ABSTRACT

Examining the Ethics of Prenatal Genetic Testing

Elizabeth M. McElroy

Director: Matt Bradshaw, Ph.D.

With new methods emerging in the field of prenatal genetic testing, it is essential to evaluate the ethical significance and consequence to these discoveries. This thesis explores such topics as: the extent to which genetic screening is utilized, what constitutes “disease” in cultural context, the public opinion of widespread genetic testing, and the ethically-ambiguous nature of newborn organ donation, amongst others. For each of these ethical questions, the issue will be evaluated in its current status, as well as the historical and predicted future utilization. Whenever possible, public opinion polls are integrated to gauge how aware the public is to these new methods, and how our governments could best protect patients and our broader society in this new era of genetic testing capabilities.

APPROVED BY DIRECTOR OF HONORS THESIS:

_____

Dr. Matt Bradshaw, Department of Sociology

APPROVED BY THE HONORS PROGRAM:

_____

Dr. Elizabeth Corey, Director

DATE: _____

EXAMINING THE ETHICS OF PRENATAL GENETIC TESTING

A Thesis Submitted to the Faculty of

Baylor University

In Partial Fulfillment of the Requirements for the

Honors Program

By

Elizabeth M. McElroy

Waco, Texas

May 2019

TABLE OF CONTENTS

Fear not, for I am with you;

Be not dismayed, for I am your God;

I will strengthen you, I will help you,

I will uphold you with my righteous right hand.

Isaiah 41:10

CHAPTER ONE

Introduction

With the recent advancement in genetic testing, specifically for prenatal care, arises a new set of ethical questions as to their clinical utility. Under which circumstances should these testing methods be used? What are the societal impacts of a whole generation of children impacted by these new genetic tests? This thesis aims to examine these issues and more by focusing on: what constitutes genetic disease from a societal perspective, determining if healthcare interventions are a right or a privilege, considering the ethically ambiguous nature of newborn organ donation, and examining the evolving public view on genetic testing and its moral correctness.

This thesis will utilize research articles from the National Institutes of Health, ethical and religious directives from several denominations of the Christian faith, and several resources that expecting families have at their disposal to gauge the perspective of the major parties involved in these ethical decisions. This literature review approach intends to demonstrate the scientific discovery that led us to this clinical genetic testing protocol, the ethical and religious foundation for those deciding the morality of this intervention, and the basis of knowledge prospective families enter this discussion with. By bringing each of these perspectives together under the outline listed above, this thesis aims to unpack the ethical considerations of prenatal genetic testing in a manner that allows experts and the layperson to understand this issue from the perspective of others. Hopefully this, in turn, will

allow us as a society to stand united in our goals for the future utilization of these and other genetic testing advances, especially in regards to the complex realm of beginning-of-life medical ethics.

For the purposes of this thesis, prenatal genetic testing refers to testing procedures performed on either a pregnant woman or her fetus, with the intention of determining if the child has certain genetic markers for disease, including an abnormal chromosome count. These procedures have greatly improved in recent years, and can be utilized by physicians to determine more about the problems a child might face in development. Specifically, this thesis examines Non-invasive Prenatal Testing procedures, known collectively as NIPT. As these tests are gaining in accuracy, and have developed to provide reduced risk to the fetus, the increased use of these tests in regular prenatal care necessitates a real conversation as to what to do with these results. While some conditions can be treated with neonatal surgical procedures or an advanced birthing plan to decrease infant and maternal mortality during childbirth, others have no contemporary treatment plan or cure. In these situations, is knowledge of the condition really beneficial, and what should parents do with the knowledge that their child's life will be short and/or painful?

This thesis aims to explore these issues, somewhat removed from the emotional distress of a parent's decision-making, and towards a larger view of how we should approach this issue as a society. The foundations of medical ethics include beneficence, nonmaleficence, autonomy, and justice - these four pillars must be upheld for any medical decision to be sound. This thesis will attempt to

uncover how this issue impacts our larger economy and healthcare framework, and how this delicate situation can be tactfully approached with distraught parents. The aim of this thesis is to gain a richer and more comprehensive view on the ethical issue of prenatal genetic testing, with grounding in both academic discipline and Christian doctrine.

CHAPTER TWO

What Constitutes a Genetic Disease?

One of the first questions to be debated in this thesis is what constitutes a genetic disease. Some groups find a genetic abnormality to be an asset, notably those impacted by dwarfism or members of the deaf community. These groups are generally proud of their unique culture and community, and tend to be hesitant to view their situation as a disability. Iceland broke headlines recently by having the lowest rate of Down's Syndrome births in the world - just one or two per year of their 330,000 population. This statistic largely resulted from the common practice of terminating pregnancies with abnormal genetic markers. Iceland allows abortion past the 16-week mark if the child is found to have a genetic condition, such as Trisomy-21, that is expected to diminish the child's quality of life. In the United States, where the rate of Down's Syndrome births are closer to 6,000 births per year with its 325 million population, Iceland's practice is largely seen as unethical. Down's Syndrome results from the child receiving three copies of chromosome 21; it is one of the only trisomy genetic conditions that are not fatal.

While patients with Trisomy-21 have a characteristic facial structure that makes them recognizable, they can have differing abilities and disabilities. Many adults with Down's Syndrome can have full and happy lives, though perhaps not as independently as they might without the trisomy. Parents in the United States tend to form support groups with one another, and the government can help in funding the immense medical bills this child will inevitably require - from additional

help in schooling to speech therapy, occupational therapy, and physical therapy. Well-adjusted adults with Down's Syndrome can gain employment, though they are often limited to lower-paying occupations in the service industry. The financial impact of having a child with Down's Syndrome to the total economy is almost certainly a net expense to taxpayers, and these children require a great deal of additional resources from healthcare and educational institutions.

But should a child's life be determined in this black-and-white fashion? There is no procedure or medication to "fix" a child with Down's Syndrome; the extra chromosome is a part of their inherent framework. This child could not exist in any way other than with an extra copy of chromosome 21, and there is a possibility for some affected children to go on to live a happy life with a loving relationship with their family and friends, even with this abnormality. Is Iceland ethical in their widespread practice of terminating these births, in order to save resources for other children and divert funds to other areas of the economy, or are they cruel in deciding that some lives should not be lived? One genetic counselor from Iceland shares her viewpoint, saying:

> We don't look at abortion as a murder. We look at it as a thing that we ended. We ended a possible life that may have had a huge compilation... preventing suffering for the child and for the family. And I think that is more right than seeing it as a murder -- that's so black and white. Life isn't black and white. Life is grey." (Helga Sol Olafsdottir)

This is quite different from the typical opinion shared by clinicians in the United States, but it sheds a unique light on the issue that may be worth considering. As our society increasingly becomes globalized, we must confront these foundational differences in culture when creating a framework for genetic testing moving

forward. This technology can do incredible things to save lives, but with cultural difference, it may be difficult to decide how and when to use these tests, and what should or should not result from discoveries.

CHAPTER THREE

Are Healthcare Interventions a Right or a Privilege?

Whenever dealing with children, especially newborns and fetuses, the question emerges - is healthcare a right or a privilege? Infants certainly have no ability to pay for their own healthcare, and many interventions such as surgery are very expensive. It is not unheard of for children to accumulate tens of thousands of dollars in medical bills just over a few weeks in the Neonatal Intensive Care Unit (NICU)[1]. We, as a society, are at a turning point in our view of healthcare delivery. As proponents of Medicare-for-All are pointing to successful models in Europe and other nations, it is worthwhile to apply this question of to what degree healthcare is a fundamental human right applies to the topic at hand. Most everyone would agree that infants and the unborn are not responsible for their medical conditions, that no lifestyle choices led them to a life-threatening condition. By asking the larger question of should healthcare be seen as a universal right to all people or a privilege to a select few, we can see this issue from a broader perspective.

Throughout documented human history we see a desire within society to help others. Because of that focus, we also see the goal of acquiring knowledge to provide more effective assistance. The profession of healer or shaman is an intrinsic part of the human experience; we have a need to cure ailments of the

[1] How Plans Can Improve Outcomes And Cut Costs for Preterm Infant Care. (2017, December 15). Retrieved March 12, 2019, from https://www.managedcaremag.com/archives/2010/1/how-plans-can-improve-outcomes-and-cut-costs-preterm-infant-care

body, mind, and soul. Midwives help deliver babies and protect maternal health – a necessary role for the survival of the species. Warriors and hunters need to be strong to provide food for others, so they need to be healed when wounded or ill. Even dismissing the virtue of the ideal of healing others, there is an objective necessity to provide healthcare in order to live as part of a larger group. There are also many examples in our modern society that illustrate our fundamental desire to help each member of society and to treat healthcare as a basic right. When in an emergency, first responders assist, regardless of your socioeconomic status or ability to pay. Few people would insist on the presentation of a health insurance card before firefighters use the jaws of life to pull you from a burning car and transport you in an ambulance to the nearest emergency room. It is not practical to take the time and possibly risk a life to ensure proper paperwork is in order.

We also tend to agree that children deserve basic healthcare, even if their parents are unable to pay. Children from all socioeconomic backgrounds are required to receive basic childhood vaccinations before beginning in public elementary schools. This law implies that we, as a society, believe that all children should benefit from the scientific research of how to prevent disease. Most of us concur that no child in the United States should suffer from polio, measles, mumps, or a variety of other preventable childhood diseases; all children should benefit from scientific discovery. Jonas Salk refused to patent the polio vaccine because he wanted it to be accessible to all people; he believed every child should benefit from his work and he hoped to dispel this crippling childhood disease[2]. These

[2] "About Jonas Salk." *Salk.edu*, Salk Institute for Biological Studies, 2019.

examples are clear indicators that our society in the United States, at least in some situations, sees healthcare as a fundamental right. It can be argued that at least in terms of children, our public welfare systems highly prioritize access to healthcare interventions and basic primary care.

There is a substantiated argument that healthcare may not be a right in every situation – many elective procedures are argued to be a privilege that should only be available if the individual is willing to sacrifice resources in that endeavor. Primarily cosmetic procedures, such as elective plastic surgery, should not be paid for by the taxpayer's dime. But these clear-cut examples of a child receiving a polio vaccine and a wealthy middle-aged woman seeking breast enhancement are on either extreme. The real burden is deciding what to do for cases in the middle of these examples, such as patients who previously led an unhealthy lifestyle requiring expensive surgeries because of their past choices. Several ethical issues arise, for example: should an individual with advanced-stage liver disease receive an organ donation only if they were never an alcoholic? Are there some people who do not deserve healthcare, say inmates of corrections facilities? These specific areas are ethically ambiguous, but do not detract from the fundamental view that healthcare is a right. Moreover, the fact that these are seen as ethically confounding indicates an underlying supposition that healthcare is a right and that only extreme examples require special consideration.

On a global scale, we treat healthcare as a fundamental right in most cases. Medical missionaries dedicate their lives towards helping those in developing countries receive the services we see as basic primary care in the United States.

Even outside of healthcare interventions, tax dollars from the United States go towards other relief organizations, providing food and shelter after natural disasters and during times of war. In this manner, we are prioritizing their health by providing these other interventions to promote basic well-being.

The United Nations High Commissioner for Human Rights states that the right to health is a fundamental human right and must include four interrelated elements: availability, accessibility, acceptability, and quality. That is, healthcare services and "underlying determinants of health" such as sanitation and clean drinking water, should be provided to all in a manner that is geographically accessible, without discrimination, and within the appropriate confines of cultural norms and medical ethics. Healthcare services should also be of "good quality" and be scientifically appropriate. While much of this report was created for use in very impoverished nations or for use in UN-sanctioned refugee camps, this document also provides an excellent framework for a philosophical argument. The Office of the United Nations High Commissioner for Human Rights (OHCHR) describes its mission: "to work for the protection of all human rights for all people; to help empower people to realize their rights; and to assist those responsible for upholding such rights in ensuring that they are implemented" (OHCHR and UNAIDS, 2007).

Stanford's Encyclopedia of Philosophy sees public health as a "commitment to secure a sufficient level of health for all and to narrow unjust inequalities."[3] In

[3] Faden, Ruth and Shebaya, Sirine, "Public Health Ethics", *The Stanford Encyclopedia of Philosophy* (Winter 2016 Edition), Edward N. Zalta (ed).

addition to the virtue of healing others, underlies a devotion to social justice and deterrent to disadvantage. In this manner, one can view the concept of justice directly in light of social determinants of health. John Rawls' definition of justice includes a discussion of the fair distribution of social goods, which can be logically expanded to healthcare as a fundamental element of justice in a society that has the economic ability to provide basic services to its populace. The overarching argument is that "health is an essential pre-requisite for the realization of the fundamental rights and freedoms"[4] that is necessary to pursue any other sort of social good in a just society. Individuals must possess a reasonable level of good health and intelligence in order to fully participate in society; this is the basis of viewing basic education and healthcare as fundamental rights. This too, can be mirrored in our current society - public education is largely free to schoolchildren through the twelfth grade; if they wish to continue their education they can do so at their own expense. We, as a society, value education and its role in creating a more equitable society and a skilled workforce. Expanded further, a democratic society requires that voters have a certain level of education to fully participate in the democratic process - this too can be undermined if citizens are not of adequate health. Ideally, society should provide sufficient healthcare to its citizens so that they can participate in commerce, the democratic process, and some degree of socialization with others.

[4] Ekmekci, Perihan Elif and Berna Arda. "Enhancing John Rawls's Theory of Justice to Cover Health and Social Determinants of Health" *Acta bioethica* vol. 21,2 (2015): 227-236.

Basic healthcare must be seen as a fundamental human right in order to justify our current beliefs in justice and democracy. Individuals need the ability to provide for themselves and contribute to a larger society; this is impossible if the individual is not of adequate health, and a disservice to the larger community if this individual is not taken care of. Whether seen as humanitarian relief, a kindness, or a necessity for a well-functioning society, it is clear that healthcare should hold a high priority in any society which has the funding and the expertise to provide these services.

There are procedures and instances wherein it is not economically stable for the larger society to cover the expense, that the cost greatly outweighs the benefit, but these cases represent a small fraction of a larger model for healthcare delivery, especially in developing countries. It is the collective responsibility of any society to provide adequate healthcare to its members, to support each individual so they can contribute meaningfully to others, and to enjoy a greater quality of life. Therefore, it is essential that the basic rights of these fetuses and newborn children are protected by providing basic foundational care. Obviously, due to disparities in the resources of the healthcare center, the same care may not be able to be provided in developing countries that in a state-of-the-art surgical centers in wealthy nations. Should the burden fall entirely onto the parents to provide the funding for this foundational care, which may include expensive surgeries, or should society step in to allow that young baby to have a chance at survival? This is a moral question that is increasingly asked, especially as religious groups and others insist that abortion and termination of the pregnancy not be an option.

If termination is not an option for moral reasons, should it not also be a moral requirement of society to provide medical care for these children? If we continue to view the life of a child to be precious before being born, we must also assume the financial responsibility of caring for them after birth. Society must assist parents with the financial liability of high medical expenses for this child, particularly when the family might be of low socioeconomic status. Money should not be the determinant of a child's survival. This is an issue that our society has not been able to answer in a functional and comprehensive manner.

CHAPTER FOUR

Newborn Organ Donation

One specific disease to note is that of anencephaly, the incomplete formation of the brain and spinal cord while in gestation. Depending on the severity of this condition, children can either live with significant disability or never leave the hospital. One practice that is becoming increasingly utilized is electing for these most severely affected children to serve as organ donors. In most cases, the child typically suffers from no other structural abnormalities other than the neurological malformation that prevents independent life. Because their other organs are formed healthily, some parents choose for their child to become an organ donor, to donate their internal organs to children born without functional use.

Many parents see this as a unique opportunity for their unborn child to contribute meaningfully to the world. One common sentiment from parents is that the news that their child was unable to live was the worst news in their life, then if their child can spare another family from that tragedy, what a meaningful sacrifice. For many children with a complicated organ defect, organ donation can be the only option of survival. Newborns and premature infants can only accept organs from other infants that are incredibly young, so one child that serves as an organ donor can save multiple lives, donating its heart, its lungs, each kidney, or each lobe of its liver. Many adults cannot say that they have saved as many lives.

The American Medical Association (AMA) specifically stated in 1995 that they do not approve of the ethical circumstances surrounding organ donation from an encephalitic infant to provide their organs to other newborn children[5]. This, however, has not stopped the procedure from increasingly gaining in common practice, especially with parents who know earlier in the pregnancy that this is an outcome that is expected for their child. Parents who have gone through this experience often see newborn organ donation as a way to have the child's life be meaningful, even though it was of a short duration. They share the sentiment of fulfillment that part of their child saved the lives of others. Parents can also choose to wait for a natural death and then donate the child's body to a research foundation to prevent conditions such as this from happening to future children. This is often seen as less controversial as it does allow for natural death; the problem with this, however, is that this limits the organs' availability to be used for organ donation because the tissues are not fully oxygenated. in many cases, children born with a severely malformed heart can only survive if they receive a donated heart from another newborn, as any other-aged heart would be too large to fit in their thoracic cavity. This necessitates some system of newborn organ recovery to provide these infants with one non-functional organ to survive. Newborn organ donation has an extra layer of ambiguity in terms of its ethical nature, and there are few explicit protocols demonstrating under which circumstances this practice is appropriate. Decision-making is more difficult due to its emotionally-charged nature and the limited time frame in which parents can make this life-altering call.

[5] Anencephalic Newborns as Organ Donors. (n.d.). Retrieved April 3, 2019, from https://www.ama-assn.org/delivering-care/ethics/anencephalic-newborns-organ-donors

A framework to guide parents and their physicians through this ethically-ambiguous and emotionally-charged period needs to be established, and the AMA needs to help in this process. Parents need to be counseled early, as in many cases, parents must make the decision before delivery, as the most successful organ recovery is done during a partial Cesarean section. This procedure allows the mother's umbilical cord to serve as a sort of life-support machine providing oxygen to the child during recovery. This is the element that makes the procedure extremely controversial and is seen as unethical, as the AMA resists interventions on newborns that occur before natural death. Ultimately, protocol must be put in place to ensure physicians and parents make sound minded decisions on this matter, one that gains consensus from ethicists and scientists alike.

CHAPTER SIX

Public View on Genetic Testing

It is important to recognize that the public remains apprehensive about genetic testing, even in the case of autonomous adults, so this moral question is exacerbated for children, especially the unborn. A VU University Medical Center Amsterdam study published in the European Journal of Human Genetics shows the change in the public's view of genetic testing from 2002 to 2010.[6] They chose these years because in 2002, the Human Genome Project had a large voice in the news that promised a new future for genetics. In 2010 however, the public was left a bit disenfranchised with the progress science had made, and more ethical issues continued to arise. Below is a table that illustrates the data they collected from surveying individuals in both 2002 and 2010 to better understand the public's view on genetic testing, and the situations wherein it is acceptable and unacceptable. The greatest marker for a society's collective ethical conscience is the general opinion, and for this thesis is the most compelling evidence of where we, as a society, have historically and currently stand on this issue.

[6] Henneman, L., Vermeulen, E., van El, C. G., Claassen, L., Timmermans, D. R., & Cornel, M. C. (2012). Public attitudes towards genetic testing revisited: comparing opinions between 2002 and 2010. *European journal of human genetics : EJHG, 21*(8), 793-9.

Table 3

Beliefs about genetic testing (2002 *vs* 2010)

	2002[a] **(n=964) *Mean (SD)***	***2010* (n=811) *Mean (SD)***	***F (df)***	**P-*value*[b]**
To prevent disease I would want to know my risk of getting certain diseases	3.42 (1.17)	3.46 (1.25)	0.67 (1, 1718)	0.413
(Completely) disagree, %	21	25		
Not agree/not disagree, %	27	22		
(Completely) agree, %	52	53		
I am curious about my genetic make-up	2.95 (1.26)	3.17 (1.26)	11.47 (1, 1718)	0.001
(Completely) disagree, %	36	31		
Not agree/not disagree, %	30	26		
(Completely) agree, %	34	44		
I do not want to know what kind of diseases I could get in the future	3.61 (1.25)	3.23 (1.33)	32.94 (1, 1715)	<0.001
(Completely) disagree, %	20	30		
Not agree/not disagree, %	23	23		
(Completely) agree, %	57	45		
Genetic tests deprive people's freedom to live as they want	3.08 (1.26)	3.02 (1.20)	0.10 (1, 1716)	0.747
(Completely) disagree, %	31	33		
Not agree/not disagree, %	33	31		
(Completely) agree, %	36	36		

All answering scales to statements 1=completely disagree to 5=completely agree.

[a]See reference Henneman *et al.*[10]

[b]Significant differences between 2002 and 2010 (analysis of covariance with educational level and age as covariates).

Table 3

Beliefs about genetic testing (2002 *vs* 2010)

	2002[a] (n=964) Mean (SD)	2010 (n=811) Mean (SD)	F (df)	P-value[b]
Knowledge about the genetic background of disease will help people to live longer	3.32 (1.09)	3.70 (1.03)	39.91 (1, 1714)	<0.001
(Completely) disagree, %	20	12		
Not agree/not disagree, %	37	24		
(Completely) agree, %	43	64		
Genetic testing does more good than harm	3.01 (1.03)	2.90 (1.11)	3.01 (1, 1704)	0.083
(Completely) disagree, %	26	33		
Not agree/not disagree, %	46	38		
(Completely) agree, %	28	29		
Genetic testing is tampering with nature	2.58 (1.22)	2.24 (1.15)	13.62 (1, 1714)	<0.001
(Completely) disagree, %	50	60		
Not agree/not disagree, %	29	28		
(Completely) agree, %	21	13		

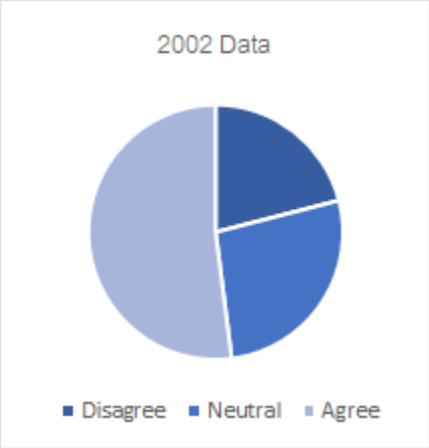
All answering scales to statements 1=completely disagree to 5=completely agree.

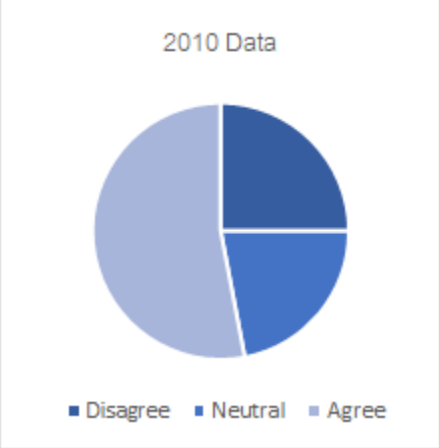
[a]See reference Henneman *et al.*[10]

[b]Significant differences between 2002 and 2010 (analysis of covariance with educational level and age as covariates).

RESPONSES TO NOTE:

Q1: *To prevent disease I would want to know my risk of getting certain diseases.*



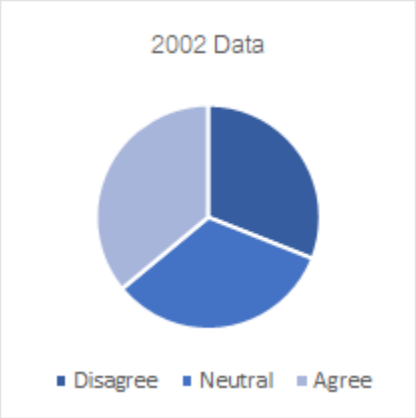


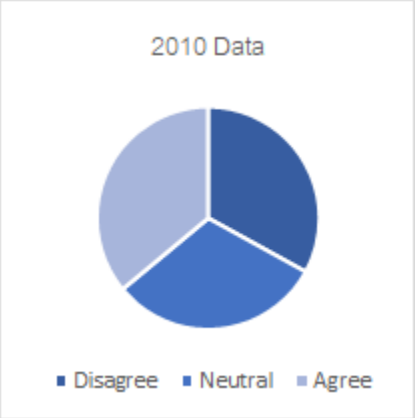
21% Disagree
27% Neutral
52% Agree

25% Disagree
22% Neutral
53% Agree

The majority of respondents agree that they would want to know their risk of inheriting disease in order to prevent it. This was consistent across both survey periods. This result indicates that the majority of individuals would like to take control of their future healthcare outcomes by empowering themselves with knowledge regarding their genetic predisposition. This finding confirms that the majority of the members in our society deem it ethical to have testing done to know more about their genetic predisposition to future disease.

Q4: *Genetic tests deprive people's freedom to live as they want.*



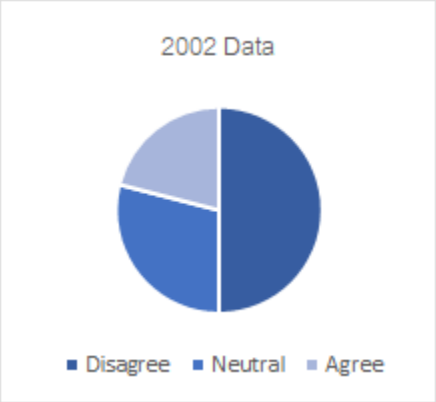


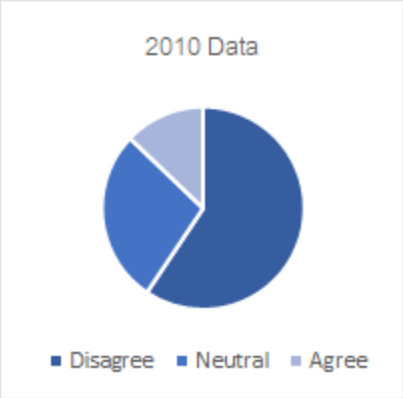
31% Disagree
33% Neutral
36% Agree

33% Disagree
31% Neutral
36% Agree

This response set adds controversy to the subject - it is, in fact, an even split between survey respondents as to their stance on genetic tests depriving individual freedom, a view that did not change after eight years. This may be of grave concern to the future of genetic testing, as individuals are still wary that the documentation of their genetic information may lead to a deprivation of their freedom. There is a fear that companies may exploit this information, by possibly denying health insurance or an employment opportunity to those predisposed to health issues. If genetic testing is to advance for the general public, advocates must be involved in policy discussions to promote the general liberties of the populace. This broad testing can help individualized medicine gain prevalence in healthcare.

Q7: *Genetic testing is tampering with nature.*





50% Disagree
29% Neutral
21% Agree

60% Disagree
28% Neutral
13% Agree

The majority of respondents in 2002 disagreed with statement seven, stating that they believed that genetic testing is not tampering with nature. This opinion was strengthened in 2010, where 60% of respondents found genetic testing not to be tampering with nature, with only 13% of respondents viewing genetic testing as tampering with nature. This is a strong indicator that the public is open to genetic testing and sees it, if not entirely helpful, also not harmful. Genetic manipulation, such as clinical gene editing or genetically modified food, might be seen differently, but this response is encouraging for proponents of widespread genetic screening for diseases with clinical validity, wherein treatment and/or prevention protocols may be in place.

There are many ethical considerations when examining genetic testing. One popular test that has emerged recently is this idea of ancestry-type genetic testing services like “23andMe”, which can help individuals understand where they come from, and in certain cases, what types of diseases they might have a predisposition towards. This is due to an analysis of their genetic similarities to individuals in their database that have hereditary-linked diseases such as cancer, Alzheimer’s, Parkinson’s, and Huntington’s disease. They generate this data from pre-established research regarding cancer-linked genes, such as BRCA1 and BRCA2 linked to breast and ovarian cancer, and informing clients of this heightened risk. For many types of cancer, there are strong links between possessing this gene and developing cancer, especially if preventative measures are not taken. Ideally, clients informed of their heightened risk are prompted to take control of their genetic destiny and better prepare themselves for a long and healthy life.

It is important to note that the sensitive information garnered from genetic test results can influence not only the individual, but their blood relatives and potential mates or offspring as well. What one individual learns about their own genetic traits might cause unwanted impacts on other individuals and family members that shared their genetic predisposition. For instance, one may not want to know if they are predisposed to have an incurable genetic condition, like Huntington’s disease, but if a sibling, parent, or child of the individual has the test, the individual learns unwanted information about their own future, as there is a very high chance of them also having this gene. It is worth considering whether or

not people have a right to test for certain conditions and disclose that information to their family members. Should it only be confined to consenting adults and not children, or should family members be left in the dark if the condition has no preventative measures? Should related individuals be forced to undergo genetic testing if a blood relative is found to have a preventable disease if regular screening and lifestyle changes can eliminate symptoms? These are ethical questions about adult genetic testing that we do not have concrete answers for, and this issue is exacerbated when speaking in terms of children.

For instance, if one child is diagnosed with ta genetic disease that may be onset later in life, should they be told about this disease? And, if so, at what age? Should other children in the family be tested? Are these answers different is the disease is easily prevented or has no cure? These questions are very much worth considering, and especially important to view in light of the results by the VU University Medical Center Amsterdam's study. Adult respondents tend to view the information gained from genetic testing as a positive addition to their healthcare. Can we extend this to children, or is that a different moral circumstance? And should any additional thought go into individuals with family members who are morally against genetic testing, a member of the 13% that, in 2010, responded that genetic testing is tampering with nature? Should these individuals be forced to reconcile their genetic predisposition because a family member obtained pertinent information without the reluctant individual's consent?

Some of these ethical questions can and should be asked for a variety of genetic testing scenarios, pertaining to prenatal care or not. For some diseases

there is no cure, or even a treatment protocol, that can greatly reduce the symptoms of the disease. In these cases, should people be informed or do they have the right to deny testing? What if other people in their family know that they will have this disease in the future and it can prepare them to make better plans? This also follows for people who are wanting to have a child - if they know that they are a carrier of a particular disease, they may have a lengthier discussion when deciding to procreate, especially if their spouse may also have a family history of the disease. Should a mother who wants to have a child with her husband be given the right to know that her husband is a carrier for a particular genetic disease? Are people who are intending to have children with one another be obligated to divulge any information they know about their own genetic disposition? Should sperm donors or other people that provide surrogate parenthood either with a sperm or an egg donation should they be required to undergo genetic testing to ensure that they are not carriers for genetic diseases, and if they do undergo this type of testing, should they be required to divulge that information to their supposed employers, who would then raise this child? There are all questions worth considering, and should be considered when deciding to undergo genetic testing.

CHAPTER SIX

Conclusion

With the emergence of new types of genetic testing comes the heightened need for our society to state clear guidelines as to how and when these services should be utilized. It is critical that we examine these issues within a wider context of their cultural significance, religious connotations, moral elements, and philosophical perspectives. This thesis was meant to unpack each of these ethical issues and analyze them from a broad perspective. Many times, clinicians view these issues in a very emotionally-charged context, with patients that they know personally. Hopefully, by seeing these issues from an objective perspective, it is more realistic to establish preconceived notions as to what is and is not ethical in the clinical application of prenatal genetic testing.

There is no clear right or wrong answer as to how these new testing procedures should be used, but hopefully this thesis gauges the issue in a way that is considered reasonable and realistic in its utilization within the healthcare setting. Many of these issues are quite controversial, both amongst healthcare professionals and the general public. Hopefully policy makers, healthcare providers, religious leaders, philosophers, and ethicists can find common ground as to how we should move forward as a society with these new health care interventions and testing capabilities. We must all come to an agreed policy for these issues if we are to provide a safer and healthier future for all citizens, while still maintaining the integrity of the patient, and recognizing that

there is ambiguity in these ethical decisions. There are circumstances in which these interventions should take place in light of prenatal genetic testing, but there are also situations in which interventions based on learned information is not in the best interests of the patient and their family, and is therefore unethical. The information garnered from this thesis can be applicable to clinicians, patients, genetic counselors, and a variety of others who are hoping to engage in this conversation in an educated and meaningful manner. This knowledge will allow us to better support one another as we face these difficult decisions, sometimes on a daily basis. The issues discussed in this thesis are highly personal as the diagnoses examined can be emotionally and financially devastating to families expecting a healthy child. Hopefully this academic perspective can help to make these decisions, which are inherently difficult, somewhat more manageable for clinicians to address. There are no doubt countless other ethical implications of these types of decisions, but hopefully this thesis begins to unpack these issues in a way that is helpful for everyone involved.

It is reasonable for there to be a serious discussion as to what is and is not classified as a genetic disease. As seen in chapter two of this thesis, even with a common disease relatively prevalent in the human population, there can be very diverse cultural belief systems about diseases like Down Syndrome, otherwise known as Trisomy-21. The view of Icelanders is quite different from those of United States citizens in terms of how people with this disease should be treated and how a diagnosis of Trisomy-21 early in the pregnancy can impact its outcome. There are no doubt other groups who might feel similarly about a disease not being seen

as associated with a happy and successful life. This decision should be made with all sides considered; physician should especially take note because the news of a particular diagnosis can mean very different things for different families, depending on their viewpoint. There are no doubt countless other diseases with groups of supporters from patients and their family members who hopes to shift this diagnosis into something that feels more livable.

The third chapter of this thesis examined an issue that seems constantly in the news, that is certainly not confined to prenatal care. The question of whether healthcare considered a right or privilege is one that many politicians, civil rights activists, and physicians find themselves asking constantly. Is it more just to provide care to everyone, or only those who can afford the care; and are there any inventions that should only be seen as a privilege, or is all medical care equally necessary for a happy and healthy individual as part of a larger society? While this thesis surrounds almost entirely prenatal care, this chapter provided an opportunity to look at the larger scope of healthcare and how we view it, both within the United States and with a larger global health context. Hopefully some insights discussed in this chapter can influence policymakers and philosophers in areas outside of prenatal care. Chapter four focused on the odd case of newborn organ donation. The American Medical Association strictly voted that this was not an appropriate procedure for physicians perform in the United States, yet there are many who deem it to be an acceptable practice, both morally and medically. Some parents find great peace after this procedure is done with their child, and find it to be an incredible sense of closure. Should parents be denied this closure because

of another individual's moral beliefs, or should they be allowed to deal with their child's passing in the way that they see fit?

Finally, this thesis examines how these types of genetic testing issues are viewed by the greater public, who typically do not have an advanced understanding of genetic testing procedures or clinical implications of these decisions. With new genetic testing possibilities, including commercial products such as 23andMe and others, it is important to understand how our public opinion is changing as these genetic testing platforms are gaining public attention. Ethical considerations should be given to other members of the family, and any others who may be impacted by the discoveries made by any of these testing companies. The manner in which discoveries are related to patients is not as heavily regulated by the federal or state government as other healthcare professionals who provide similar information. This is an oversight, as this information is more and more often involving medical information, especially when testing for genetic predispositions for particular diseases. The manner in which patients are educated on this information should not be left to a commercial company with little oversight to interpret the diagnosis, or the odds of gaining a particular disease without any formal medical training or adequate understanding of the patient's previous medical history. These tests should either come along with a clinician to sit down with the patient and clearly tell them how they should be done, or it should not be done at all. Ideally, these tests should be limited to pre-existing genetic clinics with experts who are already trained on how to approach these situations with grace and medical wisdom, rather than leaving consumers interpreting these results via

the internet without any larger scope besides sources like WebMD, which can sometimes lead to scary and unrealistic ideas of disease.

Hopefully this thesis provided an overview of the various ethical considerations that should go into the new fields of genetic testing, specifically for prenatal care, and investigated some of the ethical implications inherent to this increased usage in our society. These issues are largely thought about retrospectively, as these procedures are coming out in greater frequency and earlier in the stage of disease that ever previously utilized. This thesis aimed to provide commentary on current procedures and their ethical implications, and initiated a discussion on how to go about treating similar types of procedures and, in the future, hopefully setting up a framework so clinicians and patients can have peace of mind that they are using these methods appropriately.

BIBLIOGRAPHY

“About Jonas Salk.” *Salk.edu*, Salk Institute for Biological Studies, 2019.

Anencephalic Newborns as Organ Donors. (n.d.). Retrieved April 3, 2019, from https://www.ama-assn.org/delivering-care/ethics/anencephalic-newborns-organ-donors

Baptist General Convention of Texas, & The Christian Life Commission. (n.d.). Infertility & Medical Options; Getting Well: Christian Perspectives on Health, Sickness, and Ministry series. Retrieved April 3, 2019, from https://texasbaptists.org/ministries/clc/ethics-justice/life-health-dying/infertility-medical-options

Byrne, P. (2005). Use of anencephalic newborns as organ donors. *Paediatrics & Child Health, 10*(6), 335-337. doi:10.1093/pch/10.6.335

Ekmekci, Perihan Elif and Berna Arda. “Enhancing John Rawls's Theory of Justice to Cover Health and Social Determinants of Health” *Acta bioethica* vol. 21,2 (2015): 227-236.

Faden, Ruth and Shebaya, Sirine, "Public Health Ethics", *The Stanford Encyclopedia of Philosophy* (Winter 2016 Edition), Edward N. Zalta (ed).

Henneman, L., Vermeulen, E., van El, C. G., Claassen, L., Timmermans, D. R., & Cornel, M. C. (2012). Public attitudes towards genetic testing revisited: comparing opinions between 2002 and 2010. *European Journal of Human Genetics : EJHG*, *21*(8), 793–799. doi:10.1038/ejhg.2012.271

How Plans Can Improve Outcomes And Cut Costs for Preterm Infant Care. (2017, December 15). Retrieved March 12, 2019, from https://www.managedcaremag.com/archives/2010/1/how-plans-can-improve-outcomes-and-cut-costs-preterm-infant-care

Kagan, K. O., Sonek, J., Wagner, P., & Hoopmann, M. (2017, July 12). Principles of first trimester screening in the age of non-invasive prenatal diagnosis: Screening for chromosomal abnormalities. Retrieved from https://link.springer.com/article/10.1007/s00404-017-4459-9

OHCHR and UNAIDS (2007), *Handbook on HIV and Human Rights for National Human Rights Institutions*. Geneva: Office of the United Nations High Commissioner for Human Rights and the Joint United Nations Programme on HIV/AIDS.

Parham, L., Michie, M., & Allyse, M. (2017, February 11). Expanding Use of cfDNA Screening in Pregnancy: Current and Emerging Ethical, Legal, and Social Issues. Retrieved from https://link.springer.com/article/10.1007/s40142-017-0113-x

Sarnaik, A. A. (2015, November 17). Neonatal and Pediatric Organ Donation: Ethical Perspectives and Implications for Policy. Retrieved February 12, 2019, from https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4646954/

Silcock, C., Liao, L., Hill, M., & Chitty, L. S. (2014). Will the introduction of non-invasive prenatal testing for Down syndrome undermine informed choice? *Health Expectations*, 18(5), 1658-1671. doi:10.1111/hex.12159

Trisomy 13 - Genetics Home Reference - NIH. (n.d.). Retrieved January 5, 2019, from https://ghr.nlm.nih.gov/condition/trisomy-13#

Trisomy 18 - Genetics Home Reference - NIH. (n.d.). Retrieved January 5, 2019, from https://ghr.nlm.nih.gov/condition/trisomy-18#

23andMe. (2019). BRCA Genes. Retrieved April 5, 2019, from https://www.23andme.com/brca/

United States Conference of Catholic Bishops. (2009, November 17). Ethical and Religious Directives for Catholic Health Care Services Fifth Edition.

What to Expect, E. (2017, March 08). Noninvasive Prenatal Testing (NIPT). Retrieved March 12, 2019, from https://www.whattoexpect.com/pregnancy/pregnancy-health/noninvasive-prenatal-testing/