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Degree of Knowledge, Potential Barriers, and Reasons for Finding Out About Genetic Testing

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DEGREE OF KNOWLEDGE, POTENTIAL BARRIERS, AND REASONS FOR FINDING OUT ABOUT GENETIC TESTING

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ABSTRACT

Genetic testing is a resource that continues to evolve and change the world of medicine. The availability of such testing has created opportunities for many to better understand and take control of their health. However, studies show that the general population is still lacking in awareness toward genetic testing and what it has to offer.

The purpose of this study was to assess the knowledge about genetic testing and participant's feelings towards finding out about a possible inheritable disease. A nine question survey was distributed to 156 participants at two Minnesota schools and one Minnesota clinic.

The study found that there was a statistically significant difference in awareness of genetic testing with females being more aware than males. There was a statistically significant difference in the knowledge of genetic testing with females having more knowledge than males and participants who had a family history of an inheritable disease having more knowledge than participants who did not have a family history of an inheritable disease. The majority of participants would want to know about a possible inheritable disease and the most answered reason for wanting to know was to protect themselves. Also, various percentages between different demographical factors and their relation to being knowledgeable about genetic testing/counseling were found and among these percentages, they were not significantly different from one another.

This study showed that although the general population has become more aware of genetic testing, there is still room for improvement. By expanding on studies such as this one, more information can be obtained for medical professionals to educate and guide their patients in their medical care. This information can also help patients identify possible self and family altering inheritable diseases.

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CHAPTER 1: Introduction

Introduction

The following chapter will highlight the basic understanding of genetics such as inheritance patterns, the human genome project, common genetically related diseases, and genetic testing. Furthermore, chapter one contains the background, problem statement, purpose, significance, research questions, and definitions of terms.

Background

This background section contains a general description of the evolution of genetics. The information provided ranges from the development of Gregor Mendel's three laws of genetics to the discovery of the Human Genome and how that lead to the possibility of performing specific genetic tests. Most of the information in this section is derived from the book, *Genetics: A conceptual approach* by Benjamin A. Pierce (2012).

The study of genetics is continuing to change the lives of many people in the world of medicine. The father of genetics, Gregor Mendel, in the late 19th century, used his knowledge and insight to study trait inheritance patterns and how these traits were handed down from generation to generation (Pierce, 2012). His work later evolved into understanding the gene, alleles, chromosomes, genotype, and phenotype. He also developed what is known as Mendel's Laws, which represent how genes segregate from, assort with, and dominate over other genes when being passed down from parents to progeny (Pierce, 2012).

Mendel's third law of dominance opened the door for determining whether diseases are genetically passed from one generation to another. Determining whether diseases are genetically passed from one generation to another is accomplished with a pedigree, a simplified readable document created by geneticists that provides insight for family members about their family

history and the possibility of passing a trait to their offspring (Pierce, 2012). A pedigree also shows if a trait is on a non-sex (autosomal) or sex chromosome, a vital factor that can either increase or decrease the chances of passing a disease onto the next generation (Pierce, 2012). The basic understanding that Mendel created has continued to grow and eventually led to the development of medical genetics and more specifically, genetic testing (Pierce, 2012).

Genetic testing is the identification of changes in chromosomes, genes, or proteins that can help determine whether a participant has, or can pass to their children, a certain genetic disorder (U.S. National Library of Medicine, 2015). Through the progression of genetics, thousands of genetic tests have been developed to find predisposing conditions to diseases such as Cystic Fibrosis, Huntington's Disease, and many types of cancer (Pierce 2012). Methods such as molecular, chromosomal, and biochemical genetic testing can help show specific mutations that can lead to the development of such life threatening diseases (What is genetic testing?, 2015). Since the launch of the Human Genome Project in the fall of 1990 that revealed the complete human genome, technology for genetic testing has grown significantly (Pierce, 2012).

The Human Genome Project was a public project that involved 20 different research groups along with hundreds of researchers. The main focus of the project was to develop new automated methods for cloning and sequencing DNA and create physical and genetic maps of the entire human genome (Pierce, 2012). Since its completion in the spring of 2003, the sequence has provided crucial information about development and basic cellular processes (Pierce, 2012). The Human Genome Project also has shown through genetic tests where a participant's genomic makeup differs from others, and the specific location of genes that cause disease and affect traits in humans (Pierce, 2012). The advancement of genetic testing has brought tremendous

opportunities for people to know their possible risk of developing a disease, to make decisions on reproduction, and to help with their anxiety of possibly inheriting the disease (Pierce, 2012).

From the gained knowledge of the human genome sequencing, more genetic disorders and diseases are being identified. This identifiable information through genetic testing brought concern about discrimination against participants who were carriers of disease-causing genes (Pierce, 2012). This discrimination was addressed in the passage of a U.S. Federal law, the Genetic Information Nondiscrimination Act in 2008, which keeps insurers and employers from using such information to make decisions on health-insurance coverage and employment (Pierce, 2012). Other questions that have risen from an individual's genetic information are, who has the right to have access to a participant's genomic information and is it appropriate to use a participant's genetic information to determine certain traits in their potential offspring (Pierce, 2012)?. These reasons, along with others, have contributed to people choosing or not choosing to have genetic testing performed (Pierce 2012).

The purpose of genetic testing is to provide answers for participants who may be predisposed to life threatening diseases. Although these tests are available, whether participants want to be tested for these diseases is still unclear. For participants who want to become more educated about their genetic predisposition, there is evidence that some physicians are skeptical about integrating genetics into their practice, which causes many of the available resources of genetics to be unknown by their patients. Also, the view of testing varies based on age, gender, and education level; these demographics contribute to both a negative and positive public opinion of genetic testing, which may hinder or promote genetic testing (Henneman, Vermeulen, van El, Claassen, Timmermans, & Cornel, 2012). The availability of information about whether participants want to be tested, knowledge of available genetic resources, and demographical

factors are vital parts in promoting the benefits of genetic testing for people with or without a life threatening inheritable disease.

Statement of the Problem

Multiple studies show that the general population is still not sufficiently aware of genetic testing and its benefits (Mai, Vadaparampil, Breen, Mcneel, Wideroff, & Graubard, 2014; Bosompra, Flynn, Ashikaga, Rairikar, Worden, & Solomon, 2000; Macconaill & Garraway, 2010). As of 2010, less than 50% of the United States adult population knew about genetics (Mai et al., 2014). Genetics is an emerging field of preventative medicine that has the potential to significantly reduce the risks of several diseases with the implementation of prophylactic treatment (Meiser et al., 2012). The lack of an participant's awareness of the access to genetic testing can affect the outcome of a life (Bosompra et al., 2000; Mai et al., 2014). Some common demographic barriers that have been found to influence the awareness of genetic testing include age, sex, education, income, a family history of disease/cancer, provider contact, and health insurance status (Bosompra et al., 2000; Mai et al., 2014). Furthermore, participants who are knowledgeable about genetic testing do not always express positive feelings towards finding out if they are at risk for a pre-existing condition (Ngoi, Lee, Hartman, Khin, & Wong, 2013). This problem does not only pose a risk for the participant themselves, but it also poses a risk for their future generations. Therefore, a current assessment of awareness and educational level about genetic testing, and the reasons for wanting to find out about a possible inheritable disease needs to be done to understand more about this field of medicine in the general population.

Purpose of the Study

The purpose of this study is to assess the knowledge about genetic testing and participant's feelings towards finding out about a possible inheritable disease. Although similar

research studies have been conducted, no study has been done in the past six years. The goal of this study is to acquire the knowledge of the current general population in 2016.

Significance of the Study

Misconceptions exist regarding genetic testing. With the increasing knowledge about genetics and the possibilities that genetic testing can predict disease, public opinions toward genetic testing may have changed. Public opinions may also continue to change and genetic testing has the potential for prophylactic treatment in genetic disease (Henneman et al., 2013). Research by Henneman (2013) discussed the importance of the knowledge that the public has on genetic testing regardless of age, gender, or educational level in order to maximize medical efforts in disease prevention. Also, closing the gap of concern and misunderstanding about genetic testing was discussed in research by Wroe, Salkovskis, and Rimes, (1997) and can help genetic services accompany participant's health needs. Closing the gap of concern and misunderstanding about genetic disease can also help implement the most effective strategies to have success in genetic testing and counseling (Wroe et al., 1997). What genes are passed on can determine the future health of a participant and results of this study will give a better understanding of the general population's knowledge of genetic testing.

Research Questions

The research questions addressed in this study are as follows:

1. What level of awareness does our population have in regard to genetic testing?

2. What level of knowledge does our population currently possess about genetic testing?

3. For those in our population who want to know about a possible inheritable disease, what are the reasons why?

4. What are some of the barriers and demographical factors (gender, socioeconomic status, race, age, education) that are leading to a lack of knowledge in regard to genetic testing?

Definitions

The following definitions are important terms, which are provided below so the reader understands this chapter and the entirety of the study.

Allele: One or two or more alternate forms of a gene (Pierce, 2012).

Autosome: Chromosome that is the same in males and females; nonsex chromosome (Pierce, 2012).

Biochemical Genetic Testing: the study of the amount or activity level of proteins; abnormalities in either can indicate changes to the DNA that result in a genetic disorder (U.S. National Library of Medicine, 2015).

Chromosome: Structure consisting of DNA and associated proteins that carries and transmits genetic information (Pierce, 2012).

Chromosomal Genetic Testing: analyzing whole chromosomes or long lengths of DNA to see if there are large genetic changes, such as an extra copy of a chromosome, that cause a genetic condition (U.S. National Library of Medicine, 2015).

Gene: Genetic factor that helps determine a trait; often defined at the molecular level as a DNA sequence that is transcribed into an RNA molecule (Pierce, 2012).

Genetic Counseling: Educational process that attempts to help patients and family members deal with all aspects of a genetic condition (Pierce, 2012).

Genome: Complete set of genetic instructions for an organism (Pierce, 2012).

Genotype: The set of genes possessed by and individual organism (Pierce, 2012).

Molecular Genetic Testing: the study of single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder (U.S. National Library of Medicine, 2015).

Mutation: Heritable change in genetic information (Pierce, 2012).

Phenotype: Appearance or manifestation of a characteristic (Pierce, 2012).

Sex Chromosomes: Chromosomes that differ morphologically or in number of males and females (Pierce, 2012).

Conclusion

The work of Gregor Mendel and the discovery of the Human Genome, set a foundation of what is now a growing field in genetic testing. These discoveries, along with many others, has raised questions of whether participants are aware and knowledgeable about genetic testing and what genetic testing has to offer. By finding out the answers to these questions, steps can be made to determine how to raise awareness of the benefits of genetic testing. Also, such answers can aid in the efforts to educate participants who are or could be affected by life threatening diseases.

CHAPTER 2: Literature Review

Introduction

Genetic testing of patients when determining suitable treatment options is an expanding area of medicine that both researchers and the general public need to know more about. This literature review will outline studies focusing on genetic testing, genetic counseling, how educated the general public is about genetic testing, reasons for wanting to find out about an inheritable disease, and the factors that keep the general public from becoming educated about genetic testing. Additionally, the human genome, processes of genetic sequencing, and the mutations of normal cellular genes, are discussed to further explain the basis of genetic testing.

Genetic Testing

Three billion base pairs of genes are in the human genome (Macconaill & Garraway, 2010). A reference genome was established in its entirety by scientists in the early 2000's, but the functionality and purpose of these three billion genes is still being expanded on today (Macconaill & Garraway, 2010). In the early 1900's there was debate about whether cancer originated from a deranged genome or caused by viruses. This debate sparked further research on the origin of cancer, and led to the current understanding that cancer does not arise from deranged genomes, but rather it arises from the mutations of normal cellular genes (Macconaill & Garraway, 2010).

Researchers have stressed the importance of determining which of the normal cellular gene mutations are drivers, and which of these mutations are passengers. Genes that are considered drivers are those that are most commonly seen in correlation with a particular disease state, whereas passenger mutations are those that are either silent or rarely seen in correlation with a disease state (Macconaill & Garraway, 2010). The best way to determine which genes are

most clinically relevant are by taking DNA samples from participants that display the phenotype of a particular disease. As the number of DNA samples increases, researchers are able to more definitively assess the phenotypic display of a disease state and how the disease state correlates to alterations in a disease state genome (Macconaill & Garraway, 2010).

One of the most effective methods in determining genetic mutations is by targeting Single Nucleotide Polymorphisms (SNPs) through direct sequencing of Polymerase Chain Reaction (PCR)-amplified segments of the genome from several participants with the same disease presentation (Bentley, 2000). These disease-altered genomic sequences can be compared to that of the reference genome in determining the loci of concern. Furthermore, the method of determining genetic mutations by targeting SNP's through direct sequencing of PCR-amplified segments can be used to compare the genetic mutations amongst participants with the same phenotypic display of disease to determine the reoccurring mutated gene(s) (Bentley, 2000). Determining genetic mutations by targeting SNP's through direct sequencing of PCR-amplified segments, is commonly used in determining the presence of mutated genes in participants with a family history or a current presentation of an inheritable disease(s). Other methods of targeted gene sequencing in genetic testing include denaturing high-pressure liquid chromatography, single-stranded conformation polymorphism, microarrays, and gene chips.

One study in particular pointed out multiple downfalls of the genetic testing technique mentioned in the previous paragraph. The researchers state that targeted gene sequencing fails to reveal the complete landscape of all the genomic changes that occur in cancer (Lizardi, Forloni, & Wajapeyee, 2011). These researchers express that doing a full genomic analysis rather than targeting a specific gene sequence can make gene mutations or other genome alterations applicable to any human cancer. With enough data and subjects, Lizardi, et al. (2011), suggest

that we should be able to notice trends of cancer and other diseases on a broad-based spectrum rather than a loci-focused approach. The same researchers did go on to acknowledge the fact that genome-wide cancer sequencing is much more costly and time-consuming. As of 2010, the cost of complete cancer genome sequencing was $5,000 (Lizardi et al., 2011). This cost is significantly reduced when compared to the cost of sequencing ten years ago when it cost upwards of 1 million dollars (Lizardi et al., 2011). This trend of decreased costs is thought to continue into the future until complete human genome sequencing becomes a truly cost effective approach in disease heritability research (Macconaill & Garraway, 2010).

Genetic Counseling

The identity of genes, the mutations of genes, and the possibility of conducting a genetic test are handled by genetic counselors. Genetic counselors are trained professionals who work with participants and families in discussing a medical history or increased risk for a genetic condition (umn.edu). Genetic counseling is a rapidly growing profession and is becoming more accessible to the general population each year. It is projected that this field of medicine will grow 41% by 2022 as genetic research continues to show advancement (umn.edu). These professionals typically have educational backgrounds in biology, genetics, nursing, psychology, and/or public health.

Genetic counselors are not only skilled in communication, compassion, and listening, but they are also educated about the hereditary risks and preventative measures of each inheritable medical condition along with being able to relay often disappointing news with empathy and respect. Genetic counselors often work together with a clinical geneticist. Clinical Geneticists are medically trained professionals that may perform clinical exams and order lab tests to

diagnose the causes of birth defects and other genetic conditions (cdc.gov). These professionals will typically deal with the treatment and prevention counseling of patients.

Reasons that participants may seek out genetic counseling include: a family history of a genetic condition, diseases that are more common in certain ethnic groups, and to find out if there is a genetic cause for developmental delays or other underlying health problems (umn.edu). The most interest for advancement in genetics research for reasons related to pre-conception (umn.edu). Many couples will seek out genetic counseling to learn about risks of becoming pregnant if the mother is older, to learn about the effects of being exposed to x-rays, chemical, illness, or prescribed or illicit drugs while pregnant. Furthermore, couples will go through genetic counseling to discuss infertility, miscarriages, genetic conditions or birth defects occurring in a previous pregnancy, and to take general steps to get ready for a healthy pregnancy and care of a newborn baby (umn.edu).

Level of Education with Genetic Testing

Many studies provide information about the types of genetic tests that are available and how they can be of benefit to various participants. But, not many studies expand on the level of education that participants obtain when it comes to genetic testing and what is available to them. A few studies cited below provide useful information about the varying levels of participants who have the possibility of obtaining an inheritable disease and the demographical factors that can affect this level.

Advancements in genetic testing have provided information for patients about the specific risks and inheritance of Parkinson's Disease (PD). However, what is unknown is whether those who are at risk would want to have genetic testing done, especially knowing that there is no preventative therapies available for PD (Scuffham, Mclnerny-Leo, Ng, Mellick,

2014). Scuffham et al., (2014), investigated the knowledge towards genetic testing with those affected by PD. This study consisted of a cross-sectional postal survey of patients who were diagnosed with PD by using a standardized questionnaire. This questionnaire contained 41 items that included four different sections, one of them being knowledge about genetics and PD (Scuffham Mclnerny-Leo, Ng, Mellick, 2014).

The results of the knowledge about genetics and PD showed that the majority of the respondents (77.5%) did not have much knowledge in the area of genetics and the vast majority of the respondents (97%) supported genetic testing for PD. (Scuffham, Mclnerny-Leo, Ng, Mellick, 2014). These results showed that the lack of knowledge in the area of genetics had little effect on the actual performance of genetic testing for PD (Scuffham, Mclnerny-Leo, Ng, Mellick, 2014). These results also gave insight into what factors contribute to a lack of knowledge about genetic testing and what still needs to be accomplished to reach an individual's health needs.

The aim of genetic testing is also to gain more knowledge about inheritable diseases, help early detection of possible inheritable diseases, and develop possible future prophylactic treatments so an participant's health concerns can be addressed. Research done by Etchegary, (2014) and Henneman, Vermeulen, Van El, Claassen, Timmermans, and Cornel (2013), highlighted the general public's knowledge about genetic testing in order to better understand and treat a participant's health concerns. Etchegary found that varying levels of health literacy affected the understanding of genetic testing (Etchegary, 2014). Another study found that having a higher education level and being a women were associated with being more knowledgeable about genetic testing (Henneman et al., 2013). These factors will also help guide in the promotion of genetic testing.

Reasons for Wanting to Know About an Inheritable Disease

An important aspect of why this research is being conducted is the incentives participants have to seek out genetic testing. Determining the reasons individuals have for wanting to know about an inheritable disease is a significant step in the promotion of genetic testing. A few inheritable diseases that are sought out and are able to be genetically tested for are Alzheimer's Disease, Huntington's Disease, and breast cancer.

A common neurodegenerative disease that is often passed down from one generation to the next is Alzheimer's disease. The REVEAL (Risk Evaluation and Education for Alzheimer's Disease) study explored reasons children of parents with Alzheimer's disease sought genetic testing (Hurley, Harvey, Roberts, Wilson-Chase, Lloyd, Prest, et al., 2005). This study included 60 interview transcripts that included 157 stories of why these participants volunteered for the REVEAL study and within these stories were reasons why the participants sought genetic testing. The research included qualitative analysis that focused on two central constructs, altruism and learning about Alzheimer's disease (Hurley et al., 2005).

Altruism, as defined by the research group, is helping others by advancing science (Hurley et al., 2005). An interviewee shared, "I was feeling, and I still feel, if there's something I can do to try to help advance the knowledge, or the treatment, or the understanding, or some greater good, if there's any greater good that can come from my father's illness, I'm happy to try to help get to that" (Hurley et al., 2005, pg. 376). The participants' reasons for their altruism included, motivation from the parent with Alzheimer's, the safety and future knowledge for their family, and personal interests (Hurley et al., 2005).

The second construct, learning, was defined by the research group as curiosity for self and/or science to search for information (Hurley et al., 2005). Searching for information under

the second construct of learning was explained through the three concepts of planning, prevention, and need to know (Hurley et al., 2005). Through the concept planning, participants considered future actions they would take for themselves and others. One participant stated, "When they wanted to know if I wanted to do the program, I go, sure, I want to see where I'm at. Because I can make some decisions in my life that I could take care of everything before and not have everybody else stress about it. I figured I needed to know because what if I get it? Who's going to take care of me?" (Hurley et al., 2005, pg. 378). The concept of prevention looked at reducing one's risk for Alzheimer's disease. One participant said, "My thought at doing this was to know so that if something comes down the pipe, that I could take that could circumvent it or prevent it, that I would be the first in line. That was my premise" (Hurley et al., 2005, pg. 378). The concept of need to know was expressed by participants because these participants expressed a fear of developing Alzheimer's disease or, of feeling anxious about symptoms that indicate early onset Alzheimer's. The fear of developing Alzheimer's disease or feeling anxious about symptoms that indicate early onset Alzheimer's disease was voiced by a female participant when she shared, "But I got scared to death that I inherited this, and that's why I was anxious to get in the study and see. I wanted to know if I had the gene." (Hurley et al., 2005, pg. 379).

Another neurodegenerative disease with genetic ties is Huntington's Disease. A study done by Scuffham and MacMillan (2014), looked at who sought presymptomatic genetic testing for Huntington's Disease and why the participants sought out genetic testing (Scuffham & MacMillan, 2014). The aims of the study included, to quantify the characteristics of those seeking presymptomatic testing for Huntington's Disease and to identify what the participant's motivations were for testing (Scuffham & MacMillan, 2014).

The results of this study showed motivations for pursuing genetic testing included, family planning, planning for future endeavors, needing to know if they carried the gene for Huntington's disease, and informing their children (Scuffham & MacMillan, 2014).

Outside of neurodegenerative diseases, studies have been performed to research genetic testing for breast cancer. Bruno, Digennaro, Tommasi, Stea, Danese, Schittulli, & Paradiso, (2010), researched reasons for wanting or not wanting genetic testing of breast cancer and compared women who were affected by breast cancer to women who were unaffected by breast cancer (Bruno, et al., 2010). The results showed that the most frequent reasons to seek genetic testing, whether women were affected or unaffected by breast cancer, were to learn about their children's risk, to help advance research, and to determine the frequency of screening tests. The most frequent reasons for not having genetic testing was, women's concerns about the effect on their families and the disruption of life and projects (Bruno et al., 2010).

Factors Leading to a Lack of Education About Genetic Testing

Although genetic testing has made large advancements in recent years, it is unclear whether participants are aware of these advancements. Being able to assess the educational level participants have on genetic testing is important when determining how to provide such tests to participants who may desire them. Factors leading to a lack of education about genetic testing that are highlighted in this section include, whether or not women were affected with breast cancer, difference in years, gender, age (18 or older), education level, marital status, region of residence, and whether or not you have children.

As referenced above in the section, reasons for wanting to know about an inheritable disease, the study conducted by Bruno et al., 2010, also researched if being affected with breast cancer or not being affected with breast cancer would contribute to a lack of education about

genetic testing. Bruno et al's., results showed that, when answering yes or no to having been educated about genetic testing for breast cancer, the women who were not affected by breast cancer said yes slightly more (56%) compared to no (44%) then the women who were affected by breast cancer who said yes slightly less (53%) compared to no (47%) (Bruno et al., 2010).

Another study by Henneman et al., 2013, also researched if the difference in years (2002 or 2010) would contribute to a lack of education about genetic testing. The results showed that, participants surveyed in 2002 had been educated about genetic testing prior to taking the survey slightly more (57%) then participants surveyed in 2010 (55%) (Henneman et al., 2013).

A third study researched how demographical factors such as gender, age (18 or older), education level, marital status, region of residence, and whether you have children contributed to being educated about genetic testing (Roy, Pallai, Lebwohl, Taylor, & Green, 2015). The results showed that, of the participants surveyed, if you were married, have children, or pursued education beyond high school, you were more educated about genetic testing than if you were not married, did not have kids, or did not pursue education beyond high school (Roy et al., 2015). There was no significant difference between being educated or not being educated about genetic testing when comparing gender, age (18 or older), or region of residence (Roy et al., 2015).

Conclusion

The purpose of this literature review is to outline the previous studies that have focused on genetic testing, genetic counseling, how educated participants are about genetic testing, participants' reasons for wanting to find out about an inheritable disease, and the factors that keep participants from becoming educated about genetic testing. Each study proved that although research has been done to explain the research questions provided in chapter one, more

research must be done to further justify these research questions. By examining previous research, this study is able to adjust its methodology to target the current population's reasons for wanting and knowledge of genetic testing. Also, with the increase in interest towards genetic testing, learning what factors keep participants from knowing or learning about genetic testing will be important.

CHAPTER 3: Methodology

Introduction

The purpose of this study was to assess the knowledge of the general population about genetic testing and their feelings towards finding out about a possible inheritable disease. The study addressed the four questions of:

1. What level of awareness does our population have in regard to genetic testing?
2. What level of knowledge does our population currently possess about genetic testing?
3. What are the reasons why our population wants to know about inheritable diseases?
4. What are some of the barriers and demographical factors (gender, socioeconomic status, race, age, education) that are leading to a lack of knowledge in regard to genetic testing?

This chapter contains the following information: study design, study subject variables, population, instrumentation, validity and reliability, procedures, data analysis, and limitations.

Study Design

The research project was a prospective quantitative study that focused on questioning faculty members from two different elementary schools and patients at a plastic surgery clinic. Results concerning demographic factors, knowledge of genetic testing, and wanting to know about the possibility of developing an inheritable disease was collected through a survey. The survey was distributed by the principal at the location of Lake Harriet Lower Community School in Minneapolis, Minnesota, a teacher at Bridgewater Elementary School in Northfield, Minnesota, and a registered nurse injectionist at Dr. Michael Fashing Clinic in Plymouth, Minnesota. See appendix A for permission letters obtained by both the schools and the clinic.

By not requesting names or personal information of the participants, confidentiality is guaranteed.

Study Subject Variables

This study gathered information to see if there is a relationship between independent and dependent variables. The independent variables of this study were, age (22-70), gender, ethnicity, education level, and family history. The dependent variables of this study were, how educated the participant is about genetic testing/counseling and finding out about the possibility of developing an inheritable disease.

Population

The participants of the survey were teachers from the locations of; Bridgewater Elementary School in Northfield, Minnesota, Lake Harriet Lower Community School in Minneapolis, Minnesota, and patients from Dr. Michael Fashing Clinic in Plymouth, Minnesota. The participants were male or female, of any ethnicity, and ranged from 22-70 years of age, making them eligible to complete the survey without permission from a guardian. The participants who received the survey could have declined to complete the survey at any time if they chose to do so. There was no repercussions from the participant's employer whether or not the participant completed the survey. The goal sample size for this survey was 100 participants.

Instrumentation

The nine question survey (see appendix B) used in the study was divided into three sections that attempted to answer the four aforementioned research questions. The first four questions of the questionnaire obtained demographic information to answer the research question that aimed to answer whether gender, race, age, and education level acted as barriers to a lack of knowledge in regards to genetic testing.

The fifth question of the survey addressed whether the participant had a family history of an inheritable disease which was defined at the top of the survey as "a disease that is passed on from parent to child." This question was in place to examine whether a family history of a genetic disorder influenced the level of education a participant may have had about genetic testing.

Questions six and seven were directly aimed at answering the research questions of how much awareness and knowledge our population had toward genetic testing. The first of these two questions was a yes/no question that addressed whether or not the participant was aware of genetic testing. The second of these two questions used a five point Likert scale to measure the degree of knowledge the participant had about genetic testing.

The final two questions answered the research question regarding the reasons why the participant would want to find out about a possible inheritable disease. The first of these two questions asked whether the participant would want to know or not want to know about an existing inheritable disease. The final question of the questionnaire gave the participant four options of why they would want to get genetically tested. The questionnaire gave the participant the ability to circle all of the reasons that may have applied for them.

Validity and Reliability

Our questionnaire had been formulated by an acclimation of multiple previous research surveys, but ultimately designed by ourselves in order to specifically answer our research questions. As a result, this was a novel survey making the validity and reliability difficult to determine at the time. In attempt to increase and determine the current validity and reliability, we selected a group of ten individuals in the community that ranged in age from 23-60, were both male and female, and had various education levels to take our survey. The demographics of

these ten participants was representative of the population that we were surveying and had provided us with feedback about their understanding of the survey's contents and clarity. Changes that were made to the survey based on this feedback included, adding a simplified definition of inheritable disease at the top of the survey for clarification, rewording question 5 to say, "Do you, or your family, have a history of an inheritable disease" from "Does your family have a history of an inheritable disease" to make sure the participants taking the survey includes themselves when answering question 5, and adding "Flip over to complete questions 8 and 9" on the bottom of the first page of the survey to assure that the participants taking the survey do not skip questions 8 and 9 on page 2 of the survey.

Procedures

The principal of Lake Harriet Lower Community School in Minneapolis, Minnesota, a teacher at Bridgewater Elementary School in Northfield, Minnesota, and a registered nurse injectionist at Dr. Michael Fashing Clinic in Plymouth, Minnesota, were asked to hand out a paper copy of the survey to faculty members at these schools. At Lake Harriet Lower Community School, the survey was handed out on Tuesday August 23, 2016 at the opening staff meeting for the 2016-2017 school year. The meeting took place in the art room at 8:00 AM with approximately 40 staff members in attendance. The principal read a script to instruct the faculty members of how to complete the survey (see appendix D) and then handed out the survey with an informed consent document attached at the top of the survey (see appendix A) at the end of the staff meeting which occurred approximately at 12:00 PM. The principal then immediately collected each survey from the faculty members that decided to participate and stored them in a secure envelope. This secure envelope was then collected by the researcher on Sunday August 28th, 2016 at 1:00 PM.

At Bridgewater Elementary School, the survey was handed out during the teacher preparation or staff development week from Monday August 29th – Friday September 2nd, 2016. The teacher placed the surveys in to faculty member's personal mail boxes on Monday August 29th at 8:00 AM with instructions for faculty members on how to complete the survey (see appendix D) and an informed consent document (see appendix A) attached at the top of each survey. Each faculty member that decided to participate had 5 days to complete the survey and return it back to the teacher's personal mailbox. The teacher collected all of the surveys completed on Friday September 2nd, 2016 at 4:00 PM and placed them in a secure envelope. A researcher then collected the secure envelope on Saturday September 3rd, 2016 at 6:00 PM.

At Dr. Michael Fashing Clinic, the survey was handed out on Monday's, Tuesday's, and Friday's starting on Monday August 22nd and ended on Friday September 16th, 2016. The injectionist distributed a survey to each patient before their scheduled appointment with instructions for the patient on how to complete the survey (see appendix D) and an informed consent document (see appendix A) attached at the top of each survey. Each patient that decided to participate was given the opportunity to complete the survey while the injectionist was drawing up their medication for their appointment. The injectionist then collected the surveys that were completed and after the appointment, placed them in a secure envelope. On Friday September 16th, 2016 at 4:00 PM, the injectionist took the secure envelope home and then a researcher collected the secure envelope on Saturday September 17th, 2016 at 1:00 PM.

Data Analysis

Data collected from the questionnaires was analyzed using the SPSS program. Descriptive statistics regarding a participants' demographics and multiple correlation regression was used on the data collected in order to address the research questions. The data representing

our population was then related to the data representing populations in the studies we analyzed in our Literature Review. The data from our study was stored on a password protected computer while analyzing the data. After the analysis was completed, our data was locked in the Bethel University Physician Assistant program storage and will be locked there for for a minimum of five years.

Limitations

The limitations for the study were: the participants who took the survey were from a small sample size of the general population. The populations of exclusion were anyone under the age of 22, participants who were illiterate, or participants who were non-English reading. The participants were from Plymouth, Northfield, and Minneapolis, Minnesota. These locations may not have represented the demographic population in the United States. The participants may not have answered the survey honestly because they did not want to look uneducated given their profession as educators. Another limitation was that the participants may not have wanted to take the time to do the survey. The participant handing out the survey could have forgotten to distribute the surveys and the participant filling out the survey could have forgotten to hand the survey in or could have lost it all together. Also, the surveys distributed by the teacher at Bridgewater Elementary School may not have been completed correctly because the instructions were included in print form on the top of the survey instead of being read to the faculty members.

CHAPTER 4: Results and Data Analysis

Introduction

The purpose of this study was to find out the current level of awareness and the degree of knowledge the general population has about genetic testing. Our first research question is "What level of awareness does our population have in regards to genetic testing?" In order to answer this question, we did a chi-square test to descriptively analyze the variance, if any, amongst the demographic groups of age, gender, education level and family history of an inheritable condition. Our second research question is "What level of knowledge does our population currently possess about genetic testing?" The score data that was collected tested the mean differences between the same four demographical groups. Since there are more than two groups being analyzed, an ANOVA test was used. Our final two research questions are "For those in our population who want to know about a possible inheritable disease, what are the reasons why?" and "What are some of the barriers and demographical factors (gender, age, education, and +/- family history) that are leading to a lack of knowledge in regard to genetic testing?" Both of these questions are answered descriptively by frequency and categories of response so no statistical tests were needed.

There were 156 participants in this study. There were only 11 participants who indicated their race was something other than "non-hispanic white", therefore the variances among race were not compared. These participants were still included in the pool of surveys because race was disregarded altogether. The three demographical factors that were compared ended up being age, gender, and education level. Furthermore, we directly compared individuals that have a family history of an inheritable disease, those who do not have a family history of an inheritable disease, and those who are unsure if they have a family history of an inheritable disease.

Demographics

Out of the 156 participants, twenty-one were 18-29 years old (13.4%), twenty-four were 30-39 years old (15.4%), forty-one were 40-49 years old (26.3%), fifty-one were 50-59 years old (32.7%), and nineteen were 60+ years old (12.2%). Out of the 156 participants, 26 were male (16.7%) and 130 were female (83.3%). Out of the 156 participants, 20 had high school as the highest level of education (12.8%), 19 had an associate's degree as the highest level of education (12.2%), and 117 had a bachelor's degree or higher as the highest level of education (75.0%). Out of the 156 participants, 45 had a family history of an inheritable disease (28.8), 71 said that they did not have a family history of an inheritable disease (45.5%), and 40 did not know if they had a family history of an inheritable disease (25.6%).

Research Question 1 – Level of Awareness

A Chi-square test was used to compare the level of awareness in regards to genetic testing of age, gender, education level, and family history of an inheritable disease. The results listed in this section are in response to the survey question: Have you heard of genetic testing/counseling? The null hypothesis is that there are no significant differences within each demographical factor in levels of awareness in regards to genetic testing. The difference in awareness of genetic testing among age groups is not statistically significant therefore the null hypothesis cannot be rejected ($p > .05$; see table 1). The difference in awareness of genetic testing among gender groups is statistically significant therefore the null hypothesis *can* be rejected ($p < .05$; see table 1). The difference in awareness of genetic testing among education level groups is not statistically significant therefore the null hypothesis cannot be rejected ($p > .05$; see table 1). The difference in awareness of genetic testing among family history groups is not statistically significant therefore the null hypothesis cannot be rejected ($p > .05$; see table 2).

Table 1

Awareness of Genetic Testing

	Age					Gender		Education		
	18-29	30-39	40-49	50-59	60+	Male	Female	HS or below	Assoc or higher	Bach or higher
No I have not heard of genetic testing/counseling	27% (6/21)	12.5% (3/24)	14.6% (6/41)	19.6% (10/51)	10.5% (2/19)	**34.6% (9/26)**	**13.8% (18/130)**	4.5% (7/156)	1.9% (3/156)	10.9% (17/156)
Yes I have heard of genetic testing/counseling	71.4% (15/21)	87.5% (21/24)	58.5% (35/41)	78.8% (41/51)	89.5% (17/19)	**65.5% (17/26)**	**86.2% (112/130)**	8.3% (13/156)	10.3% (16/156)	64.1% (100/156)

Note: HS = high school; Assoc = Associates degree; Bach = Bachelor's degree; Statistical significance is in boldface.

Table 2

Family History and Awareness of Genetic Testing (p value = .08)

Response	Yes I have a family history	No I do not have a family history	I do not know if I have a family history
No I have not heard of genetic testing/counseling	1.9% (3/156)	9.6% (15/156)	5.8% (9/156)
Yes I have heard of genetic testing/counseling	26.9% (42/156)	35.8% (56/156)	19.9% (31/156)

Research Question 2 – Degree of Knowledge

An ANOVA test was used to compare the degree of knowledge in regards to genetic testing of age, gender, education level, and family history of an inheritable disease. The results listed in this section are in response to the survey question "On a scale of 1-5 (1 being the least educated and 5 being the most educated), how knowledgeable are you about genetic testing/counseling?" The null hypothesis is that there are no significant differences within each demographical factor in degree of knowledge in regards to genetic testing. There were less than

10 participants that knew a lot about genetic testing and that called themselves an expert on genetic testing (numbers 4 and 5 on the survey), so for statistical purposes we adjusted our analysis. Rather than having a scale of 1-5, we grouped the 4s and 5s into our group of 3s to make one group that knows a "moderate amount or more about genetic testing/counseling." A score of 1 means that the participant knew nothing about genetic testing and a score of 2 means that the participant knew a minimal amount about genetic testing. The difference in degree of knowledge in regards to genetic testing among age groups is not statistically significant therefore the null hypothesis cannot be rejected ($p > .05$; see table 3). The difference in degree of knowledge in regards to genetic testing among gender groups is statistically significant therefore the null hypothesis *can* be rejected ($p < .05$; see table 4). The difference in degree of knowledge in regards to genetic testing among education level groups is not statistically significant therefore the null hypothesis cannot be rejected ($p > .05$; see table 5). The difference in degree of knowledge in regards to genetic testing among family history groups is statistically significant therefore the null hypothesis *can* be rejected ($p < .05$; see table 6).

Table 3

Age and Degree of Knowledge

Group	*Mean, St. Dev., n*	p = 0.17
18 to 29 Years	1.81, 0.75, 21	
30 to 39 Years	2.17, 0.64, 24	
40 to 49 Years	2.22, 0.69, 41	
50 to 59 Years	2.12, 0.68, 51	
60+ Years	2.32, 0.75, 19	

Table 4

Gender and Degree of Knowledge

Group	*Mean, St. Dev., n*	p = 0.05
M	1.88, 0.71, 26	
F	2.18, 0.69, 130	

Table 5

Education Level and Degree of Knowledge

Group	*Mean, St. Dev., n*	p = 0.25
High School Grad or Lower	1.90, 0.64, 20	
Associate's Degree	2.11, 0.74, 19	
Bachelor's Degree or Higher	2.18, 0.70, 117	

Table 6

Family History and Degree of Knowledge

Group	*Mean, St. Dev., n*	p value = .04
Yes	2.36, 0.68, 45	
No	2.03, 0.68, 71	
Idk	2.08, 0.73, 40	

Research Question 3 – Reasons for Wanting to Know About an Inheritable Disease

134 out of the 156 participants reported that they would want to know about an inheritable disease (85.9%) therefore our sample size for this research question is 134. The 22 people that indicated they would not want to know about a possible inherited disease were instructed to skip this question on the survey. Participants were given 5 different reasons for wanting to know about an inheritable disease and were allowed to check all that applied (see table 7). 72% of this population reported “to protect myself” as one of the reasons for wanting to know about an inheritable disease. 68% reported “to protect my children” as a reason, 57%

reported for “peace of mind,” 32% for “general curiosity,” and 10% selected “other” reasons for wanting to know.

Table 7

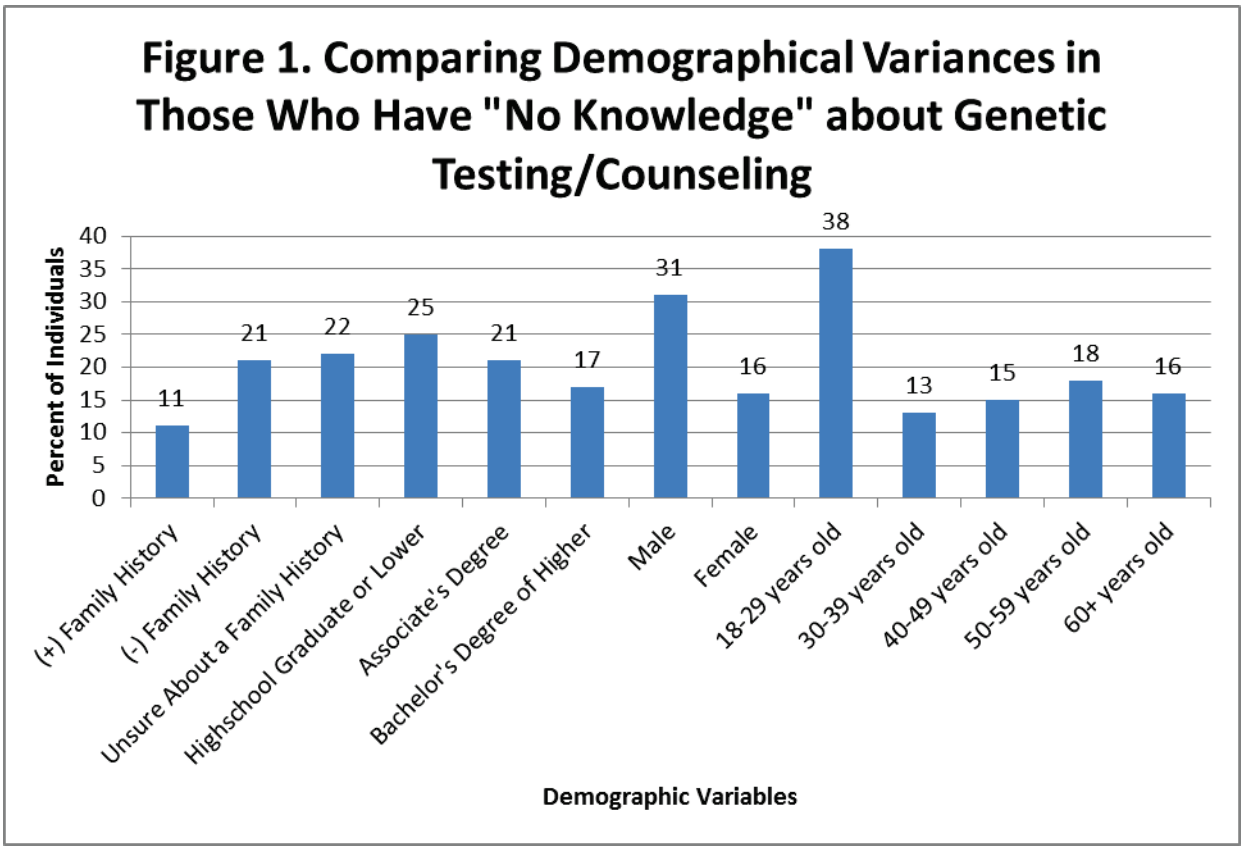
Percentages of Reasons to Know About an Inheritable Disease

Reason	Value	Percentage (conditional)
1	To protect myself	72% (96/134)
2	To protect my children	68% (91/134)
3	Peace of mind	57% (76/134)
4	General Curiosity	32% (43/134)
5	Other	10% (14/134)

Research Question 4 – Barriers Leading to a Lack of Knowledge in Regards to Genetic Testing

Our fourth research question aimed to identify any barriers that may be leading to a lack of knowledge in regards to genetic testing. The degree of knowledge has been divided into three categories: no knowledge, minimal knowledge, and moderate or higher degree of knowledge. As previously mentioned, scores of 3-5 make up a group that knows a *moderate amount or more* about genetic testing/counseling, a score of 2 means that the participant knew a *minimal amount* about genetic testing, and a score of 1 means that the participant had *no knowledge* about genetic testing. The four possible barriers being analyzed are age, gender, highest level of education, and a negative family history. We found that 38% of our population aged 18-29 has no knowledge about genetic testing/counseling, 13% of our population aged 30-39 has no knowledge about genetic testing/counseling, 15% of our population aged 40-49 has no knowledge about genetic testing/counseling, 18% of our population aged 50-59 has no knowledge about genetic testing/counseling, and 16% of our population aged 60+ has no knowledge about genetic testing/counseling (figure 1). We found that 16% of our female population has no knowledge

about genetic testing/counseling compared to 31% of our male population has no knowledge about genetic testing/counseling (figure 1). We found that 25% of our population that had a high school degree or less as their highest level of education has no knowledge about genetic testing/counseling, 21% of our population that had an associate's degree as their highest level of education has no knowledge about genetic testing/counseling, and 17% of our population that had a bachelor's degree or higher has no knowledge about genetic testing/counseling (figure 1). Finally, we found that having a negative family history of an inheritable disease resulted in 21% of our population having no knowledge about genetic testing/counseling about genetic testing/counseling whereas having a positive family history of an inheritable disease resulted in only 11% of our population having no knowledge about genetic testing/counseling. About one fourth of our population was unsure of whether or not they had a family history. Out of this population, 22% has no knowledge about genetic testing/counseling (figure 1).



Conclusion

According to the data, the lone significant finding for the question "What level of awareness does our population have in regards to genetic testing?" is that women are more aware of genetic testing than men accordingly. The significant findings for the question "What level of knowledge does our population currently possess about genetic testing?" are that women are more knowledgeable about genetic testing than men and those with a family history of an inheritable disease are more knowledgeable about genetic testing than those without a family history. It was also found that that the most common reason for finding out about a potential inheritable disease is to protect the individual taking the survey. The second most common reason is to protect the individual's children.

CHAPTER 5: Conclusion

Introduction

The purpose of this study was to assess the knowledge about genetic testing and participant's feelings towards finding out about a possible inheritable disease. In order to accomplish this purpose, a nine question survey was distributed to participants at two Minnesota schools and one Minnesota clinic. The survey asked the participants to answer demographic questions, a question about the participants' current knowledge level in regards to genetic testing, and a question about whether the participant would want to know if they had an inheritable disease or not and if so, why? This chapter discusses the findings of this study, revisits previous research studies and compares the results of those research studies to this study, addresses limitations that were encountered in this study, gives suggestions for future research, and provides a conclusion to summarize the study and its findings.

Discussion

Using SPSS and Excel, all four research questions were answered. As stated in chapter four, the results of research question one, looking for how aware the population was in terms of knowing that genetic testing was an available resource, showed a statistically significant difference in the awareness of genetic testing with females being more aware than males. Other demographics evaluated in this study such as age, education level, and family history did not show a statistically significant difference in awareness. The second research question, looking for how much knowledge the population had about genetic testing, found that there was a statistically significant difference in the knowledge of genetic testing with females having more knowledge than males and participants who had a family history of an inheritable disease having more knowledge than participants who did not have a family history of an inheritable disease.

Other demographical factors such as age and education level did not show a statistically significant difference in the amount of knowledge a participant had. The third research question, looking for reasons why a participant would want to be genetically tested for a possible inheritable disease, found that the majority of the participants would want to know about a possible inheritable disease and the most answered reason for wanting to know was to protect themselves. The fourth research question, looking for what barriers and demographical factors are leading to decreased knowledge about the resource of genetic testing, found various percentages between different demographical factors and their relation to being knowledgeable about genetic testing/counseling. These percentages were not significantly different from one another.

Previous Research

When comparing this study to others that were referenced in the literature review, there are some key differences and similarities. One difference found between this research and previous research is in the participant demographics. This study found a statistically significant difference in the knowledge of genetic testing between genders and not in relation to age, education level, and family history. The study conducted by Henneman et al., (2013) found that education level and gender affected knowledge about genetic testing. The study conducted by Roy et al., (2015) found that education level affected your knowledge level towards genetic testing for Celiac disease and age (18 or older) and gender did not have an effect on your knowledge about genetic testing. This research study found that 67.9% of participants reported they knew minimal or nothing about genetic testing (Roy et al., 2015). In contrast, Scuffham et al's research concluded that 77.5% of their population did not have much knowledge in the area of genetics (Scuffham et al., 2014).

Three studies referenced in the literature review were able to be compared to the third research question, for those in our population who want to know about a possible inheritable disease, what are the reasons why? Research question three of this study found that the top two reasons for seeking genetic testing to know about an inheritable disease was "to protect myself" and "to protect my children". A study conducted by Hurley et al., (2005) gathered qualitative answers for wanting to seek genetic testing for Alzheimer's disease and found reasons such as wanting to help others by advancing science, and curiosity for self and/or science to search for information (Hurley et al., 2005). A second study conducted by Scuffham and Macmillan (2014) found that participants sought out genetic testing for Huntington's disease because of motivations such as family planning, planning for future endeavors, needing to know if they carried the gene for Huntington's disease, and informing their children. A third study conducted by Bruno et al., (2010) found that participants affected or not affected by breast cancer sought genetic testing for the reasons of learning about their children's risk, to help advance research, and to determine the frequency of screening tests. Each study differed in the specific reasons for wanting to seek genetic testing and how they were reported, but all of the studies were similar in that the reasons related to those of protecting themselves and or protecting their children.

Limitations

There were several limiting factors in this study. One limitation was the population that the survey was distributed to. For this study, the goal was to represent the general population as best as possible. Although three different study sites were used to best reproduce the demographics of the general population, predominantly non-Hispanic white populations (145 of the 156 participants) from three Minnesota locations does not best represent the general population. Also, although there were enough male participants to report differences between

genders, with a more balanced overall population of males and females, the study could be more thorough with its results With more resources and a larger, more diverse population, this study could have better represented the general population.

Another limitation to this study was the sample size that was collected. Originally, it was a goal that the study would gather 100 participants. This study was able to collect 156 surveys which exceeded expectations, but a larger sample of participants would have provided more significant data and would have better explained the purpose of this study. With more participants for the study, the general population would have been better represented and the results may have been different.

Another limitation that affected the study was the amount of time the researchers were given to complete the study. A 27 month period was given to the researchers to complete the study; with more time, the researchers could have continued to generate a larger sample size to gather more data to further solidify the purpose of the study. The above listed limitations, kept the study from reaching its potential.

Future Research

The results obtained from the survey highlight interesting and useful information for the area of medicine. Finding that gender affected whether a participant was more aware of genetic testing while also finding that gender and whether you had a family history affects and participants knowledge level about genetic testing/counseling can guide medical professionals when educating their patients about the topic. Also, finding that protecting themselves was the most common answer for reasons for wanting to know about having a possible inheritable disease gives medical professionals insight into what might motivate their patients when it comes

to getting tested. Although this study needs to be expanded on further by incorporating a larger more diverse population to justify these findings, it provides a starting point for another study.

A study by Mai et al., found that as of 2010, less than 50% percent of the United States adult population knew about genetic testing (Mai et al., 2014). Although there are many differences in terms of the type of study conducted and the population surveyed when comparing the study done by Mai et al. and this study, it shows that awareness and knowledge towards genetic testing has increased in recent years. In the study done by Mai et al., as of 2010, 47% of the population (which was significantly larger than the population of this study), as compared to 82.7% of the population of this study, were aware of the availability of genetic testing as a resource to discover possible inheritable diseases (Mai et al., 2014). This is a small example of how much more aware participants are about genetic testing while also showing that there is room for improvement.

Although all of the studies used in the literature review differed from the outline of this study in terms of research questions, type of survey, population, etc., there are many differences and similarities to draw on. When future studies are conducted, the researchers can continue to compare and contrast studies to see what changes have or have not been made.

By expanding and continuing this study, the information can be used not only in genetics, but in other areas of medicine such as a family practice setting, for medical professionals to educate and guide their patients in their medical care. The topics of genetic testing and possibly inheriting a life changing disease can be difficult ones to encounter on for a medical professional, but with more statistical evidence a medical professional can feel more comfortable when approaching their patients. The continuation of studies such as this one is vital in terms of helping patients identify possible self and family altering inheritable diseases.

Conclusion

The purpose of this study was to assess the knowledge about genetic testing and participant's feelings towards finding out about a possible inheritable disease. A total of 156 surveys were completed by individual's aged 22-70 from three separate locations in the state of Minnesota. Using SPSS, a chi-square test and an ANOVA test were used to answer the first two research questions, respectfully, while descriptive statistics were used to answer the last two research questions. Statistically significant findings (obtaining a p value less than 0.05) were, gender (p value = .011) affects whether you are more aware of genetic testing and gender (p value = .046) and whether you had a family history (p value = .039) affects your knowledge level about genetic testing/counseling. It was also determined that wanting to protect themselves was the most answered reason for why participants would want to know if they had an inheritable disease. Limitations of this study were the targeted populations, sample size of each population, time to complete the study, and the significant difference between males and females of the participants who completed the survey. Thus, future studies can expand on this study by building on the current format, building on the results, and addressing the limitations that kept the study from reaching its full potential. This study, along with other current and future studies, will continue to impact the medical community by making positive advancements in the area of genetics that will allow participants to not only protect their families, but themselves as well.

REFERENCES

Bentley, D. (2000). The Human Genome Project - An Overview. *Medicinal Research Review,* 20(3), 189-196. Doi: 10.1002/(SICI)1098-1128(200005)20:3<189::AID-MED2>3.0.CO;2-#

Bosompra, K., Flynn, B., Ashikaga, T., Rairikar, C., Worden, J., & Solomon, L. (2000). Likelihood of Undergoing Genetic Testing for Cancer Risk: A Population-Based Study. Preventive Medicine, 30(2), 155-166.

Bruno, M., Digennaro, M., Tommasi, S., Stea, B., Danese, T., Schittulli, F., & Paradiso, A. (2010). Attitude towards genetic testing for breast cancer susceptibility: A comparison of affected and unaffected women. *European Journal of Cancer Care, 19(3),* 360-368. doi:10.1111/j.1365-2354.2009.01067.x

Budovsky, A., Tacutu, R., Yanai, H., Abramovich, A., Wolfson, M., & Fraifeld, V. (2008). Common gene signature of cancer and longevity. *Mechanisms of Ageing and Development, 130*, 33-39. doi: 10.1016/j.mad.2008.04.002

Etchegary, H. (2014). Public attitudes toward genetic risk testing and its role in healthcare. *Personalized Medicine, 11*(5), 509-522. doi:10.2217/pme.14.35

Genetic Counseling. (2014). Retrieved March 02, 2016, from http://www.healthcareers.umn.edu/health-careers/genetic-counseling

Genetic Counseling. (2015, March 03). Retrieved March 02, 2016, from http://www.cdc.gov/ncbddd/genetics/genetic_counseling.html

Genetic Testing. (2015, November 9). Retrieved from http://ghr.nlm.nih.gov/handbook/testing?show=all#riskslimitations

Henneman, L., Vermeulen, E., Van El, C. G., Claassen, L., Timmermans, D. R. M., & Cornel, M. C. (2013). Public attitudes towards genetic testing revisited: Comparing opinions between 2002 and 2010. *European Journal of Human Genetics, 21*(8), 793-799. doi:10.1038/ejhg.2012.271

Hurley, A. C., Harvey, R., Scott Roberts, J., Wilson-Chase, C., Lloyd, S., Prest, J., . . . Green, R. C. (2005). Genetic susceptibility for alzheimer's disease: Why did adult offspring seek testing? *American Journal of Alzheimer's Disease and Other Dementias, 20(6),* 374-381. doi:10.1086/502555

Lizardi, P., Forloni, M., & Wajapeyee, N. (2011). Genome-wide approaches for cancer gene discovery. *Trends in Biotechnology, 29*(11), 558-568. doi: 10.1016/j.tibtech.2011.06.003

MacConaill, L., & Garraway, L. (2010). Clinical Implications of the cancer genome. *Journal of Clinical Oncology*, 28(35), 5219-5228. doi: 10.1200/JCO.2009.27.4944

Mai, P., Vadaparampil, S., Breen, N., Mcneel, T., Wideroff, L., & Graubard, B. (2014). Awareness of Cancer Susceptibility Genetic Testing. American Journal of Preventive Medicine, 46(5), 440-448.

Meiser, B., Gleeson, M., Kasparian, N., Barlow-Stewart, K., Ryan, M., Watts, K., . . . Tucker, K. (2012). There is no decision to make: Experiences and attitudes toward treatment-focused genetic testing among women diagnosed with ovarian cancer.*Gynecologic Oncology, 124*(1), 153-157.

Ngoi, N., Lee, S., Hartman, M., Khin, L., & Wong, A. (2013). Interest and attitudes of patients, cancer physicians, medical students and cancer researchers towards a spectrum of genetic tests relevant to breast cancer patients. *The Breast, 22*(1), 47-52.

Pierce, B. A. (2012). *Genetics: A conceptual approach.* New York, NY: W. H. Freeman and Company.

Roy, A., Pallai, M., Lebwohl, B., Taylor, A. K., & Green, P. H. (2015). Attitudes toward genetic testing for celiac disease. *Journal of Genetic Counseling,* doi:10.1007/s10897-015-9867-z

Scuffham, T. M. & MacMillan, J. C. (2014). Huntington disease: Who seeks presymptomatic genetic testing, why and what are the outcomes? *Journal of Genetic Counseling, 23(5),* 754-761. doi:10.1007/s10897-013-9678-z

Scuffham, T. M., McInerny-Leo, A., Ng, S., & Mellick, G. (2014). Knowledge and attitudes towards genetic testing in those affected with parkinson's disease. *Journal of Community Genetics, 5(2),* 167-177. doi:10.1007/s12687-013-0168-7

What is genetic testing? (2015, November 9). Retrieved from http://ghr.nlm.nih.gov/handbook/testing/genetictesting

APPENDIX A

PERMISSION LETTERS

BETHEL UNIVERSITY

Drew Holm <drh62923@bethel.edu>

Fwd: New Message

Lisa Atkinson <lia69876@bethel.edu> Thu, Jun 16, 2016 at 10:27 AM
To: Drew Holm <drh62923@bethel.edu>

---------- Forwarded message ----------
From: <kndy@charter.net>
Date: Wed, Jun 15, 2016 at 10:11 PM
Subject: New Message
To: "lia69876@bethel.edu" <lia69876@bethel.edu>

- Dear Lisa Atkinson,

This letter is to confirm that I grant permission for Lisa Atkinson's research groups survey to be administer to the staff in their mailboxes at Bridgewater Elementary school on August 29th at 8:00am and receive surveys back on September 2nd at 4:00pm.

Sincerely,

Sheila Atkinson

MINNEAPOLIS
PUBLIC SCHOOLS
Urban Education. Global Citizens.

Lake Harriet Community School Lower Campus
4030 Chowen Avenue South
Minneapolis, Minnesota 55410-1096
Phone: 612-668-3210 Fax 612-668-3220

May 23, 2016

Dear Drew Holm

This letter is to confirm that I grant permission for Drew Holm to administer a survey to the staff at Lake Harriet Community School on August 23, 2016.

This is our opening meeting for the school year 2016-17 and is the first duty day for teachers. The meeting will take place at the Lower campus in the art room at approximately 8:00 a.m. with approximately 40 staff members in attendance.

Sincerely,

Merry Tilleson
Principal
Lake Harriet Community School
Lower Campus

BETHEL UNIVERSITY

Drew Holm <drh62923@bethel.edu>

Permission to hand out surveys

sandraholm@comcast.net <sandraholm@comcast.net> Sun, Jul 24, 2016 at 8:57 PM
To: drh62923@bethel.edu

This letter is to confirm that I grant permission for Drew Holm's research groups survey to be handed out to my patient's before their scheduled appointments at Dr. Michael Fashing Clinic on Monday's, Tuesday's, and Friday's starting on Monday August 22nd, 2016 and ending on Friday September 16th, 2016.

Sincerely,
Sandra Holm

APPENDIX B

REASEARCH SURVEY QUESTIONS

Inheritable disease – A disease that is passed on from parent to child

1. What is your age?
    a. 18-29
    b. 30-39
    c. 40-49
    d. 50-59
    e. ≥60
2. What is your gender?
    a. Male
    b. Female
3. What is your ethnicity?
    a. Non-Hispanic White
    b. Non-Hispanic Black
    c. Hispanic
    d. Asian
    e. American Indians/Alaskan Native
    f. Other
4. What is your highest education level?
    a. Less than high school graduate
    b. High school graduate
    c. Associate degree
    d. Bachelor's degree or higher
5. Do you, or your family, have a history of an inheritable disease?
    a. Yes
    b. No
    c. I don't know
6. Have you heard of genetic testing/counseling?
    a. Yes
    b. No
7. On a scale of 1-5 (1 being the least educated and 5 being the most educated), how knowledgeable are you about genetic testing/counseling?
    a. 1 – I know ***nothing*** about genetic testing/counseling
    b. 2 – I know a ***minimal amount*** about genetic testing/counseling
    c. 3 – I know a ***moderate*** amount educated about genetic testing/counseling
    d. 4 – I know ***a lot*** about genetic testing/counseling
    e. 5 – I am an ***expert*** on genetic testing/counseling

Flip over to complete questions 8 and 9

8. Would you rather know if you had an inheritable disease or would you rather not know about having an inheritable disease?
    a. I would want to know
    b. I would not want to know
9. If you answered yes to question 8, which of the following would be a reason for being genetically tested to determine if you have an inheritable disease? (circle all that apply)
    a. To protect myself
    b. To protect my children
    c. Peace of mind
    d. General curiosity
    e. Other

    _____

**********please return this questionnaire to the receptionist or nurse when completed**********

APPENDIX C

INFORMED CONSENT

Dear Participant,

You are invited to participate in a survey focused on how a variety of factors affect the level of knowledge genetic testing/counseling and the reasons for seeking out a possible inheritable disease. Your participation will help in the effort to educate and guide the public and professional health organizations on genetic testing. To participate, please complete the attached survey that was provided to you by Sandra Holm, Sheila Atkinson, or Merry Tilleson at Dr. Michael Fashing Clinic, Bridgewater Elementary School, or Lake Harriet Lower Community School. Your participation in this study is completely voluntary and there is no penalty by not participating. No participating will not affect your relationship with your affiliated elementary school or Fashing Clinic. To maintain anonymity, you will not identify yourself in any manner and all of the results of the study will be reported as a whole, as no specific details about any individuals will be included. This study is in partial fulfillment of the requirements for a Master's of Science Degree in Physician Assistant at Bethel University and was approved by the Bethel University IRB. Please contact us at the listed e-mail addresses below if you have any questions.

Thank you,

Lisa Atkinson PA-S, Drew Holm PA-S, Michael Jacob PA-S, Lisa Naser PA-C

lia69876@bethel.edu, drh62923@bethel.edu, michael-jacob@bethel.edu, l-naser@bethel.edu

APPENDIX D

SURVEY INSTRUCTIONS

Sheila Atkinson- Bridgewater Elementary School, Northfield, Minnesota

"This is a survey for a research project for Bethel Universities Physician Assistant Program. This survey is looking for your knowledge on the subject of genetic testing. Would you please fill out the survey to the best of your ability and knowledge without using any other sources. If you choose not to fill out the survey, it would have no bearing on your employment at this school. Please return the survey to my mailbox by September 2nd at 4pm. Thank you!"

Merry Tilleson- Lake Harriet Lower Community School, Minneapolis, Minnesota

"This is a survey for a research project for Bethel Universities Physician Assistant Program. This survey is looking for your knowledge on the subject of genetic testing. Would you please fill out the survey to the best of your ability and knowledge without using any other sources. If you choose not to fill out the survey, it would have no bearing on your employment at this school. Please return the survey to me before you leave the room, Thank you!"

Sandra Holm- Dr. Michael Fashing Clinic, Plymouth, Minnesota

"This is a survey for a research project for Bethel Universities Physician Assistant Program. This survey is looking for your knowledge on the subject of genetic testing. Would you please fill out the survey to the best of your ability and knowledge without using any other sources. If you choose not to fill out the survey, it would have no bearing on your affiliation or relationship with the clinic or its members. Please return the survey to me after you are finished, Thank you!"