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Human Genetic Technology: a Call to Mutate a Reactive Society Into a Proactive Society

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**HUMAN GENETIC TECHNOLOGY:
A CALL TO MUTATE A REACTIVE SOCIETY INTO A PROACTIVE
SOCIETY**

**A thesis submitted to
Regis College
The Honors Program
in partial fulfillment of the requirements
for Graduation with Honors**

by

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Dedications

*This thesis is dedicated to my sister, father, and mother, and in loving memory of Auntie
Machiko and Rose Vu*

Introduction

Ever[1][2][3][4][5][6] since I was young I have often found myself fascinated with movies like Star Wars™ and Star Trek™. Adventures in the vastness and ever expanding area of space have always seemed larger than life. Compared to space, us humans and our lives, have often seemed so miniscule. Many people say our minds are as vast as the universe, but we seem so confined by our size. It was not until I was 15 years old when I learned more about genetics that I realized I was not giving humans enough grandeur. Inside of all of us 7 billion and counting people is a series of four molecules (A, T, C, and G) that can bind and arrange in almost infinitesimal ways to form the blueprint for all living things; our DNA containing our genes. Four letters that make up the around 20,000 protein-coding genes that interact, regulate, and work together in countless ways to make life happen. The collection of all of our genetic material in our body is known as the genome. What is even more striking is that 99.9% of our genome is identical throughout the human population, so even if that 0.1% seems small it has in itself infinite capabilities to facilitate the uniqueness we see around us everyday. Every aspect of our being is coded in these genes. About 50 years ago humans embarked on a journey as grand as those in Star Trek™. We began discovering that many things, most compellingly many sicknesses, have a genetic basis. About 20 years ago researchers began mapping the genome in hopes of understanding it more. They slowly realized that the genome is vast and ever mutating, just like space (Nelson 2001). Although this is exciting, we must also enter this era with caution and with boundaries. Just as the U.S.S. Enterprise did in Star Trek™, we must have a mission or an oath to follow as we embark on what could be our final frontier in understanding human life

itself. What is that mission? It has been highly debated and I can foresee that it will be for a very long time. Here I join in the debate.

My story begins before I was even born. It started in 1959 when the most common birth defect disorder was first characterized as having a genetic causation. Trisomy 21, more commonly known as Down's Syndrome, is an aneuploidy (extra or missing chromosome) in the 21st chromosome causing affected individuals to have, in this case, three chromosome 21 instead of two. This syndrome that had been characterized and known to affect thousands of newborns every year since the mid-1800's had finally been clearly identified (Hickey, & Summar 2012). Sadly, this was merely a baby step on the road to help those affected by the syndrome. As research progressed, before anyone knew it, 20 years had passed, and it was the 1970's and the best predictor available to help physicians with risk assessment was maternal age. Women above 35 had exponential increased risk of having children with Down's Syndrome. So women knew to be a little more cautious to have children at later ages, but this was not a high confidence predictor. Once the 1980's and 90's rolled around, researchers had narrowed the search further and found that within the amniotic fluid of mothers carrying affected fetuses there would be a higher amount of the serum protein AFP (Canick 2012). Little did they know, this high-confidence diagnostic test would be the springboard that would set many in society at odds. Back then and still currently there are two choices when a fetus is diagnosed with Down's syndrome: 1) continuing the pregnancy or 2) terminating the pregnancy (Bianchi 2012). Much of society was and is still split into individuals who are excited supporters/advocates and individuals who are opponents/nervous skeptics of genetic technology and diagnostic tools. Even those people who would normally choose to not involve themselves in scientific issues would not

be able to avoid the far reaching effects of genetics. The 1990's was just the beginning of the development of the technology.

My mother was one of those people, who before this time was not particularly interested in the issue of genetics. My mother became pregnant with my older sister in 1987 at the age of 32. Although her older maternal age put my sister at a slightly higher risk of Down's Syndrome, her doctors chose not to confront her about it. Then in 1992 my mother became pregnant with me at the age of 37. Due to her age, her obstetrician/gynecologist suggested that she consider to have an amniocentesis test to determine whether or not I would have trisomy 21. My mother is not one of those people who are unreasonably resistant to anything they do not know about, but she does take into account her family in any decision she makes. My mother asked to learn more about the procedure and was shocked by how invasive it sounded. On top of that her options were very limited, she would have the choices of terminating the pregnancy or having the baby regardless of test results. My mother opted not to take the test because she said either way she was going to have the child. When I heard this story years later I asked her what her reasons were. She said that first and foremost she did not care whether or not I was born disabled because her personal philosophy of being a mother is that she would be responsible for her children their whole lives no matter if they are capable of living on their own at adulthood; the difference in the amount of care she was willing to do was out of the question. In addition she is of a catholic background and she is devout in the thought that terminating pregnancies is never an option.

Some might say that it was malicious of her to risk bringing a child into the world that would be handicapped the rest of his life and that she should not have let her catholic beliefs play a role in her decision-making process. Also that if she allowed for a prenatal genetic test and it

came up positive, she would have had much more time to seek counseling and help in planning to raise a child with Down's Syndrome. I concede that some of these points are valid, but absolutely refuse to concede that my mother had any malicious intent. First, my mother had very limited knowledge of Down's Syndrome; so how was she supposed to take that into account while family planning? Also she did not plan to have children at a later age, but she just knew she wanted kids and her life did not provide her the opportunity before this point in time. She took into consideration that she was in a marriage with a man that would love and take care of her and their children, and took into account their assets to assure that she could support and provide a comfortable life to her children. My sister and I had the best childhood anyone could ask for. My mother's choice has always resounded as a decision that was both truly ethical and moral, and illustrated the ultimate statement of love.

Then this perspective was complicated when I was 19 years old. My sister, who was in medical school, had recently learned of a genetic disorder called Lynch Syndrome (LS), which is linked with an increased risk of contracting endometrial cancers, especially colon cancer, before the age of 50 and a drastic increase in risk after 50 (Learn & Kahlenberg 2008). In 1966 Dr. Henry Lynch at Creighton University Medical Center discovered and characterized the autosomal dominant syndrome and the susceptibility to colon cancer (Learn & Kahlenberg 2008). In the 1990's LS was found to be caused by a germ-line defects in MisMatch Repair genes (MMR), primarily MLH1, MSH2, and MSH6 leading to genomic instability and inhibition of tumor-suppressor genes, which accelerates cancer formation in patients (Markowitz & Bertagnolli 2009). Every individual gets two copies of each gene, one from the mother and one from the father, and since LS is inherited in an autosomal dominant pattern, an individual receiving one copy of this mutation has LS. However if a person has only one copy, the disease

has a 70-80% penetrance pattern. Penetrance, is the genetics term denoting for the proportion of individuals that carry a gene and are affected. Since the penetrance of LS is less than 100%, the syndrome is not guaranteed to affect an individual diagnosed with LS. Therefore, about 20-30% of people diagnosed with LS will not develop cancer in their lifetime, while the remaining will (Learn & Kahlenberg 2008).

This alone may seem like an interesting fact, but becomes very relevant in my family's story as I started to tell. It begins with my father in 1978 when he was only 39 years old, he was diagnosed with a stage-three primary colon cancer. After having a colectomy that left him with half a colon, chemotherapy, and some unforeseen complications he was cancer free. Not too long after, both his younger sister and younger brother both had a primary colon cancer at the age of 45. Both of them were cancer free after treatment. Soon after the younger brother relapsed and was diagnosed with liver cancer at age 50, treatment was again successful. Some time past and all seemed well, until my aunt was diagnosed with a stage-four pancreatic cancer at age 68. Pancreatic cancer is essentially untreatable at that stage, and she eventually lost her life. The most recent development of this on going history, was when my father's older brother was diagnosed with colon cancer at age 75; luckily the cancer was found early and was treated easily. My father's family history has been riddled with cancer evidenced by my father and his two brothers and sister all having colon cancer in either their 40's or 50's, with the exception of one brother at a later age. Even more strikingly, my father having his bout at 39 years old and his sister having a relapse of cancer.

My father's family history raised much concern to my sister and I, and we insisted that my father go talk to a physician about his family history of colon cancer and potential LS. My father did not want to; his reasoning was that he had cancer already, so what would learning of a

genetic predisposition help him with? His Japanese culture also instilled in him an ideal that as a man, he should not show weakness, and he believed that being labeled with a disorder would mark him with weakness permanently. Although he never said this, I inferred that from his devastation with his sister's death, he was scared to find out if he had the risk of also getting cancer again. My mother, ultimately, was the one to convince him. She urged him to think of my sister and I, because even from their basic understanding of biology they knew that genes are passed on from parents to children. My father realized that he needed to do it and was happy to proceed with testing afterwards.

His physician referred him to a genetic counselor, who informed him about how his strong family history and personal bout with cancer at such a young age made him a prime candidate for Lynch Syndrome. The goals of the session with the genetic counselor was to further educate him on the basics of gene inheritance and also that the disease was not a guarantee of being affected by cancer, but rather would help set up a modified screening process that would more effectively help diagnose individuals at risk earlier. This educational session was meant to allow the patient to make an informed decision about genetic testing. Ultimately, leaving the decision up to the patient, would account for each individual's different perspective towards knowing vs. not-knowing and ideally understands what the psychosocial consequences are for themselves from the decision they make. He chose to take the test; his tests results came back positive.

My entire immediate family later met with a genetic counselor to discuss my father's positive result and its implications on the rest of us. The counselor, first told us that this is nothing to be scared or concerned about, rather we should be glad that my father's result was positive for LS in particular. She said this because due to our family's striking history, it was

highly likely that the cancers had a hereditary cause. There are many mutations that are known to be correlated with colon cancer, but only a few mutations have been researched enough and had enough cases to solidify an association between the mutation and the colon cancer found in those patients. LS is very well documented, in fact it is the most common form of hereditary colon cancer syndrome. Without knowledge of an existing mutation in our family to act as a basis for comparison, our tests, even if negative for LS, would be said to be uninformative. Since that is not the case for us anymore after our father's test, if my sister and I were to get tested we would know which mutation to test for. The counselor advised my sister and I to get a genetic test. Generally, in patients with Lynch Syndrome, screening for cancer, including colonoscopies and endoscopies, start in mid 20's due to the increased risk of developing cancer at a younger age. Regardless of our decision be to get tested or not, the counselor still suspected that we would have a hereditary risk and screening should begin 10 years before the age of my father's primary cancer. Which for us mean around 30 years of age. Both the LS diagnosis in my father and potential LS screening for the rest of the family was very exciting, particularly to my sister and I, because we have both been studying science for many years and we saw the LS genetic test as a tool to prevent cancer rather than a death sentence.

However, this state of mind was complicated when the counselor cautioned us to try to purchase life insurance before we get tested. She said this because currently the laws and legislations in place to protect an individual's genetic information from third parties, such as health insurance companies or employers, do not include life and disability insurance companies. Therefore, life and disability insurance are able to access information about genetic testing done by individuals and use risk-assessment to either deny or raise premiums. In parallel my father looked into getting life insurance for my sister and I, and found out that it is very difficult to get

a good policy as a student and without a good salary. This discovery was particularly concerning because my sister was in medical school at the time, and my future goal, was and is still, to go to medical school as well. So the earliest my sister or I could possibly get a life insurance plan is when we are between the ages of 25-27. 25 for my sister because she will be graduating from medical school and entering the workforce at that age, while in my case, I will be taking off one to two years before attending medical school, so the youngest I will begin my career would be at 27.

This really got under my skin. Life insurance is a fundamental option for the lives of all people and their families and it upset me that our lives could be valued less just because we have a higher risk of a disease that sporadically affects over 1 million American every year. The difference in extent of treatment and survival between individuals diagnosed in the early stages of cancer versus later stages is drastic. For my sister and I, the ability to take the LS genetic test could play a pivotal role in our own diagnoses in the future. Although we would still start screening early without the test, being able to truly know our efforts are not superfluous would be vindicating. This seemed so outrageous that insurance, something that is available to help people, is what is holding us back.

Now, at the age of 21 and I believe these experiences have led me to a point where I can contribute something of substance to the discussion of genetic ethics. A theme that threads through my stories is a dichotomy in views about the use of genetic technology. Just from my experiences we see that while my mother does not judge by genetics, my father was absolutely terrified by the idea of being labeled. In the end they either had to say yes or no to the use genetic technology in their situations. This illuminated the fact that there are two polar opposite

responses in this issue, neither of which being objectively right or wrong with an infinitesimal grey area in between.

Although I said earlier that I viewed my mother's choice not to test me for Down's Syndrome as the ultimate act of love, my father's, my sister's and I's current situation with LS has complicated the picture greatly. Comparing the two situations my mother's and my father's decisions were both made from a love for their children. Each decision had its own moral and ethical worth. My mother chose not to characterize any children as not worth having and had planned to support any children she had as long as she lived. On the down side from not knowing she risked not having the time to plan on and learn about raising a Down's Syndrome child if I had been born with it. This would have ultimately brought challenges to my parents financially and socially, but from my experience I believe my parents could have raised any child well. In my father's case he ultimately chose to get tested interestingly because of discussing this with my mother. They both have always had my sister's and I's best interest in mind. Now, when I look back at the position my father was put in, I realize the immense pressure he was put under. It is understandable that he had gone through cancer already before, and really did not want to know that he could be labeled with another sickness. My mother had to remind him that this was not just for him, but also for his children. My father's act of love also does not come without some cons. By knowing the presence of a mutation that leads to an increased likelihood and rate of developing cancer, he, my sister, and myself would all have to deal with the psychosocial and political/economic issues involved. The fear of getting sick, the fear of stigma and discrimination, that is all real too. So from my families story, we see this dichotomy in views on genetics. As we can see this technology is very situation specific, but as we know medicine has limits in how specific it can be and protocols need to be put in place. So how do we move

forward in a way that we use genetic technology for the best interest of both individuals and society without devaluing the integrity of our morality and ethics?

The dichotomy in views, displayed by my parents is very important and is not exclusive to them. In fact, this debate extends into the larger context of society illuminating conflict between individual choice and public health. Where does this conflict stem from? I believe that a large contributing factor is the fear of genetic discrimination and stigma. Unfortunately, another dichotomy also exists in society about genetic technology; should it be used for treatment or enhancement? This idea stems the present and theoretical power of genetics. We must answer the questions: for what, why, and how should genetic diagnosis be used? And should society be put before individuals and their family in regards to genetics, or vice versa? These questions encompass the parameters of ethics, morality, and sociology. We can tell if someone is going to be sick before they are born now, but as you are reading this essay, researchers around the world are working on ways to manipulate genes in different ways to change not only illnesses, but anything in the body that has to do with genes. The thing is, genes affect everything. This idea of manipulating something that is believed to be a concrete blue-print is an exciting, but equally worrisome thought.

My thesis will begin with a chapter that provides the necessary background of fundamental genetic theory, the discoveries within genetics, and the uses present today. Following this will be a second chapter that serve as a comparative analysis of past discrimination using race and science in comparison to more recent and present day ethical issues surfaced by the field of genetics. This chapter ultimately, will strive to discover underlying ideologies that people may knowingly or unknowingly hold that facilitate discrimination, prejudice, and stigma that may be causing the reservations that many people have towards

genetic testing and diagnosis. After this chapter I will explore the realm of science fiction, with an emphasis on *Brave New World* by Aldous Huxley, in order to further illuminate the tendencies of our society that may lead to irreparable consequences when coupled with the rise of genetic technology. In my penultimate chapter, I will attempt to prescribe a first step in our path to the proper and ethical use of genetics by explaining the need for furthering the education of health care professionals and public in the field of genetics. My final chapter will serve to reiterate the themes I hope you will take away from this work as well as inspire hope for a better future.

Thesis Statement

This thesis will explore the following: I see that the use of biology as a basis for discrimination is nothing new, but the developing technologies in the field of genetics add a nuance of being able to predict and change aspects of biology that were thought to be fixed. A society that is largely, genetically illiterate, also begs the question: how do we move forward making sure that the practices are done by the right people educated in both the sciences and ethics of the technology? Public Health, until now, has been widely focused on infectious disease and injury, but we will soon be faced with the question of how does genomics fit into public health? Genetic disorders, unlike infectious disease or injury, are not passed between individuals or self-inflicted. They are the result of an individual's family history at a molecular level. Therefore, genetic disorders cannot be treated the same, and I believe that as of right now we have the opportunity to design a future safe from genetic discrimination. Genetic technology has the potential to change and personalize medicine, but like anything else, must have its limits. Whether the answers to these questions are for health care professionals, research, the market, the government, or most likely a mix of the preceding, if we do not explore this issue,

consequences will inevitably arise. I will call upon the ideas of the science fiction novelist Aldous Huxley in his book, *Brave New World*, to affirm that our societal tendencies are enough to make these issues of genetic discrimination of prime concern that we must be proactive towards. I hope to provide insight with my stories of how genetic research and technology have affected my life and put my story in dialogue with science, sociology, ethics, morality, justice, and philosophy. This dialogue will hopefully illuminate that truth and power behind the ideologies our society hold that lead us to value certain people over others, and how we must address them in order to ensure that our society, that will inevitably be influenced by our genetic discoveries, be free of discrimination.

Ultimately, I hope to find at a least a first step in a plan of action in moving forward with the ethics of genetics, but more importantly I simply wish to promote the conversation. As we have seen so far from my story and introduction, there is a growing unease about genetics that is hard to define and before we find a solution, an underlying cause or theme must be illuminated, and I believe that can be achieved through conversation.

Genetics: Background and a Survey of Genetic Diagnosis Technology Today and in- Development

Genetics Background

If we look around us and we classify what is alive and not-alive, we could probably come up with many differences; such as: changing vs. unchanging, natural vs. unnatural, mobile vs. non-mobile, etc. Although these are all important distinctions, another one has stuck out for thousands of years in documented history: Life is exclusive from all other things on this earth with its ability to reproduce. Not only can living organisms reproduce, but curiously the offspring resemble their parents more than other individuals of the same species. This phenomenon of transmission of traits is known as inheritance or heredity (Campbell & Reece 1999). The curiosity sparked by these seemingly obvious observations started the study of heredity and hereditary variation, now known as genetics.

Mendelian Genetics/Chromosomal Theory of Inheritance

It is hard to talk about the basics of genetics, especially heredity, without discussing the father of the basic principles of heredity Gregor Mendel. He is most notable for his work with garden peas in unlocking some of the earliest genetic hypotheses. In his experiments he would observe attributes of the peas: 1) the character, which is a feature that can be inherited, such as flower color and 2) the traits, which are the variants in a character, seeds as white or purple.

Mendel chose to work with garden peas because he was able to have strict control

over which plants mated with which. This is because all flowers of peas contain both male and female sex organs. Normally, the peas self pollinate producing fairly identical offspring, but since Mendel could control mating he would cross-pollinate, which is much more applicable to the genetics of most organisms that must mate with others of their own species, like us. Using his controlled mating process he could monitor generations very easily. His designations for generations were the following: Parents (P), offspring of parents (F₁), and offspring of hybrids (F₂). In various experiments with the peas, observing different characters, Mendel came upon a distinct pattern. For sets of the characters, he noticed that the F₂ generation had a 3:1 ratio of traits. For example, from a purple (P) and a white (P) purple (F₁) offspring are produced, and then the F₂ generation has a 3:1 ratio of purple to white flowers. The 3:1 phenomenon was entitled the law of segregation (Campbell & Reece 1999).

From the law of segregation Mendel developed four hypotheses. The first was that heritable factors (we now classify these as genes and I will refer to them as genes from now on) all have alternative versions that account for variation. We now call these alternate versions, alleles. The second hypothesis was that each character is determined by two alleles in an individual organism, one from each parent. The third hypothesis was that there is a dominant and recessive allele for each pair. Dominant alleles require only one copy in order to be fully expressed by the individual, while recessive alleles require two. The last hypothesis was that during the formation of the sex cells, or gametes, the pair of alleles for any given gene will separate so that each gamete only has one copy. This happens because these gametes are the cells that come together during fertilization (i.e. egg and sperm), and since all individuals only have two alleles for each gene, this law ensures that one comes from each parent. This segregation of alleles into the sex cells is now known as the law of segregation (Campbell & Reece 1999).

These novel hypotheses of Mendel are still going strong and have been extended into some of the keystone terms of basic genetics. The term homozygous means that an individual has a pair of alleles of the same variant and heterozygous, meaning a person has one of each variant. The combination of alleles found in an individual determines that individual's genetic makeup, also known as genotype. The appearance or difference in expression dictated by these alleles is known as the phenotype (Campbell & Reece 1999).

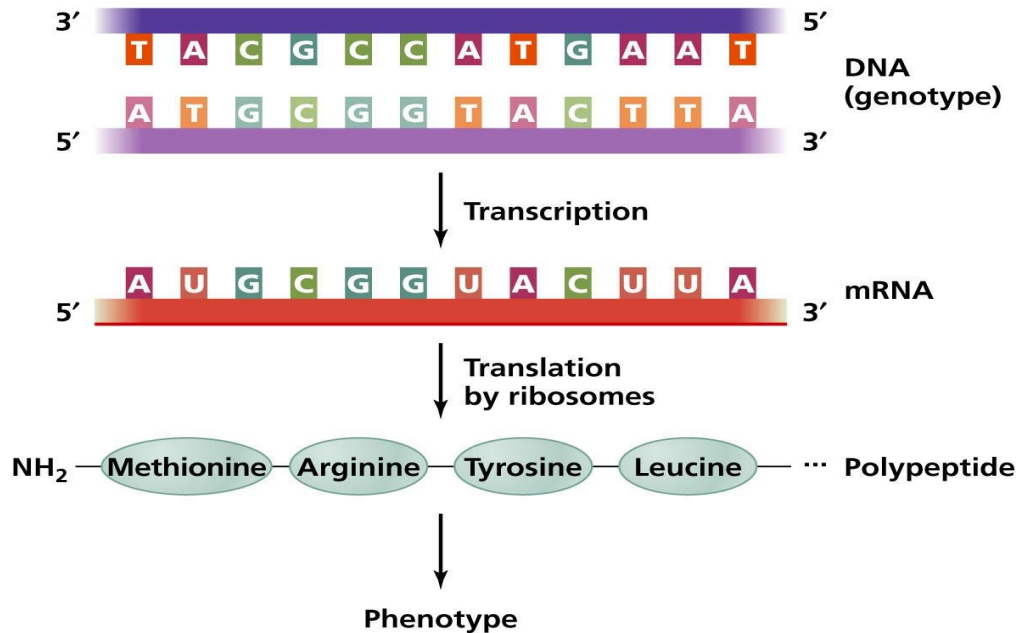
Mendel's discoveries were made all the way back in the 1850's. Even though they were convincing, his ideas about the way genes are inherited at a cellular level were still theoretical. During the late 1800's, cell biologists and cytologists, started deciphering the processes of cell division, mitosis and meiosis. They observed large structures shaped like an X near the center of cells before and after they divide (Campbell & Reece 1999). Most notably they noticed that the nuclei of male and female reproductive cells seem to converge after fertilization. This marked the integration of cytology and genetics. Mendel's fourth hypothesis that alleles segregate into the sex cells of individuals began to come to fruition (Hartl & Ruvolo 2012). In 1902, two geneticists named Walter Sutton and Theodor Boveri discovered the chromosome theory of inheritance. They found that chromosomes structures within the cell that carry genetic information. Mendelian genes are found on specific loci on chromosomes. Genes that are linked are transmitted together during cell division (Nussbaum, McInnes, & Willard 2007).

DNA/Molecular Basis of Inheritance: Central Dogma of Biology

In the story of genetics there is a theme of ideas going from bigger objects to smaller. We have already seen that genetics has gone from ideas based on observations of organisms to cells. For a while after the chromosome theory, little hints of what may be behind the chromosomes role in inheritance were left from bacterial research and microscopic imaging. Watson and Crick

in 1953 proposed a three-dimensional structure of deoxyribose nucleic acid (DNA) (Hartl & Ruvolo 2012). DNA is built of two long twisted chains containing four subunits called deoxynucleotides containing bases. Deoxynucleotide Triphosphates (dNTP) contain a deoxynucleotide unit with a ribose sugar and 1-3 phosphate groups attached. There are four variations of the dNTP subunits of DNA: adenine (A), thymine (T), Guanine (G), and cytosine (C); the four letters I mentioned earlier. Due to the properties of these bases, A-T and C-G exclusively bind. This discovery of the complementation and sequencing of bases led scientists to eventually develop the central dogma of biology: DNA → RNA → Protein (Figure 1). The genetic code held in the script of DNA codes for all the protein in our body ultimately giving us the ability to live. The processes by which these conversions occur are called transcription and translation. Transcription is the process of converting DNA into an intermediary molecule called ribonucleic acid (RNA). RNA must be made because the code in DNA cannot directly produce protein. Transcription begins with the splitting of the initial DNA strand into single template strands followed by the formation of single stranded RNA that complement those strands with the help of enzymes. RNA is very similar to DNA, except for the base T is replaced uracil (U). Once RNA is formed its sequence can be used to create proteins in a process called translation. When translation is occurring the RNA produced from transcription is demarked as messenger RNA (mRNA). mRNA is taken to an organelle in the cell called a ribosome, which then recruits another molecule called tRNA. tRNA contains three bases that complement with specific parts of mRNA; the three bases on the mRNA in which they bind are called codons. Codons are a triad of bases that code for a particular amino acid, which are the building blocks of protein. As specific tRNA read and bind the mRNA, the amino acids produced form a chain called a polypeptide, which in the end becomes a protein (Hartl & Ruvolo 2012). Ultimately, the central dogma is the

peak of Mendelian genetics in that it explains how we go from our genotype or DNA to our phenotypes expressed by proteins.



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Figure 1: Illustration of the steps of the central dogma of biology

Human Genome Project and ENCODE

After Mendel's hypotheses and Watson and Crick gave us the knowledge of inheritance and the molecule that carries the information, the next step for science was to be able to explore the DNA that makes us human. Initiated by the National Institute of Health and the Department of Energy in 1990, the Human Genome Project focused on the development of genetic and physical maps of the human genome. The project was aimed towards the ultimate goal of having more productive hypothesis-driven research of diseases and genes that may be causing them (Collins & McCusick 2001).

After the success of the Human Genome Project, a new exploration took off called Project ENCODE. The Human Genome Project gave us the book of the human genome, but ENCODE has and will allow us to define the words and their functions. Started in 2003, the

Encyclopedia of DNA Elements (ENCODE) Consortium is an international collaboration of research groups all working towards finding functional DNA sequences that act at the protein and RNA levels, as well as regulatory elements that control gene expression (Qu & Fang 2013). Both the Human Genome Project and ENCODE are having a profound impact on human genomic studies by rapidly expanding database of sequence information accessible online. ENCODE takes the Human Genome Project a step further, because knowing the sequence of DNA does not tell us much about the function, but ENCODE is attempting to close the gap between genotypes and phenotypes. In consideration with diseases, currently around about 5000 diseases that are associated with a genetic cause with approximately 1000 genetic or protein markers having been isolated.

Genetic Counseling

With the collection of knowledge growing of what genotypes cause what phenotypes, scientists and public health professionals used the information to study populations and the incidence of phenotypes. Association describes the situation when a particular allele is found either significantly more or significantly less frequently in a group of affected individuals compared to the frequency of the allele expected in the general population. This idea of association can be used at a smaller scale in order to address families and individual risk; this is called linkage (Nussbaum, McInnes, & Willard 2007).

As the knowledge and population statistics of genetic disorders grew, the field of Genetic Counseling was created. Genetic Counselors are health care professionals who provide information and assistance to affected individuals or family members with a possible genetic predisposition for a disorder. Also, they provide counseling on the consequences of the disorder,

the probability of developing/transmitting, and ways in which it may be prevented or treated (Nussbaum, McInnes, & Willard 2007).

The origin of genetic counseling is typically thought to have arisen in the setting of prenatal counseling. Counseling has drawn many obvious influences from prenatal practices most importantly the idea that couples have the right to make any reproductive choice they see fit. This ideal is at the base of nondirective counseling. Patients are never told what decisions to make when considering between the many testing and management options, but are rather provided the proper information and support in order for the patients to come to a decision that they deem to be appropriate for him/herself and his/her family.

Pedigrees are figures used by geneticists, especially genetic counselors, that display family history of hereditary condition(s). Often pedigrees are catered to the patient seeking genetic counseling, usually referred to as the proband. The proband may be currently affected, past affected, or unaffected when coming in for counseling, but the family history must be striking enough for their primary physician to have recommended genetic counseling. The pedigree when used in these cases, is not just useful for showing a family history. Genetic counselors pay special attention to the relationship of the family members to the proband, so that they can diagnose their status with respect to a particular hereditary condition. The family history is then compared to population statistics to determine the proband's risk of developing or transmitting the disorder; this process is called genetic screening (Nussbaum, McInnes, & Willard 2007).

Population statistics will include symptoms, possible conditions, affected age range, and penetrance of different genetic disorders. Most of the time the proband will come in for genetic counseling with a specific concern such as down's syndrome or hereditary cancer. This narrows

down what genetic disorders the counselors will compare and contrast his/her family history with. For example, if a patient comes in with concerns for hereditary breast cancer, the genetic counselor will ask the patient to provide his/her family history medical conditions. After collecting the information the counselor will organize the history into a pedigree of as many generations as the patient provides. For an individual concerned about hereditary breast cancer the counselor will look first at whether or not the individual was affected by breast cancer. If not, then the counselor will look at what family members have been affected. The most prevalent known mutations that are linked with breast cancer are BRCA1 and 2. Population statistics show that individuals with this mutation have a higher probability of developing breast or ovarian cancer before the age of 50. The counselor will look for these details and if the family history fits the criteria the counselor will ask the patient if he or she would like to be genetically tested for the BRCA1 and 2 genes. Also if there is someone that is currently affected by the cancer, whether it be the patient or a relative, the genetic counselor will suggest genetically testing the tumor cells. That is the most effective way to narrow down the mutation in order to know what to test the rest of the family for. After analyzing family history and possible genetic testing, genetic counselors will discuss with the patient what options he/she and his/her family have and provide suggestions on lifestyle choices. These will vary depending on the genetic disorder in consideration.

Earlier in my introduction, I detailed my striking family history of cancer on my father's side. Figure 2 is my family pedigree from our consultation with our genetic counselor. The pedigree shows my father, Seiji Tanaka, as the proband because he was the patient of interest. The genetic condition of interest was hereditary colon cancer because the high incidence of the disease in my father's family. My father provided the family history of cancers and the ages of

diagnosis. Three cancers have occurred in the family history provided by my father; colon cancer (denoted by the half pink shading), breast cancer (denoted by the color blue), liver cancer (denoted by the half green shading), and pancreatic cancer (denoted by the half gray shading). Under each individual in the pedigree is either the current age or deceased status and age of death (denoted by slash through). Below are the past disease(s) with the ages of diagnosis. Note, my mother had breast cancer at age 50, but since my father's genetics were the interest, the counselor did not take her disease into account when comparing the history with population statistics. With every single one of my father's siblings having colon cancer, mostly at a younger age, the genetic counselor had strong evidence that the cancer was hereditary. As said earlier, the most common hereditary colon cancer syndrome is Non-Polyposis Hereditary Colon Cancer, also known as Lynch syndrome (LS). LS is caused by mutations in Mismatch Repair genes (MMR), primarily MLH1, MSH2, and MSH6. The mutations lead to genetic instability in tumor suppressing mechanisms, which allows tumors to form easily and quickly. LS increases risk of colon cancer and other endometrial/abdominal cancers before the age of 50. With my father's family history the genetic counselor suggested testing for LS using genetic testing. As I said earlier, the test came back positive. This meant my father carried this mutation for LS, and although he had cancer once before, he was still at higher risk of relapsing. This prompted the genetic counselor to revise his current screening regimen to assure that if his cancer were to return it could be caught early on. In fact, during my father's last colonoscopy a pre-cancerous polyp was found and excised, avoiding a possible catastrophe.

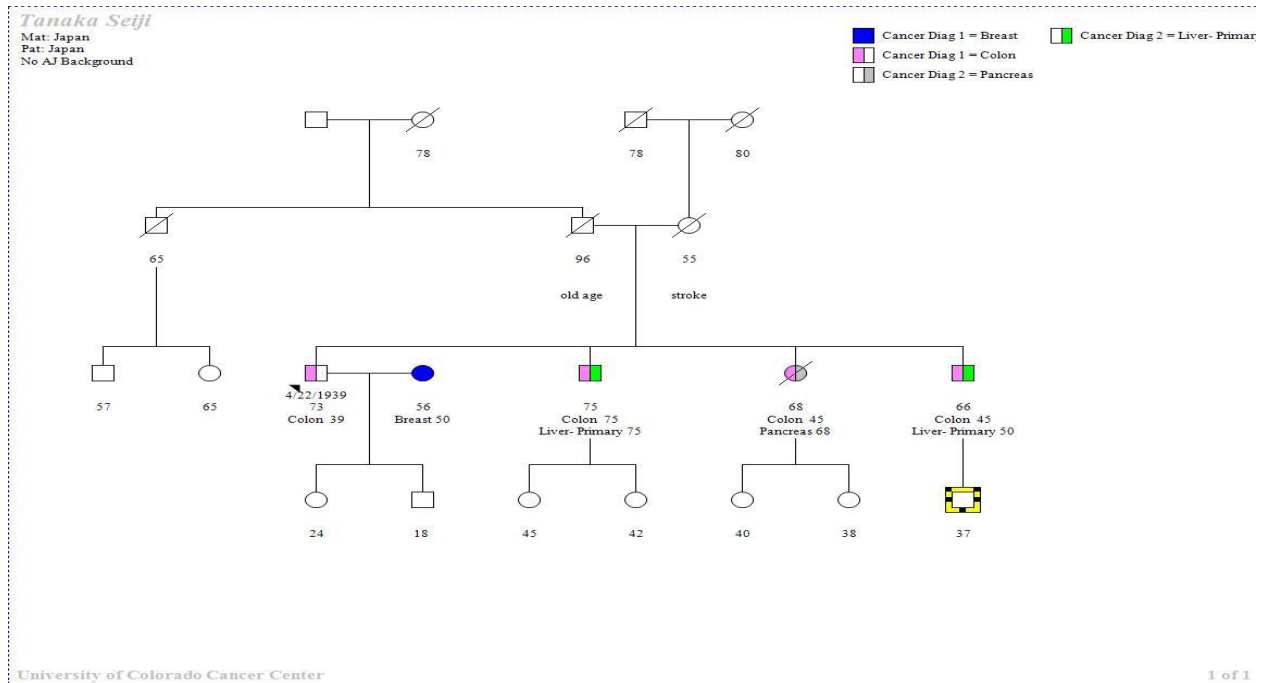


Figure 2: The Tanaka Family pedigree including age of diagnoses of cancer. Names not included. (squares=males; circles=females)

During the summer of 2014, I was able to shadow my genetic counselor for a day in the clinic. I learned that the main philosophy that genetic counselors follow is to determine relative risk for their patients and determining if the risk is clinically actionable. Clinically actionable, entails known medical screening for different genetic disorders. If a patient is shown to be at lower risk, but still higher than average, counselors provide advice for prevention and risk reduction including lifestyle and early screening.

Genetic counseling has many different specialities including prenatal, cancer, and other genetic disorders often associated with immunity. Each speciality has unique situations that do not directly translate to the others, I will focus on a couple examples from the prenatal and cancer field. For example prenatal counseling either tests the parents, a fetus, or an artificially inseminated embryo for genetic disorders giving the parents the following choices: to have children regardless of results, contraception or sterilization, adoption, artificial insemination, to

proceed with current pregnancy, or to abort a pregnancy. Cancer counselors use family history and genetic test from tumor tissue of family member (if available) to determine the risk for the individual receiving counseling. The counselor will use symptoms associated with certain mutations to determine the underlying mutation in the family, if any is present, and then give that information to the patient. The patient has the following choices: not to or to get tested for the mutation, increase screening or preventive measures if tested positive, receive information about lifestyle changes that can be taken, or employ prophylactic surgery (if possible) to reduce risk for cancer (example: removal of breast tissue and reproductive organs for women positive for BRCA1 or 2 mutation, which leads to 70% increase of cancer risk at an early age).

On top of providing scientific and risk information to patients, counselors must be thoroughly trained to provide consultation on the cost of testing, coping with adjusted or new diagnosis, and options in informing other family members to whom the results may be relevant. Also they must be deliver psychological support either themselves or by recognizing when the patient should be referred to a psychiatrist/therapist specialized in the field of disease. Psychological/social effects include but are not exclusive to: anxiety and distress, depression, guilt, altered self-image, altered relationship with family, need for support, life choices, and overestimation of risk (Mounsour 2007).

An example of an emotional and social stress that can be surfaced by genetics is a patient's attitude about being informed of a disorder or risk. To be informed in general means to know that a disorder either will affect the individual or the family and to have to decide about reproduction. Some patients can deal with this better than others because they are the type of person that would rather hear bad news than stay in the dark. Other patients may be ridden with fear, guilt, indecisiveness, and confusion. The psychological aspect of genetic counseling and

genetics in general is vast and I will make an effort to illuminate the principle aspects under concern and the ethical issues they entail later on.

Genetic sequencing and diagnosis technology

During the Human Genome Project there was a drastic increase in the development of systems with large computing power in order to sequence genes more efficiently and cost effectively. Since then, this trend has continued. Next Generation sequencing has increase the speed of sequencing drastically. This technology has made it more practical to make genetic testing and sequencing available to more people (Desai & Jere 2012).

Genetic testing is a process with the goal of detecting presences, or absences, alteration in certain genes as well as testing for protein residues found in the blood or tissues that are related to genetic disorders. There are 5 major types of genetic testing: prenatal diagnosis, newborn, presymptomatic, diagnostic, and carrier testing (Mounsour 2007). Prenatal diagnosis is used to conduct tests on a developing fetus, usually done by testing amniotic fluid for protein markers or blood from the umbilical cord. A perfect example of prenatal diagnosis is the case of Down's Syndrome, which I had brought up earlier. Newborn testing aims to diagnose and find markers of metabolic disorders because often early treatment is vital to stop the progression or the control these diseases. A commonly seen test is that for phenylketonuria (PKU), which is a disorder caused by a mutation in the gene that codes for an enzyme meant to digest a certain amino acid obtained from food causing developmental deficits. PKU if diagnosed early on can be treated by changing the diet of the individual. Presymptomatic testing is a test aimed to test healthy individuals to determine if they carry a genetic mutation that leads to increase probability of developing certain conditions. If my sister and I were to undergo testing for Lynch Syndrome it would be considered presymptomatic testing. Diagnostic genetic testing is used when someone is

suspected of being affected by a condition that may have a genetic causation. A good example of this kind of testing is that done for Huntington's Disease, which is a late onset neurodegenerative disorder caused by an alteration in one gene (therefore individuals with one copy of the mutated gene are affected). Although the disease is late onset, it has a 100% penetrance in individuals with one copy of the mutated genes. Therefore individuals with a family history of this disease will often have to make the choice to either get tested or not around their 20's and a positive test is a diagnosis of the disease. Carrier testing is used to determine if a person has one copy of a recessive genetic disorder. Cystic Fibrosis is a genetic disorder that affects about 30,000 Americans that is caused by a mutation that makes the mucus produced by the body unusually thick and sticky, eventually leading to lung, digestive, and pancreatic problems. A person who tests positive as a carrier is not affected by the disorder, but may pass it on to their offspring if their spouse is also a carrier. Ergo, carrier testing is usually used to aid reproductive decisions.

Gene Therapy

Gene Therapy is the process of manipulating the genome of an individual in the following ways: 1) Replacing a mutated gene that causes a genetic disorder with a healthy copy 2) Knocking-out, or inactivating mutated genes that are not functioning properly or 3) Introducing a new gene in the body to help combat disease (Gilbert & Tyler 2005).

Somatic cell gene therapy is done by transferring therapeutic/non-mutated genes into the somatic cells (non-sex cells), of a patient and only affects the individual treated (Gilbert & Tyler 2005). An example of somatic cell gene therapy is one for severe combined immunodeficiency (SCID), a genetic disorder caused by a mutation in the genes that code for vital immune cells. Patients that are diagnosed with SCID that go without treatment have a high mortality rate at early ages. SCID is normally treated with bone marrow transplant, but this requires that a donor

be found. SCID was the first genetic disease to be cured by gene therapy. Bone marrow cells from affect patients were infected in culture with a retroviral vector (DNA implanted from an engineered virus) to express the gene for which the patients had loss the function.

Germ-line gene therapy is when germ cells (sperm or eggs) are modified by the introduction of functional genes, which are integrated into the entire genome of an individual. The therapy changes both the treated individual and their offspring (Gilbert & Tyler 2005). Even though the theory behind it is possible, there has not been many developments in germ-line gene therapy because the lag in our current knowledge and technology as well as the ethical concerns of altering an entire family line. Recently, the United Kingdom parliament passed a legislation that legalized a derivative form of germ-line gene therapy. The method is called “Three-Parent *In Vitro* Fertilization” (IVF), which is aimed towards preventing the transmission of mitochondrial diseases from affected mothers. Mitochondrial dysfunction disorders have been linked with various multi-organ diseases. The mitochondria is an organelle within the cells of the body, which is the primary site of metabolism providing energy for the body. Mitochondrial DNA, for the most part, is passed down to offspring from the mother’s egg, so if a mother is affected then the child will also be affected. Three-Parent IVF works by transferring the genome of an early stage fertilized egg into an enucleated egg of an unaffected women. Therefore, the child will have all of the nuclear DNA from the biological parents, but will have the mitochondrial DNA from the third parent (Amato, Tachibana, Sparman, & Mitalipov, 2014).

After the advent of genetic advancements prompted by the Human Genome Project, there have been many advancements in genetic diagnosis and even preliminary versions of genetic therapy. That being said, the technology is quite beyond our knowledge, because although we can sequence any person’s genome much faster and cheaply now, the function of the majority of

our genetic material has not been defined yet. For example, although the causation of the BRCA genes have been linked to high risk of breast cancer, there are about 20 other genes that have been correlated with breast cancer with limited population statistics to quantify the relationship. Therefore, it is foreseeable that genetic research will yield new discoveries constantly for an indefinite amount of time. With these discoveries, our ability to define and manipulate all the traits and predispositions of an individual will grow, and that is why defining a protocol for this technology to be used is of the utmost importance. As Voltaire as well as Uncle Ben from the *Spider-Man* comics have said, “With great power, comes great responsibility” (*Spider-Man* 2002).

Ethical Issues seen in the Past and Today

Discrimination

In the discussion of the dichotomy in attitudes towards genetic screening in our society today it would be remiss if I did not discuss the effects of discrimination and stigma. These ethical issues are perhaps the most potent and influential psychological and social issues that have shaped both the history of the world and many today's societies. Discrimination, as defined by the oxford english dictionary, means: "The action of perceiving, noting, or making a distinction between humans" (Simpson 1989). In itself, this is a natural tendency of humanity. We like to categorize anything; for example, somebody may look around while driving and say, "that is a blue jay and that is an eagle or that car is a Toyota and that car is a Ford." Simply, that is the process of making distinctions, so why would I mention it as one of the most detrimental issues to pervade history and the present?

Discrimination, just based off its definition, is neither harmful nor unethical, but it has a bad connotation because of its association with movements that have caused unfair treatment based on ethnic origin, religion, gender or disability. Also often in these discussions of ethics it is beneficial to use paradigms in order to have a spectrum of comparison to determine whether the present situation being discussed lies on one end versus the other; this is a technique called casuistry. A simple paradigm of discrimination can be set in the setting of applying for a job. Imagine that there is a pool of applicants

with all the same credentials, but half of them are “normal”, while half of them have premature baldness. If any employer has half the number of positions open than the size of the pool of candidates at least two possible scenarios could happen: 1. 50/50 of both groups of individuals are hired it shows that the employer did not discriminate based on a stigmatized trait. 2. A majority or all people hired are normal weight, showing that that the employer discriminated against the trait of baldness even though it was not a qualification for the job. Although this paradigm is highly exaggerated, it offers clear-cut extremes. This will serve as my paradigm of comparison for my discussion of genetic discrimination, with the spectrum extending from no discrimination to the presence of discrimination. So as we can see, discrimination becomes a problem when we allow it to inflict stigma upon people because of the prejudice.

Ultimately, categories like race, culture, sexuality, genotypes, etc. do not have any inherent bias, but discrimination results from meaning being placed on them by the social and cultural context they are found in. Two major factors of discrimination are stigma and prejudice. Stigma is a mark of disgrace or infamy; a sign of severe censure or condemnation, regarded as impressed on a person or thing. Prejudice is a feeling, favourable or unfavourable, towards a person, thing, or class.

The evolution of discrimination to stigma and prejudice has brought forth some of the most concerning ethical issues that society has and still is facing: racism, sexism, caste systems, gender/sexual orientation inequality, and the list goes on. Perhaps one of the most potent of that list to affect society is racism. Racism often lies at the route of imperialism, colonialism, the slave trade, genocide, and war. I understand that in a longer work that of the other examples of negative discrimination could be discussed, but I have chosen to concentrate on racism for this discussion due to its history and situations that are analogous to my discussion on genetics. I

believe that before engaging in how the use of genetics is and could lead to discrimination, a historical and philosophical exploration of racism and racism using science is necessary. In my exploration I will tease out details that pertain to science or more specifically to heredity/genetics.

Origins of Racism

The ideas of discrimination and dominant forms of humans is quite old. Pinpointing the origin is difficult, but it is fair to assume that ideas predating Christ have a high probability of being the origin. Many people are familiar with the greek philosopher Aristotle, who is best known for his ideas of the “good-life” in his work the *Nicomachean Ethics* and his idea of gaining knowledge through deductive reasoning and experience, also known as empiricism. Overall, Aristotle sought to provide a process of reasoning for people to learn and understanding reality and the natural world.

Aristotle did not particularly begin the thought of drawing distinctions due to physical features, but he furthered developed the idea “essence,” first introduced by another greek philosopher, Plato. He believed that man could be identified by his “essence,” which is the immutable and unchanging truths behind the physical being. He also asserted that the essences of all beings and organisms on earth are arranged in a *Scala Naturae*, where organisms are intrinsically “lower” or “higher” beings. Underlying these ideas is teleology, an idea positing that all things have an innate goal or limit. Aristotle believed that there are two types of men: the “political man” and the “barbarian man.” Political men are those capable of participating in society to help make ensure a civilized and peaceful society. The political man also must be able to govern its own actions. The barbarian men were men unfit for self-governance and therefore were natural slaves. So although Aristotle did not use physical markers, he did begin a train of

thought that placed unchangeable attributes on people that allowed for one to feel superior to another (Jackson and Weidman 2004).

Christianity, for ages was governed by biblical literalism. Christians were of the earliest to begin enslaving Africans, and their purposed reason for doing so was the “Curse of Ham.” The curse originates from the story of Noah. Noah had three sons, one of whom was named Ham. One day while Noah was in a drunken state, Ham mocked his father, but in the process awoke him. It may seem like this is of little offense, but many analyses of the Curse of Ham mythos suggest that Noah was nude in his drunken stupor and therefore Ham had revealed himself as being immodest and deviant. Noah, by the power of God cursed Ham and his descendents. Ham was assigned sovereignty over Africa and his descendents were cursed with black skin (Jackson and Weidman 2004).

Jacques Nicolas Poillat de Montabert, a painter and theorist, in 1837 wrote, “White is the symbol of Divinity or God; Black is the symbol of the evil spirit or the demon...” (Goldenberg 2005). This was written in a manual for painters. This starkly demonstrates how engrained the idea of the opposition of black and white. While I cannot be certain that all of the writers that cited this story were of Judeo-Christian religious background, it is unarguable that the bible is not confined to its influence over the religious and is widespread throughout the past western culture. The Curse of Ham has in fact been cited during the enslavement of Africans. During 1838, a pro-slavery author wrote, “The blacks were originally designed to vassalage by the Patriarch Noah” (Goldenberg 2005). Vassalage, can easily be replaced with the word submissive or subordinate. Blacks were seen as lesser, and basically sub-human. Note, that these writings are only from about 200 years ago, showing that the use of this story is not far from the present in the scope of history.

Many people in the present may be skeptical about whether this is proper evidence for racism in Judeo-Christian Western Culture because this story is not often if even recited in the present. Like I said above, we must consider that the tradition of biblical literalism is not as prevalent now as it has been in history. In fact, I am willing to argue that racism in Christianity, is now marked as sinful. In 1985 Pope John Paul II organized a council to construct the catechism, in hopes to thoroughly summarize and make clear the principles of the Catholic religion. In the articles of Social Justice the catechism directly confronts the problem of racism. Passage 1935 goes as follows: the equality of men rests essentially on their dignity as persons and the rights that flow from it: “Every form of social or cultural discrimination in fundamental personal rights on the grounds of sex, race, color, social conditions, language, or religion must be curbed and eradicated as incompatible with God’s design” (U.S. Catholic Church 1997). Not only does the passage address race, but also extends to other social conditions that are often victim to prejudice. I cannot say that every Catholic or Christian reads or follows the catechism, neither can I say that problems with racism and other forms of discrimination have been removed among Judeo-Christians or Western society by the release of the catechism. I can say that this shows that those in power made an effort to disclose and make no room for interpretation over the values of the church.

My example of the Curse of Ham, although not culturally relevant now, has great utility in analyzing the patterns and effects of racism and prejudice in the past. What I wish to show with this piece of evidence is how an idea can spread through a society without necessarily rational reasoning. Analogous to my discussion of genetics is the theme of interpretation. It is more than likely that the original author of the story of Noah did not intend for social prejudice to ensue, but he/she also did not do anything to say otherwise. I have to chosen to write about

genetics because we must address this issue to attempt to leave no room for interpretation. We must decide how we want to integrate and utilize this growing and powerful technology into our society without wavering from our moral and ethical responsibilities.

Racism/Discrimination using Science

Carolus Linnaeus, a professor of botany, during the 1700's developed the modern classification method in which organisms are arranged broadly (kingdom) gradually more specific eventually reaching the classification of species. Linnaeus proposed the idea that although humans are members of the same species there are four varieties with their own unique attributes. The four types are: Americanus, Asiaticus, Africanus, and Europaeus. His classifications provided further evidence supporting the notion that the people creating a system will create it in their favor. Examples of his classifications: Africanus= black skin, frizzled hair, indolent, women without shame, governed by impulse; Europaeus= white, long, flowing hair, blue eyes, gentle, inventive, covers himself with close-fitting clothing, and governed by laws. His classifications, although not noted at the time, were the first notions of race and culture. His system would lead to the monogenism school of thought, which recognized that humans are one species and races formed from environment. Many of the race scholars at the time, like Linnaeus, used europeans as a standard with other races as degenerations of the norm (Jackson and Weidman 2004).

In the 19TH century the idea of polygenism arose, which asserted that different races actually are a result of human speciation into distinct species. One of the most notable polygenecists is the American physician and anatomist, Samuel George Morton. He coined the idea that races have observable differences in cranial sizes and proportions, which reveal essential qualities and mental worth/capacities of their owners. With the monogenist and

polygenic movements we see racism, or discrimination in general, being more focused on generalizing the inner qualities of a group of people rather than just physical attributes (Jackson and Weidman 2004).

A true turning point in the use of science for discrimination was the publication of *Origin of Species* by Charles Darwin in 1859. Darwin came up with the idea that all organisms came from a common ancestor and evolved into the many different species. He defined the motor of evolution as natural selection, in which organisms with favorable traits are more likely to succeed and live to reproduce, therefore passing on those traits to their offspring. Evolution added the nuance of inheritance as an equally influential factor over traits, when environment was thought to be the only mode. Gregor Mendel, whom we have already discussed, further made the distinction that heredity is independent of environment. The idea of genes controlling the essence of a person, gave racists or anyone who wanted to discriminate the notion that favorable/unfavorable essences of people are passed on regardless of where they are. This leads well to the discussion at hand in my thesis, people who have reservations towards genetic screening are not without good reason, because now that we have mapped the genome and are working on mapping the functions of genes, a person's entire essence has the possibility of being known. We no longer only have to worry about people discriminating racial stereotypes, but now there is the possibility of discriminating against anyone with unfavorable genes regardless of race. Seemingly, no one is safe from scrutiny (Jackson and Weidman 2004).

Often people are unaware that they hold the ideology of discrimination or any ideologies at all. Ideologies are the combination of our conscious and unconscious ideas, which are unquestioned by those holding them. These ideas influence the motivations and actions of those who hold them. The 5 tenets of the ideology of biological discrimination are:

1) People can be classified in distinct, biological groups on the basis of physical characteristics, either phenotypic or genotypic. 2) These groups can be ranked on a hierarchy with some better than others. 3) Outer characteristics of people are linked to inner characteristics. 4) These outer signs and inner capacities mentioned above are *inherited and innate*. 5) These differentiated capacities/characteristics are *fixed either by nature or God* (Jackson and Weidman 2004).

Discriminatory ideology often uses words such as “innate” or “fixed” meaning the thing being discriminated against is inescapable by the person or group. What happens when you introduce the ability to map every single trait or change the genes within a person, which is a reality now that is being furthered everyday?

The novelist Annie Dillard warns us of the power of the ability to communicate and put words to what we see as natural phenomena. Ultimately, our words can overshadow what is happening naturally, and although I concede that if used in an ethical and moral way words can shape society for the better, they also hold tremendous destructive power. “Biological evolution takes time...the unit of reproduction is the mortal and replicating creature. Once the naked ape starts talking, however, ‘the unit of reproduction becomes’... ‘the mouth’” (Dillard 1999).

Present Day Genetic Discrimination

Barash loosely defines genetic discrimination as: differential treatment based on an identified or presumed genotype. Differential treatment includes exclusion from disability service, being labeled as having a pre-existing condition affecting insurance, stigmas arisen affecting relationships, valuing one genotype over another, distributive justice of gene testing, and many other variables (Barash 2008).

As I mentioned earlier on in my story, one of the biggest issues in genetics is insurance. The Health Insurance Portability and Accountability Act (HIPAA) has regulations that mandate that a patient's authorization is required to release his or her medical information, including the results of genetic testing. However, exceptions exist. The first exception is that genetic practitioners can disclose a patient's protected health information if it is seen as a serious threat to another person's health or safety. This first exception is reasonable in that it sets out to secure public safety if the patient's intention is to harm others. The point must be made clear that the patient has still made the choice to be threatening giving just cause for disclosure of his or her information. The second exception, is one that I had mentioned earlier, life and disability insurance are not yet included in HIPAA's regulations. This leads to insurance underwriting, which is when insurers insist that they require access to all relevant genetic information about an individual. Life insurance companies claim that calculations show that their premiums plotted on an age-specific survival are skewed by individuals with hidden genetic information. They follow the assumption that current premiums cannot cover losses accrued from individuals with hidden risks for disease. Therefore, people with genetic disorders or predispositions must buy extra life or long-term disability insurance. This is referred as the process of adverse selection, where premiums would have to increase for the majority of customers to subsidize the minority of individuals. For many, if they think in the realms of economics, this exception seems reasonable (Nussbaum, McInnes, & Willard 2007). Why should people that are genetically "healthier" have to pay for those who are not? I put quotations because the term "healthier" in this case should not be used in the same as the way we would use it typically. Health is something that should be able to be observed in the present, not anticipatorily. I call upon the term "symptomology" to further this discussion.

Insurers routinely obtain smoking history, blood pressure, serum cholesterol, and urine glucose testing in order to determine premiums. So they ask why should genetic information be constrained? The insurance companies are not considering all the variables that play a part of most genetic disorders, specifically the distinction between affected individuals with an observable phenotype versus individuals with predictive risk. A phenotype as defined earlier is the expression of genes observed as physical traits. Although that is the traditional definition, phenotypes can also be simply any observable characteristics even if they are a result of environment. So things like obesity, diabetes, hypertension, high cholesterol, or tar in the lungs from smoking are also phenotypes. A genetic disorder that is inherited in a dominant fashion will manifest itself usually at an early age between birth and adolescents with early 20's being the latest. Other genetic disorders, especially those associated with cancer, just entail an increased risk, but never have 100% penetrance meaning that the affected individual may never express the phenotype (Nussbaum, McInnes, & Willard 2007). I would like to draw a clear distinction between genetic disorders and the observable phenotypes listed above. First, phenotypes that are a result of environment are a result of a person's choice (for the most part), while a dominant genetic disorder is not the choice of the individual. Secondly, having risk of disease is not the same as having a disease or being unhealthy. I would like to draw on a specific case to explore this topic.

Symptomology entails how we determine whether something is a disability or not? Often genetic tests can reveal something that puts people at risk later in their life, but for the most part they are asymptomatic (meaning they do not express any symptom for the time being). Consider the scenario when one person's cholesterol test results indicates a level of cholesterol that functionally provides a measure risk of cardiac disease. Then a gene test for the Apo enzyme E

gene indicates another person's risk of cardiac disease later on. As of now insurers would be allowed to raise the premiums for both, but could deny a insurance plan to the latter. Insurers argue that genetic tests are nothing short of a medical test and prevent them from getting cheated, but many people fear that insurers will use tests to exclude anyone short of genetically "good" health (Barash 2008).

This issue is particularly pervasive in countries like the United States or those that share our model of insurance, where the practice is carried out primarily by private companies. Ultimately, these companies want to turn a profit, while the concern for public health and assuring that all people have access to both preventative and therapeutic healthcare often comes as an after thought.

One of the most topical subjects of the last 5 years has been access to health care in America. In response to this need for access the Obama Administration and congress passed the Affordable Care Act, which has the goal of making sure the percentage of uninsured Americans goes down. The law includes portions revising existing systems like medicaid and medicare along with mandating that no companies deny policies to people with pre-existing conditions. Although this law meant to be comprehensive, there were no stipulations regarding genetic information or testing. As of now the only legislation in place is the aforementioned HIPAA, which protects genetic information from being accessed by health insurance companies. There are no moves to input the cost of genetic counseling or testing into health insurance plans by either the government or private insurance companies. So the choice of some individuals to seek genetic counseling and genetic testing is for their own preventative purposes, and often comes at their own financial cost. The access to these services is not equal. This brings up the issue of

financial/social class discrimination in regards to genetic testing. I will explore this issue with a case study.

In the United States breast cancer is the number two cause of deaths in women, second to lung cancer. It was discovered about 20 years ago that about 5-10% of breast cancer along with 15% of ovarian cancer is due to a mutation in either the BRCA1 or BRCA2 gene. Women with confirmed genetic tests with either of these mutations can undergo increased screening or prophylactic removal of the breasts and/or ovaries to mitigate their risk. This scientific discovery is complicated by the fact that the location and function of these two genes was first discovered by an independent company called Myriad. Based on their discovery they developed a medical test to detect the mutations. After their discovery Myriad was able to obtain multiple patents giving them the exclusive rights to any technology involved in their process of testing for BRCA1/BRCA2. When other institutions began developing and offering tests for BRCA1/BRCA2, Myriad had ordered them to stop. For 20 years now, Myriad has been the only company to be able to administer the test. As of 2013 Myriad was charging about \$4,040 for the test and had an annual profit of \$57 million. Other companies that wished to offer the test had set prices as low as \$1000-2300 for the test (Rosenbaum 2014). Although neither the Myriad test nor the other companies' tests are cheap, the difference in price is about 200 fold. This makes it clear that only individuals willing to spend the money could receive the possible benefits of this test. Without federal legislation concerning the patenting of genes and the limits of genetic testing in the private sector, the technology will become subject to the market. As the demand of testing goes up with only one or two companies being able to administer tests, therefore not changing supply, it is a simple law of economics that the price will soar. This will further build upon the already transcending and pressing issue of inequality in our society.

Eugenics

Although what we have seen thus far is that genetic discrimination in its infancy primarily affects access to life/disability insurance and has fallen into the snare of the free market causing socio-economic inequalities, the issues have not been life and death. Or so it may seem. At one point in the world's history before our knowledge of genetics had expanded, an idea so influential was proposed that it was found in both Nazi Germany and the United States. This was the idea of eugenics. Eugenics is popularly thought to be an ideal of the past, but I will explore the history of this ideology and link it to the present very tangibly.

Francis Galton in the mid 1800's, proposed the idea of using scientific selection to replace the Darwinian process of natural selection stating, "What nature does blindly, slowly and ruthlessly, man may do providently, quickly, and kindly. As it lies within his power, so it becomes his duty to work in that direction (Glad 2006). He asserted that because of modern medicine, selection is not defined by mortality any longer, but by the people who choose to reproduce. Unfortunately, the well endowed and intellectual people seemed to reproduce less, which horrified him. He saw that the impoverished and the diseased weakened society because their genetics makeup, which influences both their behavior and health, has led to their societal position. Ultimately, it is our responsibility as society to ensure that our future generations are free of these ailments resulting in a more prosperous society.

Eugenics has two methods that it proposes to improve society: positive and negative eugenics. Positive eugenics entails raising fertility among the genetically advantaged; this includes financial and political stimuli, targeted demographic analyses *In Vitro* Fertilization (IVF), egg transplants, and cloning. Negative eugenics entails lowering fertility among the genetically disadvantaged; family planning, abortions, and sterilization (Ludmerer 1972).

Eugenics history is most often associated with Nazi Germany. In 1895 Alfred Ploetz wrote a book entitled *The Fitness of Our Race and the Protection of the Weak* that coined the word “Rassenhygiene” or “race hygiene.” This led to the formation of the Society for Racial Hygiene in 1909, which eventually evolved into the Kaiser Wilhelm Institute for Anthropology, Human Genetics, and Eugenics (KWI) in 1927. The KWI insisted that racial purity was the only way for an organism to function correctly and that the people of Jewish ancestry were parasites to the German/Aryan bloodline. This first led to Hitler’s signing of the “Nuremberg Laws” in 1935. The laws stated that no Jew and Aryan could be married to ensure purity of the race and in 1939 the law was extended beyond preventing conception to eliminating any children in existence that the Nazis found not worthy of life. In 1939 Hitler signed an order that commanded physicians to evaluate institutionalized and ill patients and if they were determined to be “un-curable” to grant them a mercy killing. By 1941 this had led to the euthanization of over 70,000 individuals. Lastly, the ultimate act of Nazi eugenics was the Holocaust in which approximately 9 million people of Jewish descent in Europe were killed in concentration camps (Jackson and Weidman 2004).

Many people who claim to be true eugenicists say that Nazi Germany is the only case of eugenics being used maliciously, but that is simply not true. The effects of eugenics are much closer to home than one who is an American might be comfortable with. One of the most prominent American eugenicists was Madison Grant (1865-1937). Grant was also an author and lawyer. Grant was a member of many exclusive high class clubs in the Northeast that had many other profile members, many of whom were politicians including President Theodore Roosevelt. Grant published many works promoting eugenics in America and even proposed at one point mass sterilization, “beginning always with the criminal, the diseased, and the insane, and

extending gradually to types which may be called weakling rather than defectives, and perhaps ultimately to worthless race types” (Jackson and Weidman 2004). The correlation between his statement and acts that followed is not confirmed, but regardless many states soon passed sterilization laws. In 1907 the first law was passed in Indiana requiring the sterilization of “criminals, idiots, rapists, or imbeciles” (Jackson and Weidman 2004). By the year 1922, seventeen more states had followed suit and had passed laws primarily aimed towards those who had been institutionalized. By the time these laws were repealed between 60,000 and 90,000 Americans had been involuntarily sterilized (Jackson and Weidman 2004).

Although eugenicists have the ability to claim that the genocide and mass murder was not an original intention of eugenics, it also leaves the mechanism of achieving a superior society somewhat ambiguous to the idea of assuring the cessation of unfavorable offspring. On top of that, involuntary sterilization is much different than giving certain individuals incentives to not procreate. This is also not me conceding that their stated methods of reproductive control and genetic manipulation are any more ethical, but I wanted to illuminate that by leaving limits up to interpretation, such as with the Curse of Ham, leaves the fault in the hands of origin regardless of intention.

Eugenics, although in its scientific stance of disease, is correct (still unethical for the reasons cited above and to come), also falls to the fallacy of genetic determinism. Eugenicists believe that the behavior or a person is completely due to genotypes as if it is expressed like a phenotype. In recent years there has been an emerging field of neuroscience called behavioral genetics where scientists are now trying to determine genes or gene sequences that correlate with people of certain personalities and behaviors. This field is in its infancy, but has shown that there is some predictive power. Although there are influences and predictors to predispose behavior, as

we can plainly observe, environment and nurturing influences behavior just as or more heavily. This is evidenced by cases of identical twins. The case of Eng and Chang Bunker is one of the most famous cases about conjoined twins attached at the sternum. These twins shared the closest thing to identical heredity and also shared the same environment, yet one twin was “morose and enjoyed his liquor, while the other was cheerful and abstained from drinking” (Gilbert, Tyler, & Zackin 2005). This suggests that each of them interpreted their environment differently and therefore developed in completely different ways.

Eugenics may have lost its fervor after WWII, but its tendencies have placed a firm root globally in a new and much more subtle age of eugenics. In 1993 in Colorado a bill was put into consideration by the legislature to mandate testing of Fragile X due to the fact that affected children cost the state well over a million dollars during their lifetime (Barash 2008). This case shows the direct correlation of a human life with a monetary value. Eugenics stresses functionality in society, and a primarily capitalistic society where self reliance and individualism is stressed, individuals that cost money without the ability to reciprocate is the modern day definition of dysfunctional. Although I am sure these local politicians were not eugenicists, they inadvertently supported and almost reincarnated the practices.

Another case of modern eugenics is in the directive genetic counseling practices in Israel. Many well documented genetic disorders occur within a specifically population of people of Ashkenazi Jewish descent. The high frequency is a result of inbreeding as well as the size of the community throughout history. Even if members were not marrying within the family, the community was so small that the gene pool lacked much diversity. With the advent of genetic testing and counseling, Rabbi Joseph Eckstein founded the international genetic testing program in 1985. Rabbi Joseph created this program in hopes to eradicate all the genetic disorders in his

population. The program tests a large number of orthodox Jewish students and couples. In the case of the couples, if only one prospective spouse is shown to be a carrier, couples are not simply advised to not have kids (which is already a violation of the ideal of genetic counseling), but they are advised not to get married. This ideal has laid siege in the way Israel goes about genetic screen as evidenced by the statistic that the country has the highest screening rates in the world. Naomi Stone, a Jewish writer, expressed her feeling towards the issue: “Perhaps, the disease[s] can be eradicated entirely from populations...who could reasonably express qualms?...I am Ashkenazi Jew, and I know that it is my obligation...” (Glad 2006).

Modern day eugenicists have shifted some of their attention to *in vitro* fertilized egg implantation. Currently the process of artificial insemination involves fertilizing multiple eggs in a laboratory and implanting them into women from infertile couples. Scientists use multiple eggs because the chances of an egg being able to implant into the uterine wall is low. As technology from genome testing advances to the point where we can screen fertilized eggs we will be able to determine which eggs we want to implant. Also as germ-line gene therapy arises eggs and sperm cells could be manipulated before even going through fertilization ensuring that all the fertilized eggs are “favorable” (Glad 2006).

Eugenics places society above the individual, but fails to ask the question: how can we determine what genes/traits are favorable over others? What gives anyone the right, both scientifically and philosophically? With the completion of the Human Genome Project and the departure to define the function of genes with ENCODE project we will be discovering new genes along with the phenotypes associated with them constantly for years to come. I also believe that the discoveries will never cease because mutations occur for various reasons everyday and that is simply a part of microevolution. Eugenics suggests that human evolution is

somehow directed towards some ideal, but evolution is quite the opposite. Eugenics follows the Aristotelian ideal of teleology, which has been proven not to be relevant in the study of evolution. Evolution occurs due to chance and from environmental factors. Also, setting science aside for a moment, the alterations in our genes provide the diversity we see within the global society. With the never ceasing process of evolution and the massive diversity in the world, if we buy into the idea of eugenics how could we ever define what the ideal human is?

There is evidence that many diseases that are genetic arose during human evolution as a result of external diseases caused by pathogens (virus, bacteria, and other external sources of disease). For example, sickle cell anemia is a condition that is seen by many as a degenerative disease that affects oxygen delivery in the body, causes painful blood flow, and can lead to amputation of extremities and early death. Interestingly, the condition provides individuals with a heterozygote genotype for the disease immunity to malaria. Heterozygote individuals suffer from mild sickle cell anemia, but gain the immunity from their altered blood cells. Even more interesting is the fact that the genetic condition is found in high frequencies in areas that have been affected and are still affected by malaria.

Although many people would try to argue that eugenic tendencies have lessened ever since its associations with Nazi scientist, I would argue that it still exists in new and unique forms. I believe that the two most prominent forms in today's society both have to do with reproduction. The first is a free-market driven eugenics in the market of *In Vitro* fertilization (IVF) and the other is the stigmatization towards disabled individuals and their parents whom did not elect to prenatally test or abort diagnosed fetuses.

In recent years old eugenics has taken root in an intricate part of our culture, consumerism. The advocates for this newer form of eugenics, refuse to admit that it is eugenics because it is no

longer coercive and leaves the freedom of choice. Although I concede that the coercion associated with the past eugenics was troubling, I do not think it was the only concerning aspect. I believe that underlying all eugenics is an idea of genetic exceptionalism in some individuals over others, which still exists today. There was story on the news in 2007 about an advertisement posted in some Ivy League University's newspapers "...offering \$50,000 for an egg from a young woman who was at least five feet, ten inches tall, athletic, without major family medical problems, and with a combined SAT score of 1400 or above." Eggs like this have been auctioned online since then for prices ranging from \$15,000-\$150,000 (Sandel 2007). Many of the egg and sperm banks participating in practices of collecting "designer" specimens have claimed not to be doing so for eugenic purposes when asked; but this begs the question, what is the difference between designing children for eugenic purposes as compared to designing children in line with what the market dictates? Although these practices are not scientifically grounded because in no way is genetics as simple as "what you see, is what you get," the underlying ambition to control the outcome of a child to have traits the parents deem as good and to carry out the wishes of the parents, is extremely troubling. This shows the overwhelmingly common misconception of genetics, and without proper understanding, a technology as powerful as genetics becomes more costly compared to its possible benefits.

With the birth of prenatal testing we have been able to allow parents to have knowledge of certain genetic conditions their child may have before they are born. I am not arguing that this is not a useful tool because this knowledge can provide parents with information that may lead them to different reproductive decisions, those being carrying the baby to term or terminating the pregnancy. I agree that some parents that are in certain financial and social/psychological situations are probably not fit to raise these children that require more care and their choices to

terminate or put up the child for adoption are logical. That being said, those kinds of situations are not the norm anymore. A perfect example of a disease that falls in this category is one that I have already discussed, Down's Syndrome.

Currently there are two choices when a fetus is diagnosed with Down's Syndrome: 1) continuing the pregnancy or 2) terminating the pregnancy (Bianchi 2012). In 2010, an annual report of pregnancies terminated after positive diagnosis in the U.S. revealed that 67% were terminated. More staggeringly, in the Netherlands there was a 93% termination rate (Veirweij 2013). Eugenicists must be very pleased with these numbers because the less Down's Syndrome individuals that are born, the less our society is burdened by them. Although I asserted that we as a society have tendencies, I am simply letting the numbers speak, but I am sure that some of the people that underwent termination did so with good reason and a heavy heart.

If the genetic abnormality for Down's Syndrome like many eugenicists believe is just a mishap in evolution that has simply produced mentally and physically handicapped individuals, why would we ever want to subject anyone to be born that way if we had the choice not to? Recent research on Down's Syndrome individuals has once again shown that evolution is not so easily placed into terms of rights and wrongs. Bratman et al. (2014) release a review article on the research done on Down's Syndrome individuals in regards to cancer risk and showed that Down's Syndrome individuals had higher risk of leukemia, but in the case of solid tumors, their risk is very uncommon with lower than expected age-adjusted incidence rates. There has been much interest in finding either environmental or genetic causation for this unusual risk. Some research is looking into how their extra chromosome may supply a third copy of a tumor suppressor gene, making it extremely unlikely to have a triple knock-out of all the copies of the tumor suppressor gene's function. DeYoung, Tress, and Narayanan (2003) found a variant of a

tumor suppressor gene on chromosome 21. The gene SIM-2 has been correlated with solid tumors, but is not well documented. It has been seen that SIM-2 becomes overactive causing increased cell-growth and tumor formation. DeYoung et al. (2003) predict that the triplication of the gene has promise to be associate with the variation in risk in Down's syndrome individuals, but they also observed that it is an abnormal form of the gene overall.

As discussed in an earlier chapter we have seen that genetic therapies are sound in theory and are possible. Personalized medicine could eventually treat these fetuses with Down's Syndrome giving the mothers another choice as far as reproduction goes, but currently the choices cause devaluation of these fetuses. Unfortunately, in recent literature all I can see is that research has been primarily going towards making genetic testing techniques more accurate and less invasive. This highlights an issue of priorities with genetic testing in general. I do concede that genetic testing has much to offer especially when it comes to early diagnosis of cancer with increased screening of hereditary cancer in individuals or allowing parents to prepare to raise a handicapped child. Although we should have ample research in providing this information we still cannot set aside the priority to cure or treat a disease. A fear that I have is that since testing for many particular diseases often is discovered far before a treatment, that the incentive or intention to find a treatment could be lost. As of now the incentive to have less of these individuals in our society rather than treating them seems much more prevalent.

To close out this section on eugenics I must discuss the fact that a majority of genetic disorders that have been discovered are not debilitating like Down's Syndrome, Tay-Sachs, or Huntington's Disease. Many genetic mutations lead to cancer, cardiovascular conditions, diabetes, etc. Children can be born with a disease as severe as PKU and be treated by diet change to be able to develop normally. My point is that the people, besides the risk of certain health

issues, are functional and contribute to society. Eugenics would mark them as crippled and not worthy of society. We would sacrifice a majority or possibly all of the human population because I can say with confidence as projects like ENCODE continue we will find genetic causation for a majority of human ailments. Humans are not the epitome of evolution and will never be perfect, but perfection should not be our goal, rather respect and care for the dignity of our fellow man.

Treatment vs. Enhancement

One of the ethical issues within genetics has recently looked to the future with the topic of treatment versus enhancement. Although the technology is not present currently, it is scientifically possible. That is why this issue is of much importance to discuss even though it is premature because many precursors that show an inclination towards enhancement exist in the present in the direction of our research as well as our attitudes towards priorities, overcoming obstacles, and parenting.

One of the classic cases in this discussion is that of muscle enhancement. Researchers have been in the process of developing a somatic gene therapy to help alleviate or cure the genetic disorder muscular dystrophy. Muscular dystrophy, is genetic disorder found in individuals that lack a gene coding for a protein that facilitates the repair of muscle cells, this leads to muscular deterioration and eventual paralysis in affected individuals. The genetic therapy in development has been tested on mice that have been manipulated to lack the same gene and has shown promising results where the muscles grow back and do not deteriorate with age. The researchers in this field hope to eventually employ the therapy on humans. Interestingly, this research has not only caught the attention of afflicted individuals, but also athletes seeking to gain a competitive edge over their opponents. I know what many of you are thinking, is that not so different from steroids, and are not those illegal? Technically, yes, but

there has actually been studies done showing that performance enhancing drugs have become so common in the field of sports that the regulation has become almost impossible. On top of that, many athletes feel as if they are “playing naked” if they do not join in when all their competitors are using the drugs (Sandel 2007). Our society in general has this hypercompetitive attitude, and say hypothetically if the gene therapy for muscular dystrophy is shown to be safe on people without the disease as well? We all like to think the reason for regulated performance enhancing drugs is fairness, but in fact the main reason is their safety. So if the genetic therapy is safe, would it be ethical? Let us explore a couple other examples before answering this question.

Other situations of treatment versus enhancement have been concerning memory and height. There has been research into treatments for Alzheimer’s Disease that entail inserting an extra copy of a gene associated with memory. Again this treatment shows great promise. In this case the attention of big pharmaceutical companies in search of memory enhancing drugs was captured. With the baby-boomers reaching an age where their memories naturally deteriorate, the market for a “viagra for the brain” would be huge (Sandel 2007). One caveat to this is that this genetic therapy has shown to permanently alter the persons germ-line, meaning that the extra gene will be passed on to offspring. If my predictions are correct, if this drug ever came into the market, the price would be high. Could we possibly make another class split by creating a memory enhanced upper class and an unenhanced memory lower class? If we make access available to all, does this morality of the situation change?

Lastly, I want to discuss height. This example is not exactly genetic, but pertains to the subject nonetheless. In the 1980’s a hormone therapy was discovered to help children born with hormone deficiencies making them shorter than normal. By 1996, 40% of the use for this therapy was “off-label” meaning it was being administered to normal children, whom were deemed too

short by their parents. Whether for aesthetic or reasons of allowing a child to be more athletic, this situation is pretty clear cut, showing how parents place their judgements of themselves on their children and treat their kids as a means to achieving their own ambitions. The worst part is that hormone therapy on normal children is done without hesitation, most likely due to the market. Again, this exemplifies the hyper-competitiveness of our culture. I think this style of parenting can be seen outside the realm of medicine as well with parents that train their kids as olympic athletes from the day they can walk, parents that have to be asked by universities to go home or stop calling because they have to assure their child is on task, etc. A frightening statistic that makes this come to life is that today, a large number of sixteen-year-old baseball pitchers are undergoing elbow reconstructive surgery; a surgery once only done on major league athletes (Sandel 2007).

I posed many questions above, all to build to a point. All those questions are the wrong ones to be asking. We should not be concerned with the safety, fairness, or financial access to these treatments, rather we should be concerned about why we are using medical treatments for non-medical uses. These treatments were all originally developed to restore health, cure or prevent disease, or repair injury. I understand that this stems from the idea that we as humans should always seek to improve, and I agree I do not think people should stop learning when they are out of school or should not exercise just because they are getting older. I also understand that most parents are not ill-willed in their efforts to help their kids, but the role of a parent is to facilitate the growth their child in order for them to be able to pursue their own ambitions, rather than those of the parents. There comes a point though, when we are crossing over from striving for improvement and striving for perfection. The questions we should be asking are: should the ambition be to perfect ourselves and try to mitigate the effects of chance? What are the

psychological and social consequences of this concentration on finding flaws, rather than achievements or gifts? What does this say about our humanity, if we are so afraid of our own human nature? I cannot attempt to answer these questions both because of the constraints of time and because these questions are beyond my current knowledge. That being said it does not take hours of research to see why these questions are troubling.

Readdressing the paradigm with Casuistry

Now that I have discussed the issues of discrimination present in the past and now due to genetics, I will go back to the paradigm I set-up in the beginning of this chapter. I proposed a paradigm where normal individuals and bald individuals were applying for the same job with the same qualifications and the two possible outcomes that showed either the absence or presence of discrimination. Like in any application there must be qualifications that are met for someone to be considered for the job. The job that is in consideration in my discussion is that of being a human worthy of value and respect. Classically, value and respect would be seen as natural rights present from birth until death. What I see as the underlying goal of the agents behind genetic discrimination, eugenics, and other forms of discrimination is to change the qualifications. That is putting it kindly, but in reality they are being prejudiced, which is simply the valuing of certain individuals over others on subjective grounds. Advocates of genetic discrimination, try to argue that their ideas are objective because they are grounded in science, but they are mistaken to view health as something that can be quantified and set to a spectrum. A person could be the healthiest person genetically, but could get in a car accident and be paralyzed, has that person suddenly lost his/her value to society? Is there any difference if something happens at birth or during life? Who has the right to decide what genotypes and phenotypes are better than others? After employing casuistry I have come to the conclusion that

the current state of attitudes towards genetics lies on the side of the paradigm with the presence of discrimination.

That being said, genetics has brought some nuances to the table of discrimination. Typically, discrimination is based on stigma towards things that are out of control of the victim such as race, ethnicity, religion, socioeconomic class, etc (I include religion and socioeconomic class because although they are changeable, people are most often born into them). Genetics has introduced the freedom of choice. Just as athletes that do not use performance enhancing drugs or supplements are seen as “playing naked,” there is a growing attitude towards those who do not test themselves or their children, as “flying blind.” This nuance can be summed as the burden of choice. What is so subversive about this burden of choice is that there are stigmas that discourage either decision. On the side of choosing genetic testing, insurance, financial burden, and psychosocial consequences weigh down on the individual or family, while on the side of not using testing has the stigma of “flying blind.” These leaves very few options for the people that find themselves in this conundrum. Before assumptions are made, I do not believe that this is justification for the cessation of any further use of genetic testing because I do see that there are many benefits to be taken advantage of now and in the future that will innovate the field of medicine. I believe that right now, we have found ourselves in an opportune time to be proactive because the use of genetics is fairly limited. In order to solve this issue of genetic discrimination, both factors on both sides of the choice must be addressed.

Brave New World and Science Fiction

If we look around us, many of the things that we see as everyday items that we may take for granted, may have never seemed possible in just a couple generations past. Science fiction writers have always sparked the imagination of their readers with fantastical inventions that had no evidence in their time, but time and time and again, these fictional items have become reality. People often cite lunar exploration as the epitome of science fiction come to life, but that example still has a sense of grandness. Would it surprise you if I said that scuba diving or escalators were examples of science fiction come to life? Well they are. Scuba diving was first conceived by Jules Verne in his novel *20,000 Leagues Under the Sea* from 1870. In the novel *When The Sleeper Wakes* by H.G. Wells published in 1899, Wells described a futuristic metropolis with “moving walkways” a.k.a. escalators. Science fiction is often taken for granted as simply novels to appeal to the imagination of the readers and entertainment, but we must remember it still falls into the realm of literature and one of the most important literary devices is “theme.” “Theme” is the exploration of the human condition. Science fiction writers simply make futuristic or far fetched settings for their stories, but often have subtle or strong undertones portraying their views on society as it was, is, or could be. George Orwell in his dystopian novel *1984* terrified readers with his theme “big brother is always watching,” but readers at the time of its publishing were reassured that cameras watching them were a myth. Low and behold only a year or two later closed-circuit

television (CCTV), also known as surveillance camera systems were introduced. The invention of the technology is probably not attributed to Orwell, but this shows that authors are more than often astute with societal trends or tendencies. *1984* is often cited as one of Orwell's many critiques of capitalism, and intriguingly surveillance cameras were first made commercial by the United States. So as we can see, fiction can often be a powerful representation or predictor of society, but more times than not, critiques about society become relevant after they have come true. Aldous Huxley in his novel *Brave New World*, written in 1931, paints a powerful picture of a utopian society based around complete control, including genetic control, by the governing powers. There are many complex themes in this book that I will call upon in my discussion of the future for genetic diagnosis and technology.

The novel *Brave New World* by Aldous Huxley explores the dichotomy of ideals between the two societies at play in the novel: The World State and The Savage Reservation. Throughout my discussion of this topic I will primarily reference the characters John and Mustapha Mond. Mond is the character used by Huxley to represent the leader and instigator of The World State, while John is a character often referred to as "the savage" because he has his origins in the reservation.

The World State represents a sterilized society where the culture of mass production has proliferated taking over the thought process over everything. The planetary motto of the World State is "Community. Identity. Stability" (Huxley 1946). The World State and their controller, Mustapha Mond, see these three tenants to be central to a civilized society where everyone is "happy." These ideas link to how the World State treats Henry Ford as almost like a deity figure that created their society. Obviously the idea behind Ford automotive, to produce reliable and similar cars in large quantities, is what the World State strives for; they wish to have a society

that is reliable and without change or surprises. People are cloned and brainwashed to “think” the same and live without sickness or struggle and die around the age of 60. The people never get married, do not have families, and have sex for recreation. In the society of this novel, the philosophy of mass production has “at last applied to biology” (Huxley 1946).

The Savage reservation is a move by Huxley to presents a completely opposite society. The Savage Reservation is one that is supposedly uncivilized and is removed from science for the most part. In the reservation traditional family units exist along with sickness and old age. People die of sickness primarily. They also live in the countryside, which is an area that the people of World State are conditioned to hate because of how close nature is. I assigned the Savage Reservation as being driven by truth and beauty due to the fact that it does not try to manipulate their people and the nature around them; they live with it.

Mustapha Mond happens to be a character that was born pre-World State, so at one point he also lived in a society like that of the Savage Reservation. The World State views families and relationships as almost pornographic and Mond explains that its because the old “...world didn’t allow [people] to take things easily, didn’t allow them to be sane, virtuous, happy...they were not conditioned to obey...all the disease...uncertainty and the poverty – they were forced to feel strongly” (Huxley 1946). This to me implies a reference to the common phrase that “a machine cannot work unless all its parts are working properly.” The World State craves for comfort and stability, but cannot do that if its people are not also in that state constantly. For these reasons, Mond asserts that war is a direct result of feeling too much. Mond says, “What’s the point of truth or beauty or knowledge when the anthrax bombs are popping all around you?” (Huxley 1946). I see this as a perfectly innate reaction when put under such a threat like anthrax bombs, to put effort into trying make life a little bit more quiet and controlled. We often do similar

things in our society today such as putting up cameras everywhere to feel safer with crime, but for me there has to be a distinction between precaution and overcompensating for a problem. In the World State the people were willing to sacrifice their own freedom to assure their security.

On top of freedom, I think we lose something that makes us human rather than instinct driven animals, we lose our obligation to discover ways to deal with our strife, with effort and moral training. Instant gratification is not real happiness; it subsides and leaves us empty and less human. John, puts my attitude in words best when he says, “whether ‘tis better in the mind to suffer the slings and arrows or outrageous fortune, or to take arms against a sea of troubles and by opposing end them...” we can not choose to neither suffer nor oppose (Huxley 1946).

Huxley’s vision may be based in fiction, but neither the basics of the technology nor the society seen in the novel are foreign in anyway. For example, the notion of cloning is not even shocking in the scientific community anymore because it was successfully done in 1997 on a sheep named Dolly (Gilbert, Tyler, & Zackin 2005). Although the book shows some science analogous to our developments today, the book explores our societal values and tendencies in a stark, but unfortunately realistic manner. In fact, we have seen these themes continue to be portrayed by a number of popular culture and media all the way to the present; such as *Gattaca* (movie), *Star Wars: Clone Wars* (movies), *Oryx and Crake* (book), and *Bioshock* (video game).

The themes of excessive comfort, stability, societal coercion by uninformed/corrupt powers, and mass production in *Brave New World* tie back to the philosophies of eugenics and genetic discrimination that I have described earlier. The World State’s mantra of “Community. Identity. Stability” seems to be opposed to our ideals of hyper-individualism seen today, but I believe that our society’s technology, media, consumerism, and culture has undermined and accentuate the unfavorable ideologies of individualism at a systemic level hindering our freedom

to think and act as we like. Our individualism has fostered an obsession with control and unwelcomeness to things that are unbidden. Therefore our society has placed value on consistency, convenience, efficiency, quantity, etc.

If this sounds familiar, it should. Look at our news outlets. A majority of the stories are now under the umbrella of entertainment, sports, fashion, etc. You name a trivial thing, and our media probably covers it. These are subjects we find comforting because they foster complacency. While many people would argue that many news outlets still report the real news by talking about crime and international crises, I would argue that those stories are often biased toward raising national pride as well as giving the unevidenced perspective that the world is becoming a more dangerous place; while in fact crime has decreased significantly (about 10%) within the last 40 years. As a result, we go out and invest in pricey home security systems and begin unintentionally profiling certain groups of people as criminals (Owens 2015). By tricking ourselves that we can avoid crime by paying for security and having the ability to “predict” who will commit crime we further foster our sense of control. In fact, statistically profiling groups of people for crime that are not actually committing the majority of crime.

To give some other examples. If you have ever had a small talk with someone that you do not know very well, what are the most common subjects of talk? The weather, a popular movie, and...gas prices. We are so attuned with the price of gas that I have met people that even notice when the price has negligibly changed +/- a few cents. All the while we are concerned with this rather than climate change or the certainty of the finite nature of fossil fuels. The thing is that those kind of thoughts are not comfortable, while discussing gas prices gives us the illusion of strife. If we want to find examples that directly correlate with the novel *Brave New World* consider online dating or the increased use of drugs and alcohol. The generation entering

adulthood now has been deemed by many as the “hook-up” generation because we are more concerned about the number of intimate partners we have than actually finding love. We also do this in a cold, calculated, and superficial way, exemplified by computer apps like “Tinder” which simply allows you to scroll through someone’s photos and either say “yes” or “no” to a “hook-up.” Along with this, our entertainment industry keeps producing romantic movies and television shows where characters are chastised because they say the dreaded, and I do mean dreaded, words “I Love You” too early on in a relationship. Just like in *Brave New World*, sex has become the norm, while love and family has become taboo. In the case of drugs and alcohol, that is an obvious analogy to “soma,” the drug taken in the novel to disrupt riots and to take “holidays” the moment stress rears its ugly head. Self-medication, exemplifies our idea that we have control over our own emotions, while in fact it is truly an act of forfeiting them.

The societal themes within *Brave New World* are too familiar and with the advent of genetics coupled with this clouded and unethical ideology that underlies much of society, the outcomes of the book are not so farfetched.

Just like in the novel, we will never actually obtain dominion or mastery over our nature. We see this by how stress still exists, requiring the invention of soma, which also inadvertently shortens lifespan so that people never have to face old-age. Or in a more recent science movie named *Gattaca*, which has been said to be a modern re-telling of *Brave New World*, we see one of the people who are genetically superior people attempt and successfully commit suicide. With genetics we can easily fall into the fallacy of being able to control our’s and our offsprings’ future health and abilities. While in fact we are all subject to the genetic lottery as well as chance. As a result, our illusion of control can place a new found burden of perfection on those said to be superior. The reason the character in *Gattaca* had committed suicide was because he

had not lived up to his projected potential when he got second place as a professional swimmer. This burden of perfection would stem from the psychological and social pressures to fulfill predetermined potentials, the inability of people to relinquish control over parts of life that should or cannot be controlled, and our insatiable hunger to be satisfied. Even if we manage to make our lives comfortable or long and free of disease, we would still be subject to our environment and external dangers. “There is no gene for fate” (*Gattaca* 1998).

We see here, although one of society’s greatest assets is the ability to unify its people under law and order, culture, and values, that once the underlying principle of any of these are distorted, it becomes a plague on society. Therefore, Huxley has illuminated the tendencies and capabilities of our society seeded deeply within the ethical issues I have already addressed. So the urgency to determine how we shall use the technology and to what institution(s) the power over will fall must happen with haste and without room for interpretation. Lest we have to react to a catastrophe that could take us decades to even mitigate, much like our seemingly eternal struggle with racial discrimination.

Genetic Education of Professionals and Public

As I have said throughout this essay, the need for a protocol of sorts to be defined towards the use of genetics in diagnosis is and will be essential as it becomes more prevalent. The ethical problems that are possible are dire, but as of right now the ethical issues are minimal because of the small group of people who are involved: professionals and patients. The size of this group will inevitably grow as the importance of genetics in the clinic grows in a field pushing towards preventive health care. Like any other solution, we cannot find a consensus unless a majority of people are on the same level of understanding and generally share the same sentiment. Therefore, the education of genetics for health care professionals as well as the public and public institutions, during this time where the use of genetics is fairly limited, will put us on a good track to avoid further ethical dilemmas.

Advances in genetics during the last few decades have resulted in a variety of health care providers specialized in genetics and genetic counseling including genetic counselors, medical doctors and PhD geneticists, and genetics nurses (Brierley et al. 2010). Later advancements like next generation sequencing that have increased the efficiency and accessibility of genetic testing recently have prompted recommendations that genetic counseling and testing be provided by all healthcare providers in fields that encounter genetic disorders. This has raised much concern in specialists because other healthcare personnel not trained thoroughly in the field may not understand the

complexity of prescribing and interpreting tests. On top of that, untrained personnel may not be sensitive to the psychosocial consequences of certain results and care-plans.

Brierley et al. (2010) explored cases where there have been negative outcomes in the field of cancer genetics when patients were not provided with pre-testing counseling by a certified genetics professional. The discovery of the BRCA1 and BRCA2 mutations and the strong correlation found with breast cancer started the trend that all healthcare providers should be able to use genetic testing. Problems have arisen time and time again because uncertified health providers are not up to date with the literature and the nuances in the field leading to unnecessary or wrong testing, misdiagnosis, or misinformed long-term treatment options. There are a few cases that they discussed that I believe are the most powerful.

In one case, a 49-year old breast cancer survivor sought for genetic counseling because she had been diagnosed when she was only 42. She had met with a genetic counselor, and after analyzing the family history, the counselor said that she had a strong chance of being BRCA positive. The insurance company did not allow her to order testing through the genetic counselor and instead referred her to another facility. The patient underwent testing at the facility and was found to be negative. Afterwards the patient sent the results to the genetic counselor she had seen and the counselor noticed that the facility ordered for testing of three common Jewish BRCA mutations because they had those patented. This patient was not of Jewish descent. The patient then elected to pay out of pocket to get testing through the genetic counselor for full sequencing of the BRCA genes, and she was found to be BRCA2 positive. The patient underwent prophylactic surgery to remove breast tissue and her uterus, and now her family was informed that they may have the BRCA2 mutation. Without the correct testing the patient may not have elected prophylactic surgery or even if she did not do that, she would have not have underwent

increased screening, making her risk of relapse much higher. Furthermore the patients expenditure was about \$3,000 higher than if the right test was ordered to begin with (Brierley et al. 2010).

In another case, a 52-year-old man with a family history of colon cancer was recommended by his gastroenterologist (GI) to be tested for Lynch Syndrome. The patient did not want to, but during a later colonoscopy appointment while sedated he was asked to sign a form, since he was not able to, his wife signed off unknowingly. The GI doctor had sent off a sample for genetic testing and found that the patient had a mutation linked with Lynch Syndrome. These results were then mailed to the patient. Written on the letter sent was a note saying, “Your children need genetic testing when teenagers. You will need a colonoscopy in one year” (Brierley et al. 2010). His children at that point were already in there mid to late 20’s. On top of that the doctor did not mention that Lynch Syndrome was also associated with increased risk of uterine, ovarian, and other endometrial cancers. At no point was the patient referred to a genetic counselor, and the patient suffered from depression and anxiety, ultimately stating that he would have never wanted the test. This case is a perfect example of how a positive result is not always favorable. If the patient had met with a certified professional, he/she would have done significant analysis of the family history to determine that the patient may have been at risk for Lynch Syndrome before testing. Also the counselor would have provided all the information about Lynch Syndrome for the patient before hand, and only would have ordered the test if the patient had chosen to. So although the GI doctor may have had the patient’s best interest in mind, he/she did not take into account that genetic testing requires mental preparation and deliberation by patients and with their families if applicable. Many patients even with all the information may elect not to get tested and that is ultimately their decision, and it is the responsibility of the

provider not to be directive because ultimately, the results only affect patients and their own family. And in cases that patients do elect to and regret the decision, counselors and other geneticists are trained to provide psychosocial support.

Some have argued against the assertion that only certified genetic counselors should be able to provide testing simply because there are not enough of them. These arguments are for the most part unfounded for two reasons. In large urban areas there are often many qualified professionals and in some cases the wait time to meet with a primary physician can be longer than the wait to meet with a genetic specialist. In rural areas or more sparsely populated areas there is the option to access genetic counseling through satellite clinics or phone/internet based services (Brierley et al. 2010).

I do concede to the fact that as the knowledge of genetic disorders becomes more well known and clinically applicable, the patient load will grow and at least one of two things will need to occur: 1. an increase in certified professionals and 2. more genetic training during the education of health care professionals. Currently, the pay grade for certified genetic counselors is quite low considering the amount of extra school required, so in order to increase the amount of people who pursue this career, the compensation may need to be increased. Also, as of now most medical school curricula do not include genetic training in testing and interpretation, so a revamp of the curricula would be required.

I believe that a combination of these solutions must be implemented. Realistically, the field of genetics is quite complex and constantly evolving, so clinicians would be overburdened if they were responsible to keep up with the literature as well as the literature pertaining to their own field. That being said I do believe that there has to be sufficient training in interpreting patient family history and risk assessment so that clinicians know when to refer patients to

certified counselors. A study was conducted by Baars et al. (2005), which subjected over 2000 medical students nearing graduation to take a genetic assessment test with questions divided into categories (essential, desired, and specialized). They found that in their three experimental groups that anywhere from 3-26% of the medical students would have passed the essential knowledge test. This knowledge was deemed by geneticists to be essential in the daily practice of non-geneticists with the growing relevance of genetics in all fields. That being said, the curricula taught in schools of medicine should include basic genetics, advanced genomics and interpretation, as well as a ethics and counseling in order to instill the values of non-directivity and sensitivity to the psycho-social well-being of the patients. Overall, there must be a more synergistic approach in the field between increasing the knowledge in clinicians as well as increasing the amount of certified genetic professionals to deal with increase patient load.

There has also been much concern as of late that much of the information being provided to clinicians about genetic testing, has been from direct-to-consumer marketing firms with special interests because they are associated with labs that have patents on genes. Often this information is misleading, incomplete, or inaccurate (Dressler et al. 2014). In order to avoid this I believe that federal oversight over genetic tests and advertisements is needed as well as laws prohibiting patenting of genes. Without the ability to patent genes, genetic technology can hypothetically be exempt from the pressures of competition and the market. This also furthers the necessity of the education of genetic testing during graduate school in a competent, ethical, and nondirective manner.

In my discussion of genetics within the field of healthcare professionals I would be remiss if I did not mention how genetics will influence and should be implemented within the field of nursing. Although we often equate health care with medical doctors, I do not think health

care could operate without the nurses. This is because nursing training along with the nurse-patient relationship is so different than that of physicians. Nursing education in contrast to physicians, places more concentration on maintaining, facilitating, and monitoring health, rather than purely treatment. This fosters a very different relationship with patients where nurses have much more frequent and regular interactions. This relationship requires that nurses not only be well versed in physical and medical sciences, but also social and psychological science. This is so nurses can approach patients of all ages in a holistic manner tailored to their emotional, physical, intellectual, spiritual, and other personal needs. Because of this context of nursing, nurses could be the key in translating genetics into health care, by being translators and interpreters for patients.

As of right now, nurses in general, do not receive adequate genetic and genomic education, unless they become a Advanced Practice Genetics Nurse (APGN) (Morrison 2013). This profession is a nurse practitioner level position meaning that nurses require a master's level education in genomics and genetic counseling to be an APGN. I believe that all nurses should be trained to assess family histories and to determine whether a patient should be referred to a certified geneticist or APGN. Daack-Hirsch et al. (2012) performed a study with the goal of determining a practical first step in integrating genetics into the nursing curriculum. The researchers believed that the first step in ensuring that genetics is incorporated into the curricula was to show nursing faculty and students what their level of understanding of genetics was. The researchers employed a Genetics Literacy Assessment Instrument (GLAI). The GLAI was a standardized test written by geneticists to test literacy in essential genetic and genomic information. Daack-Hirsch et al. (2012) suggest that this assessment should be done by all nursing programs in order to illuminate and motivate them to address the discrepancy in genetics

knowledge in their profession. This conclusion is very similar to that of the study Baars et al. (2005) discussed above about testing the proficiency in genetics knowledge of medical students.

Although I have asserted that it is the responsibility of the health and research professionals to be knowledgeable in the field, it cannot be ignored that common public misconceptions do play a large role in unnecessary or mistaken use of genetic testing. The public perception of genetic testing is that it is a clear cut test with either a positive or negative result, therefore meaning the results are easily interpreted and dealt with. It does not account for how case specific the field is and that although many things are testable, that treatments are not necessarily going to be easy. It is also notable to say that these misconceptions exist in public institutions such as the government and insurance companies. Therefore, in the near future public health professionals in policy-making positions must work to implement programs that spread awareness to the general public about genetics as well as be active advocates for proper and ethical use of genetic technology and information.

Allen, Senecal, and Avarad (2014) wrote a paper outlining the degree of public engagement that public health authorities should begin working towards. The authors do a very good job of outlining the caveats and issues that complicate moving educational initiatives too quickly such as "...reducing trust in public health authorities; increasing public anxiety; demotivating lower-risk individuals from participating in preventative programs; perpetuating the idea of genetic exceptionalism; and contributing to stigmatization and/or discrimination" (Allen, Senecal, & Avarad 2014). Therefore they propose the purposeful withholding of genetic information not immediately relevant to the public because of lack of sufficient research and the unknown implications for some newly discovered information. This withholding of information could spark conversations within the realm of health policy making, and allow for platform of

ensuring that everyone is on the same page scientifically, ethically, and legally. That being said the public should still be informed of this technology that has far reaching implications, but just in a methodological manner. In the present, public health professionals do not think we should dive into trying to launch campaigns advertising genetic counseling or genetic testing and such before we ensure that people understand the purpose of genetic testing. The first step to take is to combat the common attitude of “use it or lose it,” or in the case of genetics, “why should a person not be testing if a test exists?” This attitude is a direct result of allowing the science to guide our decision making while our overall understanding of the subject is behind. An analogous example is the use of antibiotics in farming. During the industrialization of farming, farmers stumbled upon the fact that livestock treated with antibiotics, whether they were sick or not, would grow bigger. Instead of showing discretion and ensuring that this practice was safe, this soon became a regular practice and now we are having many problems with bacterial resistance to antibiotics. So in order to avoid a situation like that, we must educate the public that there is a right not to know one’s genetic information and that it should not be frowned upon to choose not to. I think once this consensus is achieved that a public initiative can move forward to educate the public more about basic genetics and also genetics in healthcare.

Conclusion

Genetics is the new frontier of science and medicine and will be constantly evolving for the foreseeable future, and I hope this work has inspired contemplation and the necessary dialogue needed to ensure the safe and ethical use of genetic technology from now on.

What I would like to be taken from my comparative section of past discrimination with genetic discrimination is that timing is everything. We, as a society, are generally reactive rather than proactive, but right now we have the opportunity to make sure that genetics does not become subject to corruption by prejudice. Although we are in this opportune time, we also are very vulnerable because our moral and ethical understanding of genetics, is far behind our understanding of the science. History has shown us that the timing of an idea can cause it to spread through society like a germ or like cancer, and once it has spread we can only mitigate it so much, but ultimately we are fighting a losing battle.

I can foresee many critics trying to draw analogies between genetic disorders and issues of vaccination. Public health advocates and professionals have been very concerned as of late with the resurgence of parents refusing to vaccinate their children due to illegitimate scientific research. So the argument being made is that people have social responsibility to society by strengthening herd immunity to these communicable

diseases that we have vaccinations for. Eugenics and tendencies to fall into that train of thought draws a clear analogy between infectious disease and genetic disorders. However, genetic disorders are vertical in essence meaning they directly affect individuals and their family and not society as a whole.

If we have a concentration on research to diagnose individuals with genetic diseases and fixate on that ideal, we could have potentially unintended consequences. Our research into treatments and therapies may be placed on the back-burner, which ultimately places society on the side that we want to know who is sick, but it is not our priority to treat them. The individuals with genetic disorders or the potential to have offspring with disorders would essentially be devalued, and socially coerced to follow these ungrounded assumptions made by eugenics. This is much like the coercion being employed to encourage vaccination. But the analogy is false because in nature, infectious disease and genetic disease are not the same. One is horizontally transmitted, meaning it spreads from person to person no matter the degree of relatedness, while genetic disorders are purely vertically meaning that it stays within a family. Ultimately, genetic testing should not be forced and used to dictate what an individual has to do, but it simply provides information and an opportunity to make a decision for yourself and take timing and preparation into account. So I refuse to accept that the two public health concerns are analogous in anyway.

Another theme that I hope is taken away from my discussion is that the ambition towards perfection and controlled life, ultimately defeats our humanity. As seen from my discussion of treatment versus enhancement, whether or not parents are using genetic/medicinal means to improve their children, love may be stated rationale, but underneath is often an aura of control. This stems from our idea that everyone is in competition with one and another and that there is

an ideal to strive for. This is not only a direct attack on the autonomy of children, but also the diversity of society. Diversity is the ultimate source of innovation and discovery, and no child should measure success with that of another person, rather children should measure whether or not their own goals have been reached. Another aspect of our humanity at risk is our goal to promote the flourishing of the sick or injured. Our focus has become dramatically skewed towards finding the flaws, rather than addressing or fixing them. Lastly, as seen in *Brave New World* we lose something when we forfeit our freedom and choice, in order to fulfill a fallacy of stability, control, and painlessness in our lives. Our ability to face adversity is something I see as humanities greatest asset. Without adversity we would find ourselves in a perpetual state of complacency. In sum, I believe that neither the end of controlling human nature nor the means of sacrificing our humanity are justified.

Some critics may propose that my rationale is rooted too much in Kantian ethics, while our society would gain more benefit from a utilitarian perspective. Utilitarianism is a philosophical ideology that places the priority on maximizing happiness for greatest amount of people. Eugenics as well as genetic enhancement could be justified by utilitarianism because it theoretically maximizes health as a good, leading to happiness and well-being for a majority of people. Although I do concede that utilitarianism is useful in other discussions, I believe that it does not fit the scope of this one. First of all, genetics is not a good that can be quantified or objectively put on a spectrum of good to bad and the idea of setting a hierarchy cannot be achieved without subjective favoring of certain traits over others. Secondly, the ability to determine traits that are good versus bad is impossible both in the scope of science and philosophy. For example, Down's Syndrome research has led us to breakthroughs in cancer research along with the fact that working with Down's Syndrome patients teaches compassion,

patience, and empathy. Lastly, our use of people as an instrument towards perfection and control, will not lead to more happiness, rather a loss of humanity. That is why I firmly believe Kantian ethics should direct us in this discussion because it mandates that people do not treat themselves as objects to reach a desired end. With those ideals guiding us we can foster both happiness and health by maintaining our humanity and striving to treat the people with the diseases rather than the diseases themselves.

I hope that I have painted the picture so far that the use of genetics, although constrained by the progress of technology and science, is ultimately dictated by society and culture. There is an idea in the study of sociology called the Thomas Theorem that states, “If men define situations as real, they are real in their consequences” (Smith 1995). The majority of this thesis has been an exploration of socially constructed ideas of stigma and prejudice. This is because objectively there is no scale that we can reference to value one type of human over another. In the end, if there are ideologies that exist that hold such biases, it is man-made. Since both of these phenomena are subjective in nature, we must not make the mistake in believing that the world we live in is completely objective. Without realizing this, we will continue to foster the subjective reality that harbors the valuation of some people over others; this will lead to very real consequences. This is where education and raising awareness comes into play. If we properly educate all the interested parties, including the public, federal and state level governments and health-care professionals; we may ensure that the subjective reality that is currently in its infancy will never see the light of day.

Through this work or by other means, I hope a discussion will be sparked that in the end will lead to transparency and collaboration between clinics, government, and insurance companies. Clinics must provide care without being directive and be able to provide equal care.

Insurance companies must ensure that patients are able to obtain genetic treatments because it is preventative. Clinics must also ensure that genetic counseling and testing is necessary as to not waste insurance money. Governing powers must put laws in place to protect the people but not direct how people should view genetic information. Laws should protect individuals rights to health and life and disability insurance. As well as ensure that genetic material is far removed from the market system to ensure income inequality distribution is avoided. Laws should be put in place that all gene therapy must be medicinal in nature as to be for treatment and never for enhancement. Laws also must protect genetic information from institutions of education and work to avoid discrimination as knowledge of personal genetics becomes more common.

Within the field of healthcare, among all the positions, one of the core values and goals is to provide evidence based care. I believe as the evidence of genetic causation and linkage to many diseases or ailments are found, it is the responsibility of all personnel within the healthcare system to stay up to date in order to provide the best care possible. That being said, I think discretion must be shown by the administrative powers governing medicine and research to determine what evidence is relevant in the present, and what research should not be implemented until further research. For example, the discovery of genes linked to cancer(s) has immediate implications in health care to help diagnose and prevent cancer, which as of now is the best treatment. On the other hand, a test that tests for an incurable disease with lack of knowledge on at least controlling or lessening symptoms, would for the time being, not be beneficial to incorporate into diagnostic healthcare. I believe this discretion can best be executed with proper education of ethics within the field of medicine and research. On top of this, making sure that the field of healthcare research and practice is guided by a *telos*, an Aristotelian term meaning having a definitive purpose, end, or nature (Sandel 2007). Having a *telos* for any practice firmly

orients and constrains it. I believe medicine's *telos* lies within the oath that all medical doctors must recite upon matriculating from medical school, the Hippocratic Oath. Within that oath they say, "I will prescribe regimens for the good of my patients according to my ability and my judgment and never do harm to anyone" (North 2002). A lot can be inferred from that, but I think in this case the explicit meanings speak for themselves. I believe doctors, researchers, and all healthcare professionals should strive to never do harm to a patient, whether that be physical or psychological, and always strive to promote human flourishing by never losing sight of treating a people with a disease, rather than achieving control over the disease itself. If those values are kept, I believe there should be no argument in the reason why genetics should not be seen as an end to be improved, such as eugenics, rather it should be seen as a means in order to promote humanity.

I know what many of you are asking yourselves, after writing this thesis am I still considering being tested for Lynch Syndrome? My answer is still, yes. This is because the genetic use of the test is well defined as purely for medical use and more like a treatment rather than an attempt to have dominion over my life. As of now, the best treatment for cancer is early diagnosis or prevention. Cancer in the general population has an age-dependent risk factor, but in cases like LS, the risk is greater and at a younger age. That is why the discovery of certain hereditary abnormalities that lead to increased risk are important because they allow people like me to know that I need to get screened earlier and possibly change my lifestyle or undergo surgery. That being said, this situation is not and should not be taken as analogous to most other genetic disorders, especially those that are dominant and shown at or near birth. Yes, we can detect the presence of those disorders in families or prenatally, just like with cancer syndromes, but the difference lies in the fact that the options for "treatment" is abortion. Staying clear of arguments

of pro-life vs choice here, abortion still lies far outside the realm of treatment. It is an important step that we have discovered the genetic source of many diseases and can detect them, but we cannot stop there. We must keep our priorities straight and strive to find cures for affected people, rather than use reproductive control or to judge parents that choose to have children with genetic disorders. Ultimately, nobody is perfect, and people with genetic disorders are just as human as everyone else. All people have risks in life, even if they are genetically “healthy,” such as getting in a car accident, getting injured, have heart disease caused by obesity, having cancer, etc. No one is free of risk or pain, and I do not think it is what our ambition should be because ultimately that is a corruptible ambition fueled by the desire to master and have dominion over our very human nature. Rather we should be concentrated on the flourishing of our fellow man because nobody should be seen as having less value than anyone else.

Plato, in his most famous work, *The Republic*, introduced the Theory of Specialization, which means, society is at its peak when the individuals that comprise it do what they are most fit to do by their nature (Plato and Bloom 1991). I interpreted this as all humans having a purpose no matter their disposition.

From my personal experience, the sickness and people who are sick provide much more than promise in research medical treatments. I volunteer at a camp for children with cancer and their siblings called Camp Wapiyapi. The non-profit, Camp Wapiyapi, was started in 1998 to provide a summer camp experience to children between the ages of 6 and 16 who either have cancer or are a sibling of a patient. The word “Wapiyapi” comes from the Lakota Sioux language and means “healing.” The camp could not have a more fitting name because it provides the kind of healing that happens outside the hospital received from relaxation, support, kinship, and love.

Often the campers are not the only ones that leave healed in some sense. I know that as a counselor after volunteering at camp I left with more appreciation for life and a clearer perspective on sickness. The children at Camp Wapiyapi have the capability to teach us so much about what it is to live in kinship with human-kind. One of the most important things they have taught me is to question the idea as what it is to be “normal” and that life is something to be appreciated because we have no reason not to even in the worst of times. Pediatric cancer, although all the linkages have not been found, has evidence pointing towards causation by genetic abnormalities present at birth. Even though the links between pediatric cancer and genetics is complex and not understood, I do not see that as being the case as research continues. In the case that a prenatal test is released the issue will become much more complicated than it already is. Throughout this paper I have used Down’s Syndrome as my primary example of a genetic disease discriminated against, but for the most part this disease is not painful. In the case of pediatric cancer I will not blame parents if they abort their children diagnosed with it, preferably if it is through beneficence rather than financial reasons. That being said I do not think our priorities should be to only find a diagnostic test for pediatric cancer and neither do I think our ambitions are in the right place if we no longer want these children to be born at all. Of course the first thing that needs to be discovered is the genetic link(s) to pediatric cancer, but the research effort cannot stop there. Those discoveries should be utilized as the first step in finding treatments for these children. Again, I may be repeating myself, but I must make it clear that our ambition should be to treat the people with the disease, rather than eradicate the disease no matter the cost. I can speak on the behalf of all the volunteers from Camp Wapiyapi and myself and say that the time we have spent with these children has been invaluable. On top of that, my life now has direction; I want to be a pediatric oncologist. As a pediatric oncologist I would be

able to translate what I have learned from writing this essay into my practices and be able to foster these children back to health as best as I can; giving them an opportunity to spread their zeal for life, compassion, strength, hope, and love with the world. Most importantly, these children have taught me to have an appreciation for life and for everyday given to us, because life is fleeting for all of us so there really is no reason not to live life to the fullest. This thesis is not only in defense of our society as whole, but is also my unrelenting effort to be an advocate for the lives of these children.

To conclude, I know I mentioned Star TrekTM in my introduction and now I feel responsible to tie that loose end. In the television series and movies, the institution of Starfleet requires all of its captains to recite an oath that basically says, that the goal of their journeys through space is to be purely exploratory, while avoiding disturbing other civilizations and engaging in military action. In the series this often does not go to plan and things go wrong and they must figure there way out of it. Obviously, this makes for an exciting story, but what I am talking about is real life. We cannot afford to be constantly fighting an uphill battle by trying to fix what could have possibly been avoided. Therefore our society as a whole, especially our governing powers, should take an oath of genetic exploration. Genetics must only be used for medical purposes and genetic information must be solely for the benefit of the person and not for profit. Genetics must always be clear of coercion, discrimination, or other social pressures. Lastly, the goal of genetics in medicine should be used for preventive care in appropriate cases, but should not deter from the ultimate goal of finding treatment.

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