can be moved by any arbitrary whim, proving my point below about the arbitrariness of the term "health". For example, we, in the West, would not know that we live a life of social well-being if we could not witness the lack of social well-being in other countries or in other parts of our world, at least from our perspective. Thus a consideration for the lack of well-being, or the state of being ill, is implicit in any such definition. In the argument to establish health, disease is needed to define with certainty the borders of health. Even the call to species-typical functioning excludes the ill from the basis of the species. Otherwise, a decrease in life would be considered normal; that would make the concept of disease meaningless. Additionally, in reference to the World Health Organization definition specifically, this definition includes goals that are not commonly encountered in medical science and is therefore beyond the immediate scope of this paper; certainly, my arguments can be expanded and applied to the other norms that drive the search for and preservation of health, however it is defined.

[51] Canguilhem, Georges, 1989, p. 284.

The bottom line is that there is no such thing as health without such a thing as disease; these two concepts are two sides of the same coin.

As previously stated, illness is an un-modern construct, and, therefore, it is an arbitrary and unquestionable construct from a modern perspective. Disease is the conversion of illness into science (modernity), but because it relies on illness at its basis, it is just as arbitrary. Health makes itself out to be a modern construct. Since health is necessarily defined in opposition to an arbitrary and unquestionable construct, it is, in a sense, arbitrary and unquestionable. People can know that they are well just as well as they can know that they are ill; the personal norm of health is personal and un-modern.

As it guides the paradigm of medical constitutionalism, the population norm of health masquerades as a purely modern construct and that it is equivalent to any other norm in biology. However, given the mode of its derivation, it must be considered just as arbitrary as the personal norm of health. In order to derive a population norm of health, group of normal people are brought together and evaluated for similarities. These similarities are cataloged and defined as normal life signs.[53] A normal heart rate is between sixty and one hundred beats per minute. A normal respiratory rate is between eight and twenty breaths per minute. A normal fraction of CO_2 in exhaled air is between thirty-six and forty parts per million. All of these values were derived in this manner via mathematical and statistical manipulations. It is important to note that the population norm does not escape reliance on the personal norm. Normal people are those who define themselves to be not sick; they are people who satisfy the personal norm of health.

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[52] Constitution of the World Health Organization (preamble). *The First Ten Years of the World Health Organization* (Geneva: World Health Organization, 1958). Cited in Engelhardt, Tristram, p. 206.
[53] Canguilhem, Georges, 1989, p. 243-246.

They are the personally normal. It could be argued that these study subjects are routinely evaluated against population norms to prove that they are not diseased. This is a circular argument. The population norm had to be generated at some point. If it was not generated by taking the personal health status of the subjects into consideration, it is an arbitrary construction and continues to prove my point.

On a more individualized level, the system of medical education relies on the transmission of witnessed normal and abnormal forms from generation to generation. One of the critical elements of medical education is the practical experience garnered under the supervision of more experienced practitioners. This is how a budding physician-scientist learns to distinguish, for example, a murmur from a normal heart sound or a hyperreflexive joint from hyporeflexive joint. This process is guided by a person who was trained in a similar manner, guided by some other experienced mentor, and so on. In that way, the population norm is ingrained into the consciousness of every practitioner of medical science.

The population norm of health presents a second concern. The numerical values that populate the population norms come from averages of individual values measured in healthy people. The range of 36-40 ppm for exhaled CO_2 is a statistically valid range that comes from a data set of a multitude of values, many of which will fall within that range. However, a few values of normal individuals will not fall within that range; they are accounted for in the statistical manipulation of the data. Therein lies the concern. The population norm will necessarily define people who consider themselves healthy in a personal sense as unhealthy in a population sense. The population norm of health is thereby incompatible with the individual norm of health for a given parameter of health for some people. As previously argued, the individual norm of health is a necessary precursor to the population norm of health and is itself closer to the static condition of being ill. That condition of being ill is a critical component of each individual's understanding of life. Therefore, it could be argued that the population norm of health does a disservice to the scientific understanding of the human being. The act of reducing the individual into the average and the reapplication of that average onto the individual ensures that the population norm loses sight of life. It forces us to understand only those aspects of life that fit within some ideal range of values or the set of experiences of the physician-scientist in training, the aspects of life that are allowed by medical constitutionalism. The idealized and impossible paragons of the species are the only true life in medical science; all other persons examined by medicine are only varying distances from death.

The population norm also operates to shift people into the "standing reserve" as described by Heidegger. Normal people fit into society in a number of different ways. They are all cogs, but they are all different cogs. In the sense of the personal norm of health, to be ill is to not be able to do what one normally does. It is to be dysfunctional, but this dysfunction is in relation to one's previous level of functioning. That level of functioning could be in line with one's status as a cog in society or it could be completely outside of the needs of society. The critical point is that the definition of normal is still within the individual's choice. The individual could theoretically choose to be sick from a population perspective by redefining illness from a personal perspective; the individual, if he or she was sufficiently aware of his or her circumstances, could choose not to be a functional cog. Through the population norm of health, medicine exists to create perfect cogs - cogs that can fit in anywhere in society, a one size fits all person for society. Because there is only one population norm of health, medicine can only create one type of cog. As previously argued, the population norm eliminates individual variation from the definition of health. There is a population-wide standard and anyone who does not fit into that standard is unhealthy by definition. People who fit the population norm are ideally able to do anything that any one person in the population can do; the paragon of health is the perfect cog. Medical technicity uses the population norm to define the health of any given individual. The body is challenged by medical science and normalized to the population norm. People with high blood pressure, defined as greater than 140 mmHg systolic and 90 mmHg diastolic, are given any number of medicines and recommendations for behavior modification to get their hypertension back into the acceptable range, defined as less than 120 mmHg systolic and 80 mmHg diastolic depending on the presence of other medical abnormalities, because other elements of medical science say that these patients are at risk of acquiring a cerebrovascular accident or a myocardial infarction. There is no illness of hypertension per se since hypertension rarely produces symptoms; yet the signs of hypertension dictate the normalization of the patient's body. In this manner, medicine limits the freedom of people just as is true with any technicity, assuming that the patient chooses to cooperate with the directions of the physician.

Ethical Decision Making

This conflict between the un-modern and the modern can be phrased in a slightly different manner using the language of beneficence and autonomy. When a physician recommends a treatment, he or she does so out of concern for the health of the patient. This is the principle of beneficence; the physician recommends what is in the best health interest of the patient. The physician who practices evidence based medicine speaks from the perspective of population health. This physician acts to normalize the patient. The patient counters the physician's recommendation by resorting to the claim that he or she knows his or her body the best. This is the principle of autonomy; the patient has enough freedom and understanding to know when there is something wrong in his or her body and therefore has the right to participate equally in any decisions regarding the treatment of his or her body. In the words of H. Tristram Engelhardt,

> Patients frequently choose to engage in behaviors that physicians and nurses know to be dangerous, possibly disabling, and in the end perhaps lethal. Out of respect for those persons, physicians and nurses must often, if not usually, tolerate noxious lifestyles or failures to comply with treatment. Yet physicians and nurses, in joining the health care profession, have committed themselves to achieving the best interests of patients. This tension can be appreciated as the conflict between two ethical principles: that of permission [autonomy] and that of beneficence. It is in terms of the contrast between these two principles that the moral tension felt in many choices regarding abortion, treatment compliance, or refusal of health care is to be understood.[54]

The patient speaks from the perspective of personal health. The patient acts to maximize his or her freedom. There is, however, no science protecting the autonomy of the patient; science cannot be used to maximize freedom because freedom, in this case, is personal and un-modern. If science backed the will of the patient, then the physician, as the ambassador of medical science, would have to support that decision; there would be no conflict, so there would be no ethical dilemma. However, in times of conflict, the physician resorts to the definitions of being human established by medical science. Moreover,

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[54] Engelhardt, Tristram, p. 103.

> the more the characterization of *the* moral fabric is tied to the very enterprise of being a person, the more firmly it can be generally justified. This is the case, for in asking a question about morals as a philosophical question, one is seeking a rational reply that is, as far as possible, inescapable. One is seeking a clincher to a dispute concerning which of the possible ways one can live life…one *ought* to choose, where the sanction for violating the "ought" is not a threat of force or feeling of guilt, but irrationality, worthiness of blame, or the failure to realize the goods one wishes to achieve.[55]

Many ethical dilemmas come down to the question of who knows how to achieve health best, the physician or the patient; these points of view have to be mutually exclusive. Otherwise, a compromise could be reached and there would be no dilemma. Thus, many ethical dilemmas can be understood in terms of a modern scientific understanding of the world versus an un-modern understanding of the world. This conceptualization presents an intriguing reversal of the characterization of the physician as a priest of health. If the physician offers a medical directive without a reasoned and rational explanation, the reason for following the directive might as well be a resort to the divine, the same reason for following a priest's directive. This is an argument without referent in the realm of science. However, once patients are allowed to question, allowed to assert autonomy and act on an un-modern understanding of their own bodies, the argument without referent is presented by the patient. The countering point of view, that of the physician, is presented with the support of the corpus of modern medical knowledge. And, as previously stated, the consequence for defying the corpus of modern medical knowledge is the label of irrationality, a label that the patient may accept if the reason for being irrational and defending the patient's position is compelling enough for the patient.

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[55] Ibid, p. 104-5.

Revolutionary potential

Foucault opens The Order of Things with an assertion about medicine implying that medicine is a purely modern endeavor. While it is true that the scientific aspects of medicine are modern in their character, those scientific aspects are predicated on an un-modern understanding of illness. People still need to experience symptoms to bring disease into the realm of medical science, to direct the clinical gaze. Since this un-modern perspective persists, medicine cannot be considered a pure science; it is not purely modern. Medicine can only use the population norm to supercede and erase the personal norm. This is a futile effort because the personal norm is critical to the establishment of the population norm and achieving the personal norm of health is more likely to be the clinical objective of the patient. In biology, there is a place for monstrosity, anomaly, and error. That place is defined by theories of evolution and the differentiation of species, elements of knowledge in the science of biology. Monstrosity, anomaly, and error are simply observed and preserved by biologists. Monstrosity, anomaly, and error are available to produce crisis and thereby initiate revolutions. There is no such place in medicine. Physicians try to eliminate monstrosity, anomaly, and error. As long as there is this vision of health presumptively defining certain states of being as disease and other states of being as health, medicine will not be a pure science; medicine will not see revolutionary possibilities in the anomalies it encounters. Embracing the modern perspective, embracing signs rather than the symptoms that establish their normativity, is the way to allow monstrosity, anomaly, and error to return into the science of medicine; it is the way to purify science. The modern perspective allows us to discover functional possibilities in modern disease. This is the revolutionary potential of genomic medicine, explored in more detail in Part IV.

Part IV - Genetic Revolution

> The sequencing of the human genome is only the tip of the iceberg. It is the beginning of a revolution that many predict will transform medicine. How will genetics research affect physicians and patients and the practice of medicine? When investigators identify the function and association of human genes with common chronic diseases, diagnosis, treatment, and classification of human diseases will be changed forever. Genetic susceptibility testing allows patients to know their predisposition to disease long before symptoms appear. Physicians can intervene with customized advice so that the patient can prevent, modify, or avoid the predisposed condition by better understanding both his or her genetic and environmental risk for disease. The promise of a genetic approach to drug therapy involves moving from one size fits all to personalized medicine tailored to the individual patient. Physicians will become mentors and counselors, advising patients on the best treatment path given their unique genetic predisposition – even in this sophisticated, high tech field, the physician-patient relationship is likely to improve, highlighted by individual therapies and personal attention.[56]

This quotation opens a piece on e-health in the new millennium in a periodical for professionals involved in health administration; it is typical of the grand potential regularly attributed to genomic medicine, with its elimination of symptoms, customized advice, personalized medicine, and the transition of the physicians to mentors/counselors. As the quotation says, sequencing the human genome, a task that will soon be completed, is only the beginning. According to the genetic prophets, before long, physicians will be able to inform patients while they are still infants about every disease they may incur and make it so that they will never have to suffer the symptoms of any constitutional disease. This is the genomic revolution in medicine, the subject of inquiry for this Part of the essay. Before entering that realm, however, a common confusion between genetic medicine and genomic medicine needs to be resolved. Doing so is essential to characterize the borders and components of genomic medicine, the second component of this Part of the essay. Finally, the issue of ethical decision making in medicine that was raised at the end of preceding Part is brought to a conclusion.

Genetic Revolution in Medicine

Drs. Alan Guttmacher and Francis Collins draw the distinction between genetic medicine and genomic medicine:

> [F]or decades knowledge of genetics has had a large role in the health care of a few patients and a small role in the health care of many … If genetics has been misunderstood, genomics is even more mysterious – what, exactly, is the difference? Genetics is the study of single genes and their effects. "Genomics" … is the study not just of single genes, but of the functions and interactions of all the genes in the genome. Genomics has a broader and more ambitious reach than does genetics. The science of genomics rests on direct experimental access to the entire genome and applies to common conditions…These common disorders are also all due to the interactions of multiple genes and environmental disorders. Genetic variations in these disorders may have a protective or a pathologic role in the expression of diseases.[57]

Genetic medicine is already here, even though it may not be manifest to many of us. It is what allows physician-scientists to diagnose and manage diseases like cystic fibrosis, sickle cell disease, and Marfan's syndrome.[58] For the most part, genetic medicine deals with conditions that are monogenic in their causes, i.e. there is one specific sequence of DNA that is in error and that error causes other molecular level defects. These molecular level defects include the dysfunctional transmembrane regulator in cystic fibrosis, the dysfunctional hemoglobin protein in sickle cell disease, and the dysfunctional fibrillin protein produced by fibroblasts in Marfan's syndrome. For the most part these diseases are rare. In addition to diseases, genetic medicine also reveals risks for diseases. Example are the BRCA1 and BRCA2 mutations and breast cancer.[59] A handful of specific DNA sequences have been epidemiologically linked to common diseases. It is

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[56] Bottles, Kent, p. 58.
[57] Guttmacher, Alan E. and Collins, Francis S., p. 1512.
[58] Pyeritz, Reed, p. 2761-2763.
[59] Guttmacher, Alan E. and Collins, Francis S., p. 1518.

widely believed that more DNA sequences will be linked to common diseases in the future. This is the way that genetic medicine plays a small role in the health care of many.

Genetic medicine is the quintessence of the medical constitutionalism paradigm. In the paradigm, to know the disease is to understand all of the constitutional changes that define the disease. With a purely genetic disease, like sickle cell disease, all of the constitutional changes are known. The faulty gene is known, therefore the faulty gene product can be produced. The gross effect of the faulty gene product is known because this is what produces the symptoms that calls the physician's attention to the condition. Indeed, genetic medicine is strengthening the place of medical constitutionalism as the dominant paradigm in medical science. Prof. M. Susan Lindee argues that genetic medicine has been at the heart of the medical paradigm since the early 1960's, entrenched by a series of changes in the various sub-disciplines within genetics.

> This was the idea that all human disease is a genetic phenomenon that can in theory be subject to technological, rather than social, control … The idea that all disease is fundamentally genetic operates and works across many different social and epistemological fields, and has become, at the dawn of the 21st century, one of the most important ways of understanding the frailties of the human body, embedding those frailties in evolution, molecular disorders, the notion of race, and the biotechnology industry…The notion of constitutional disease or constitutional susceptibility has a long history in medical thinking and clinical practice … The same diseases that had seemed constitutional in the 19th century – heart disease, cancer, mental illness, alcoholism, and infectious disease – began to be technically reconfigured as biochemical products of underlying genetic predisposition, which could in theory be specified down to the DNA base through genomic mapping.[60]

As physician-scientists become more proficient at drawing the link between a faulty gene product and a faulty gene or set of faulty genes, more and more diseases become genetic diseases. The constitutional changes are understood from their genesis in the gene to their termination in the gross pathology, with all of the relevant epigenetic factors included along the way. The paradigm is a very linear one: a pathological gene yields a pathological gene product which yields a pathological cell or set of cells which yields a pathological body which yields a pathological person, with a set of signs that can be used to diagnose the condition. At this point, it is critical to remember the discussion of the two norms of health in Part III. There would be no medical constitutionalism without a patient, a pathological person, coming to the physician in search of resolution of the symptoms of the pathology. And without a number of such cases, there would not be enough research material to define a condition in the first place.

There are two potential revolutions in genetic medicine, but while each potential revolution recognizes a shortcoming in medical constitutionalism, neither revolution produces a true criticism of the dominant paradigm, i.e. the new and the old paradigms are not mutually exclusive. The first such revolution is the advent of individualized medicine through pharmacogenetics and personalized medical advice. "[P]harmacogenetics … is based on observations of clinical efficacy and/or the safety and tolerability profile of a drug in individuals – the phenotype – and tests the hypothesis that interindividual differences in the observed response may be associated with the presence or absence of individual-specific biological markers that may allow prediction of individual drug response."[61] Pharmacogenetics recognizes the problem that modern medicine predicts a relatively uniform response to medicines by all people in theory, yet in practice, people respond differently to the same medicines. This is a true anomaly. Like genetic medicine, pharmacogenetics is already here. We have already uncovered

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[60] Lindee, M. Susan, p. 75-82.
[61] Lindpaintner, Klaus, p. 221-.

the genetics behind person-to-person differences in drug metabolism, especially regarding the P450 enzyme family.[62] Additionally, the drug trastuzamab (Herceptin®) is a monoclonal antibody designed to attack only those breast cancer cells with an active Her-2/neu oncogene in their genome.[63] It could be argued against this claim that there is a difference between the individual and the individual's tumor. That is very much my point. The literature on this matter lauds this approach as individualized medicine, but it is just the individualization of the tumors or other pathological findings. Pharmacogenetics does not meet the Kuhnian criteria for revolution. While it purports to be individualized medicine, which would be a true criticism of the medical constitutionalism tenet that patients are similar enough to be studied as a group, it is actually only a new means of stratification of the patient population, now along genetic lines, rather than along racial, gender, age, or other lines. Stratification is fully compatible with the second part of this tenet, which states that there are differences between individuals. Modification of the dominant paradigm allows the integration of pharmacogenetics into modern medicine; it will not revolutionize medicine.

The same argument applies to the personalized medical advice cited in the opening quotation. That personalized advice will be based on the collection of genes in the person's cells and the experiences of others with a similar set of genes. In the end, it is just a further stratification, just as the genetic analysis of pathology is just another stratification.

The second unsuccessful potential revolution was initially presented in Part II of this essay. Strohman argues that the paradigm of genetic determinism is insufficient for a

[62] Ibid.
[63] Ibid.

number of reasons. First, the "genome complexity found in humans and mice, for example, is not correlated with the differences of form and function found between them." Second, "[t]here is a striking lack of correspondence between genetic and evolutionary change." Third, the research approach in genetics makes "dubious assumptions concerning the similarity of genes and gene complexes in different species – between mice, fruit flies, and humans, for example, that are related to unique functions." Finally, a number of diseases are multifactorial, meaning that the single gene-single gene product-single disease paradigm elucidated above is insufficient.[64] Strohman concludes that the "theory of the gene is complete and wonderfully so; it is beautiful and magnificent in its utter simplicity…[but we] have been lulled into the reasoning that if the gene theory works on one level – from DNA to protein – it must work at all higher levels as well."[65] Strohman proposes that epigenetic factors, such as differential modification of DNA by methylation, differential splicing, and other intervening steps in the chain from gene to disease, have to play a role. The problems that Strohman elucidates and the paradigm he proposes are both very compelling. And, this set of ideas may constitute the grounds for a Kuhnian revolution in the genetic paradigm. But, his ideas are not at odds with the paradigm of medical constitutionalism. They are only at odds with the central dogma discussed in Part I. His revolution just adds a few more steps in the chain from gene to disease; disease still has changes in the constitution of the patient at its core.

Genomic Revolution in Medicine

[64] Strohman, Richard, p. 194-5.
[65] Ibid, p. 197.

As previously presented, genomic medicine differs from genetic medicine in that it takes the whole genome, rather than a single or a handful of genes, into account when investigating disease. Genomic medicine is the true genetic revolution in medicine, assuming that the genomic paradigm can be sufficiently validated to make it normal science. By truly individualizing medical science, it would undoubtedly resolve the conflict between individual variation and group characteristics. Non-genetic signs would be eliminated from the paradigm of medical constitutionalism, a process that is already underway as previously indicated.

Numerous barriers exist to the achievement of genomic medicine largely because of the complexity of the genotype to phenotype pathway.[66] Epigenetic factors, as previously discussed, add a number of intervening steps, and most of these steps are not well understood. The genetic-genomic conflation previously discussed will need to be avoided, meaning that it will not be enough to compare genes in model species to identical genes in humans. The entire genome of an individual will have to be investigated as a whole. Somehow, the predictive validity of that genome in its entirety will have to be established without piecemeal comparison to the experience of other genomes – a research model currently being proposed.[67] In genomic medicine, we can no longer predict the future based on the experience of the illnesses that came before. Those illnesses were unique, just as the illnesses that face physicians in the genomic present are unique. People will cling to the old paradigm; but, as argued in Part II, every paradigm needs its detractors to force its defenders to produce good work to validate the paradigm. Regardless, it seems to me that genomic medicine is a pipe dream, a search for the power

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[66] Scheuerle, Angela, p. 1204-9.
[67] Sander, Chris, p. 1977-8.

to augur with accuracy and scientific reason (or lack thereof) comparable to the Oracle at Delphi in ancient Greece. Every predictive methodology that purports to be scientific, from health care[68] to economics[69] to weather forecasting,[70] depends on using the past to predict the future. This methodology is completely entrenched. It will be very difficult, if not impossible, to validate the genomic paradigm. Fortunately, it is not my task to validate an alternative paradigm to medical constitutionalism, only to discuss the revolution that might occur because of the currently ongoing examination of the human genome. The remainder of this Part operates as a hypothetical inquiry: if the genomic paradigm is validated, then how would medicine be transformed? If we could change the body in a perfectly predictable manner, would medicine view the body differently?

The clinical encounter in the post-genomic world, the individualized and personalized encounter as discussed in the quotation that opened this Part, would be radically different from the current clinical encounter.

> Let us look at how an imaginary patient will benefit from this revolution. Shortly after a person is born, her genotype is recorded at her physician's office, and the information is transmitted to a secure database. Here, genotype means the presence or absence of specific variations in genes known to be relevant for assessing disease susceptibility and predicting responses to known drug types. Assisted by a decision support system, her physician may prescribe a personal immunization and screening schedule or recommend specific preventive measures. The genotyping information is complemented throughout her life by a screening program based on biomolecular profiling. At any point, screening may lead to recommendations about life-style or nutrition, or to detection of early stages of a disease. Refined diagnosis and choice of personalized therapy follow, which take into account her genotype and patient history and details of her molecular health profile.[71]

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[68] Dublin, Max, p. 175-207.
[69] Ibid, p. 22-27.
[70] Ibid, p. 21-22.
[71] Sander, Chris, p. 1977-8.

The clinical encounter would have a strong emphasis on prevention and early detection of disease through "prognostic genotyping and diagnostic molecular profiling."[72] Therapy would so personalized and so specialized that success is virtually guaranteed. If the genomic paradigm proves to be correct, the patient would never have to experience the symptoms of the disease; the patient would never have to suffer. Medical science would be able to make everyone and anyone a paragon of health, the perfect cog to fit in science's world picture. Medical science would be pure.

Physicians will have to develop a facility with genomics in order to accurately shepherd their patients through the world of lifelong health. Primary care physicians[73] and general practitioners[74] are best positioned do the brunt of the shepherding. Medical education will also have to change to reflect the understanding that all medical conditions, including trauma and infection, are genomic to varying degrees.

> [G]enetic education must prepare health care professionals and the public to manage uncertainty in the context of a health care system informed by genetic perspectives. Components of genetic literacy include new views of genetics and of disease, and attention to the nature of science, the principles of technology, and the ethical, legal, and social implications of genetic medicine. One of the consequences of genetic literacy in medicine will be to shift the focus of medicine from the name of a disease itself to genetic individuality, to the individuality of the experience, habits, and conditions of the environment of the particular patient.[75]

It is important to note that, even though this quotation refers explicitly to genetic medicine, it is more applicable to genomic medicine. That is clear from the last sentence of the quotation; the quotation mistakenly conflates genetic and genomic medicine, showing how commonly that conflation is made. In summary, medical education cannot

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72 Ibid.
73 Collins, Francis, p. 1285-6.
74 Kumar, S., p. 1992-3.
75 Cutter, Mary Ann, p. 255.

be based on collecting experience with disease without first learning what it means to understand individuals as they are.

The quotation in the previous paragraph also hints at the most dramatic change that comes about because of the genomic revolution: the elimination of disease as a named general entity if not as a state of being. The genomic revolution does so because it eliminates the impurity in medical science. As discussed in Part III, that impurity is the reliance on the patient to bring a set of symptoms to the attention of the medical establishment. Normal medical science, under the medical constitutionalism paradigm, then absorbs the symptoms as usable data, i.e. the symptoms are translated into signs, and uses these data to generate a disease category. For example, a patient may come to the physician concerned about unquenchable thirst and increased frequency of urination. The patient may also report that the urine smells sweet and that he or she has been feeling very tired recently. The physician will translate these symptoms into signs. Excessive drinking of water becomes polydipsia. Excessively frequent urination becomes polyuria. Feeling very tired becomes fatigue. A urinalysis will be performed to verify that there is too much glucose in the patient's urine, making it smell sweet. This will all make the physician suspect the disease entity diabetes mellitus, a verifiable object in medical science, and will perform an experiment, in this case a series of blood tests to measure the glucose concentration, to verify that the patient has diabetes mellitus. Each incident of verification of diabetes mellitus validates the applicability of the medical constitutionalism paradigm to the constellation of symptoms experienced in association with the condition. The patient does not have the disease diabetes mellitus until he or she comes into the view of the medical establishment; the physician gives the patient the disease. Conversely, there would be no disease if this patient, and others like him or her, never went to the doctor. Diabetes mellitus, as a verifiable entity in medical science, would not exist if people were not concerned enough about the constellation of symptoms associated with it, including excessive thirst, frequent urination, tiredness, and sweet smelling urine, to present those symptoms to physicians; there would only be the constellation of individual experiences.

This is what happens with all of the normal variations of human existence. People are, by and large, not concerned enough about the specificities of hair color to bring that issue to the attention of the medical establishment, therefore, hair color is not pathologized. Having red, brown, blonde, blue, purple, etc. hair is not a disease nor is having a buzzcut, a beehive, or any other coiffure; it is only a matter of fashion. However, enough people are concerned about baldness to bring that to the attention of medical science. Therefore, baldness, a matter of fashion, is also a disease – alopecia. By eliminating the impurity, i.e. symptoms, from medical science, both by defining conditions based exclusively on the genome (constitution) of the person in question and by keeping people from every fully suffering from symptoms, the genomic revolution forces medicine to rely solely on signs. This produces a "reverse medicine" where constitutional factors prompt the search for symptoms,[76] rather than vice versa as it is done now. However, with no symptoms, as defined under the old paradigm, to guide the clinical gaze to conditions worthy of being assigned signs, i.e. diseases, there is no way to draw the line between pathological and non-pathological experiences of life. There is no normal. There is no disease. There are only variations, and all variations are equally worthy of investigation by medical science. Clinicians are left without guidance from medical science about what should be treated, about what services they ought to be offering their patients. Every service and no service are both fair game. Purity comes at a great cost.

In light of this understanding, the clinical encounter described in the earlier quotation is not entirely correct. The quotation explicitly references diseases; in genomic medicine, there are no diseases, only variations. However, if the words "disease," "health," etc. were replaced with a more neutral terms, like "specific variant state of being," or deleted entirely the quotation would be rather accurate. Here is the quotation again with the appropriate changes.

> Let us look at how an imaginary patient will benefit from this revolution. Shortly after a person is born, her genotype is recorded at her physician's office, and the information is transmitted to a secure database. Here, genotype means the presence or absence of specific variations in genes known to be relevant for assessing [specific variant state of being] susceptibility and predicting responses to known [chemicals]. Assisted by a decision support system, her physician may prescribe a personal [intervention] and screening schedule or recommend specific…measures. The genotyping information is complemented throughout her life by a screening program based on biomolecular profiling. At any point, screening may lead to recommendations about life-style or nutrition, or to detection of early stages of a [specific variant state of being]. Refined [knowledge of her body] and choice of personalized [intervention] follow, which take into account her genotype and patient history and details of her molecular [variant state of being] profile.[77]

Without the normative terms, it is not clear how the physician should guide the patient. In modern medicine, physicians eliminate disease because they are linked to the suffering of symptoms. In genomic medicine, the question of how clinical decisions will be made remains open.

Clinical Decision Making in the Post-Genomic World

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[76] Kaplan, J., p. 658-661.

While disease as defined by medical constitutionalism ceases to exist as an entity in the genomic paradigm, there is no reason to believe that the set of diseases that are currently defined will not persist. Indeed, since genomic medicine will be born from modern medicine, this is a very likely scenario in the short term. However, diseases are experienced differently by different people, both in terms of symptomology and in terms of desire for termination of the disease. As such, reliance on the existing set of diseases, all defined by collective experience rather than by individual experience, is incompatible with the paradigm. As presented in Part II, Kuhn writes that elements that are incompatible with the paradigm tend to be ignored as problems fit for investigation by scientists working within the paradigm. Additionally, as previously presented, genomic medicine will be very successful in keeping people from ever truly suffering from an illness. Our knowledge of the genome will be so complete that we will be able to manipulate the constitution of anyone with extreme precision, so much so that there will be few side effects. As such, people will not really have the opportunity to personally experience physical suffering; pain will, for the most part, be relegated to storybooks. Diseases will go away. Else, they risk being static and arbitrary, just like the religious commandments of old when they are viewed through a modern world picture.

Since disease and the guidance offered by the population norm of health will disappear, the existing ethical framework used to evaluate the interchange between the doctor and the patient will have to change as well. As argued in Part III, beneficence, the ethical principle that guides the position of physicians, is supported by medical science and autonomy, the ethical principle that guides the position of patients, is supported by a very personal understanding of one's own condition. Beneficence has power because it

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[77] Sander, Chris, p. 1977-8.

represents what works for most people. Autonomy has power because it represents what works for the individual. A democratic dialogue between the doctor and the patient may bring these two perspectives to a compromise, meaning that there would be no ethical conflict, only a moral course of action that only the irrational would not accept. However, if there can be no compromise, one of these perspectives must win. Even though beneficence is supported by medical science, modern medical practice is such that the physician asks permission from the patient to compromise his or her autonomy through the course of treatment; this is the purpose of informed consent. The patient must voluntarily give up his or her autonomy in order for treatment to proceed.

In genomic medicine, there is only that which will work for the patient; there is no conflict between what works for most people and what works for the individual. Under such conditions, conflict can only arise in a situation that pits the politics of the physician against the politics of the patient, e.g. in differences between the definition of "working" in the minds of patients and physicians. Moral decision making stops being a conflict between beneficence and autonomy; it becomes a conflict between the physician's autonomy, his or her willingness to perform a procedure, and the patient's autonomy, his or her willingness to have the procedure performed. There are two states of affairs that present potential conflict: the patient wants a procedure the physician does not want to do and the physician wants to do a procedure the patient does not want. There need be no discussion of the other two scenarios (both the physician and the patient want the same thing and both the physician and the patient do not want the same thing) since there is no core conflict; a compromise is achievable.

It is important to emphasize, once again, that science cannot inform either side in these conflicts; science cannot guide either side to the truth. The final course of action in any one such conflict in the world of genomic medicine will be the result of the persuasive power of one agent or the other, not because one person's politics is more reflective of the truth than the other person's politics. It could be argued that the norm of life, and the science around that norm, might not be useful in resolving this conflict, but other norms, like evolution, would be more informative. However, resorting to evolution to reintroduce science into the conflict is not feasible because doing so assumes that evolution works toward some norm. As discussed in Part III, in the Classical era, that norm was the design of God; in the modern era, after the Darwinian revolution, that norm ceased to exist. Evolution is norm-free. What about the norm of life? As previously argued, the norm of life is the organizing value in modern biology and modern medicine. The modern understanding in evolution is that the life protected by evolution is the life of the species, not the life of the individual. This is incompatible with both the genomic paradigm's emphasis on individual lives and with the modern medical practice's desire to save individual lives.[78]

In the first potentially conflict filled state of affairs presented above, the patient wants a procedure that the physician does not believe would be a good idea for the patient. This is similar to a scenario presented by Beauchamp and McCullough regarding a 26-year-old very well informed and independent woman in excellent health who is concerned that having a child would interfere with her career goals. Therefore, she asks her gynecologist to surgically and irreversibly sterilize her by performing a tubal ligation. The gynecologist tries to persuade the woman to pursue other methods of birth control,

[78] Engelhardt, Tristan, p. 202.

largely because a tubal ligation is not easily reversible and there may be some day when the woman wants to have a child. The woman is not convinced because she values her career over having children and her career goals will keep her occupied through her child-bearing years. She has carefully considered the pros and cons and is convinced that she needs a permanent solution to the problem of her fertility.[79] In that scenario, the conflict is between the physician's belief that women should be able to bear children now or in the future, a belief that normalizes women into the societal role (or cog) of parent, and the patient's belief that childbearing would interfere with her career aspirations, a belief that normalizes herself into the societal role of worker. This is an interesting conflict because it deals with the dominant paradigm regarding gender roles. However, medical science does not take a stand supporting either norm. For medicine, being able to work and being fertile are both important values. Medical science can offer no guidance to either the physician or the patient; the arguments are made entirely from the political beliefs of the two parties. In the world of genomic medicine, the procedure sought in this scenario would be sterilization on the genetic or molecular level, rather than on the anatomical level. Nevertheless, the same issues are encountered. In the status quo and in the future, medical science can offer no guidance.

Genomic medicine offers the opportunity to create "monstrosity" at the patient's request. Assume that, with the genomic paradigm, physician-scientists learn how to regenerate organs. This is not unbelievable; if we know how to fix something at the molecular level, there is no reason to believe that we will not know how to make something at the molecular level. As that is the case, it is entirely possible that a person would ask for an extra heart, lung, liver, kidney, etc. An extra lung, for example, might

[79] Beauchamp and McCullough, p. 23-25.

be valuable for competitive runners or for people who enjoy diving. Regardless of the reason, the potentiality exists. A physician might balk at this request because it would make the patient into a monstrosity, an anomaly against the background of "normal" human beings. But, in the world of genomic medicine, there is no such thing as a normal human being. Disease cannot be used to define normalcy because disease does not exist. Evolution cannot be used to define normalcy because evolution deals with the species, not with the individual. Statistical measures cannot be used to define normalcy because the world of genomic medicine is a world of individuals, where population norms do not apply. Science cannot help the physician defend his or her position. This is a form of self-eugenics – the dramatic modification of individuals of the species above and beyond the gifts of their birth. It can be as dramatic as the example given above or as subtle as the correction of an inborn error of metabolism, like phenylketonuria or glucose-6-phosphate dehydrogenase deficiency, using gene therapy.[80] But, is this the sort of eugenics that should be avoided?

In the second potentially conflict filled scenario presented above the physician wants to perform a procedure on a patient who believes that that procedure is unnecessary. A reversal of the scenario presented by Beauchamp and McCullough would suffice as an example of this scenario taken from modern medicine. Here, the physician is advocating sterilization and the patient is advocating childbirth. Here, the physician is subverting dominant societal gender roles and the patient is a slave to modernity. However, assuming that the physician is not forcing this course of action on his or her patient, is there any reason to believe that there is an apolitical reason to support one

[80] Galton and Galton, p. 99-105.

position over the other? In my opinion, there is not. Again, neither party will receive guidance from science.

Genomic medicine offers the opportunity for more fundamental transformations. The physician could be advocating, for example, the transformation of a genetically deaf fetus into a hearing baby. The parents would take the other position – that they would prefer a deaf baby to a hearing one. They may even argue that performing the procedure to make a hearing baby out of a deaf one would be eugenic. This may seemed far-fetched, but it is a realistic possibility. Deaf[81] culture is alive and well in the United States. It developed from the intersection of the civil rights movements and the special needs of those who can only communicate in American Sign Language.[82] While the vast majority of people with deafness become deaf after being born hearing[83], being born deaf is a reasonably common occurrence. "Incidence of congenital severe hearing impairment is at least 1 in 1000 births, half of which can be attributed to genetic factors."[84] Given these facts, it is not unbelievable that a Deaf couple would see having a deaf baby, a child that could fit easily into their culture, as a boon rather than something that needs correction. Additionally, On March 31, 2002, the Washington Post published a story about a Deaf (and deaf) lesbian couple, both of whom had at least one deaf parent, who conceived a deaf baby by artificial insemination from a donation from a deaf man

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[81] It is a convention in the literature to spell Deaf with a capital D when referring to a person or persons who identify themselves with the Deaf culture or to Deaf culture itself and to spell deaf with a lower case d when referring to the condition of not being able to hear. That convention is followed in this paper.
[82] Lennard Davis presents a very complete history of the treatment of the deaf and the development of the Deaf in Enforcing Normalcy (1995).
[83] Luey, H. S., et al (1995) reports that "that only 22 percent [of the profoundly deaf population] (.22 percent of the whole [deaf] population) lost their hearing before age 19" and that "about 8 percent of all people have significant hearing loss."
[84] Tekin, M., et al.

because they wanted to raise the child Deaf.[85] From this incident, it is not a far leap to our scenario of a Deaf couple refusing a procedure to correct a deaf baby. As in the extra lung example, a modern physician may balk at the couple's position, in this case, since modern medical science reports deafness as a condition worthy of correction by medicine. Physicians interested in *genetic* medicine would make the same assertion.[86] *Genomic* medicine can offer no guidance.

In this case, however, there operate two other values – non-interference with subsequent generations and functionality in society rather than functionality in life. First, because genes are heritable, changes in the genome can be transmitted from generation to generation. This does not have to be the case. Even though the term genomic medicine conjures up images of gene therapy, genomic medicine is more about fully understanding the constitution of a given person. With that understanding, the interventions could be made at a higher, non-heritable, level with the same effect. Additionally, if DNA modification is necessary to achieve the desired effect, that modification need not necessarily be made in germ cells. Somatic cells form the bulk of our phenotype, so changes that are limited to somatic cells should be very effective at changing our phenotype. Nevertheless, heritable changes may be desired. In the words of Tristram Engelhardt,

> In the future our ability to constrain and manipulate human nature to follow the goals set by persons will increase. As we develop our capacities to engage in genetic engineering not only of somatic cells but of the human germline, we will be able to shape and fashion human nature in the image and likeness of goals chosen by human persons, not by nature or God. In the end, this may mean so radically changing human nature that our descendants may be regarded by subsequent taxonomists as a new species. If there is nothing sacred about human nature (and no merely secular argument can reveal the sacred), no reason will be

[85] Mundy, L.
[86] Reardon, W. and Mueller, R F., p. 319-321.

recognized as to why, with proper caution, human nature should not be radically changed.[87]

Genomic medicine offers the possibility to change future generations and the human species itself. As such, the question of the autonomy of those potential persons should be considered by clinicians and patients who operate under the genomic paradigm. Once again, science can offer no guidance; the consideration of future generations will be a part of the physician's and the patient's politics. If the party believes strongly in the change being sought, it may believe in it strongly enough to advocate it for their offspring as well. On the other hand, the facts that changes are only beneficial in the context of the world in which they are sought and that the world will rarely remain static combine to form a very compelling argument for limiting germ cell modification. We do not know what the future will look like, so we should not make permanent changes that will affect generations in the future, or so the argument goes. Then again, if genomic medicine truly understands the body of the individual, then it will understand the bodies of subsequent individuals well enough to reverse the so-called permanent change.

This ties in with the second point – functionality within society. Individuals do not live in a vacuum, even if individualism is prized by genomic medicine. Modern medicine treats disease. Disease is defined by a set of symptoms. Those symptoms are our understanding of our bodies not functioning as they should. If Heidegger's interpretation of modern life is correct, then people are driven by the systems in which they live to make themselves fully functional in the context of their dominant system. A large part of the system is society. Thus, diseases are dysfunctions in a societal context. So, when a physician treats a disease, he or she returns societal functionality to the

[87] Engelhard, Tristram, p. 413.

patient. Improvements beyond baseline are defined as eugenic, a term that harkens back to an era when the infringement of individual liberties in order to strengthen the human species was seriously contemplated in many corners of the globe.[88] Modern medicine is able to protect patients and physicians from accusations of eugenic behavior because it has the norm of health to divide the medically necessary from unnecessary improvement. Will the same be true of genomic medicine? This is the central question addressed by Sarah Mandel in her Masters Thesis. She concludes that the issue is much too complex to resort to eugenics to explain the morality of what could come from genomic medicine. The term "eugenics" itself is loaded with so much history and misunderstanding that its use does not make an argument; rather it only clouds the issues that are involved.[89] Mandel believes that disease will persist after the revolution.

A more complete characterization of the improvement/therapy continuum in genomic medicine can be derived from a return to the discussion of modernity presented in Part III. As previously contended, science and technicity are at the cornerstone of modernity. Science is a mode of inquiry that challenges nature in order to derive a better understanding of nature. Technicity is the mindset that allows these challenges and directs the inquiries of science. Each successful inquiry expands science's understanding of nature and expands our ability to shape and order nature to fit technicity. Additionally, each successful inquiry validates science as an effective means of understanding nature. Medicine fits in this scheme in that it is the element of modernity that fits people into technicity. Each successful inquiry into the body expands medical science's understanding of the body and expands our ability to shape and order the body to fit

[88] Galton, David J. and Galton, Claire J.
[89] Mandel, Sarah, p. 93-103

technicity, specifically the technicity of health. Finally, health is defined as being fully functional in society.

Genetic medicine is an extension of the current paradigm; it is a tremendously successful inquiry into the body. In genetic medicine, physician-scientists continue to mold people into perfect cogs for society by finding and eliminating disease. Moreover, because genetic medicine purports to truly understand the individual, the voice of the patient is silenced. Science says that the course of action offered by the doctor is well-designed for the patient's individual genetic makeup. The voice of beneficence becomes too loud to be ignored. Individual autonomy is coerced out of existence; modernity compromises our freedom.

Genomic medicine, on the other hand, is a true revolution; it purifies medical science. There is no human body in genomic medicine, only human bodies. There is no disease or health in genomic medicine, only variant modes of existence. The science and technicity of health are fundamentally transformed. Physicians can only facilitate transformations of the body; they cannot dictate them. In genomic medicine, the voice of beneficence is dampened by the theft of the self-styled objective (scientific) definition of health. The only control the physician has over the patient is through the exercise of his or her freedom to refuse to participate in a transformation of the body.

The eugenic danger in genomic medicine is not that the medical institutions will force people to be a certain way. Rather, the locus of normalization shifts out of the clinic and into the society in general in the world of genomic medicine. The norm that guides changes in our bodies will no longer be health, rather it will be success – functioning at the highest level possible. Market forces, or some other element of modern technicity, could very well drive people to be the paragons of health, i.e. perfectly functioning cogs, just as modern medicine drives people in that direction now. And with perfect science and technology, there would be perfect results. However, as indicated above, physicians and patients still have the choice of whether or not to include modernity in their politics. Physicians would be able to make everyone and anyone a paragon of health, the perfect cog to fit in modern technicity's world picture. But, only those physicians with the desire to do so, will do so. Neither medical science nor the markets can dictate this decision. Admittedly, there will be motivations to move in that direction - that is the nature of technicity – but resistance by rethinking technicity is far from futile.

Part V – Conclusion

The genomic revolution may never arrive. Genetic medicine may also be proven to be out of order. We may end up investing billions of dollars and an equally tremendous amount of effort into the investigation of the human genome with no results with the power to move the Earth. My ability to predict the future is not powerful enough to know where we will end up.

Nevertheless, the process will yield information of great value to modern medicine. After all, anomalies that do not revolutionize prove the validity of the paradigm. Regardless of our endpoint, the process will give physicians and patients much more power and control over the human body. And with that control comes new ways to escape the suffering and loss of freedom associated with disease.

The process of generating this inquiry reveals a critical lesson as well. The idea of norm-free medical science is most certainly a result of the genomic revolution, but it can also be a part of modern medicine. The central crisis in modern medicine is its lack of ability to reliably predict results for a given individual. Modern medicine can say that a certain exposure increases a person's risk of disease by a certain percent or that a certain proportion of people in a community have a given disease. It can even attribute a lifetime risk of incurring a disease for a human being. All of these are population based measures, and no matter how stratified that population is, population based measures cannot predict with absolute certainty the future of a single person. A patient will either have a disease or not; the patient either has a lifetime incidence of 100% or 0%.

This crisis is what allows a debate between the physician and the patient regarding the appropriate course of action in the face of a given bodily condition. In a sense, this debate is already a debate between the autonomy of the patient and the autonomy of the physician, as it would be in genomic medicine. The difference is that the physicians have science in their political arsenal. Since science is the dominant mode of comprehending the world in modern Western society, the use of the scientific argument is invariably compelling, just as the word from a priest in the Classical era would have been very compelling. It is hard to stand against something that compelling, but the joy of having a debate is having the opportunity to do so. Even though it is hard, it is still possible to argue that sterilization before having children is an appropriate course of action or that being deaf is a healthy way to be or that death is a preferable alternative to life. The technicity of health dictates a very definite way to be a perfect cog, and thereby reduces the freedom of humans as a whole. But, the practice of medicine preserves the freedom for individual humans to rebel against the dominant paradigm, to create meaningful revolutions in each of their own lives. Physicians should never forget that modern medical science presents that which works for most people in the form of what works for every person. Each instance of taking this intellectual step preserves the freedom of the patient to be an individual.

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