AN UPDATED VIEW OF INDIVIDUALS' ATTITUDES, BELIEFS, AND CONCERNS REGARDING PREDICTIVE TESTING FOR HUNTINGTON'S DISEASE

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of Master of Science in Genetic Counseling

By
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CERTIFICATION OF APPROVAL

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DEDICATION

"School before boys."

- Ricky Farrar

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ABSTRACT

Huntington's Disease (HD) is a slowly progressive neurodegenerative disease. Individuals at risk for HD may present prior to the onset of symptoms seeking predictive testing in order to determine their gene status. Previous research has explored at-risk individuals' motivations for pursing predictive testing and its psychological consequences. However, the majority of this research predates 2010. Additionally, after literature review, there are currently no studies exploring the relationship between genetic law literacy (i.e. familiarity with the Health Information Portability and Accountability Act (HIPAA) and/or the Genetic Information Non-Discrimination Act (GINA)) and the decision to undergo predictive testing. The purpose of this study was to re-evaluate the at-risk testing population to better understand who is testing and to evaluate their attitudes, beliefs, and concerns about predictive testing. This study also explored the at-risk testing populations' genetic law literacy and how that may potentially influence testing decisions. This study revealed a similar socio-demographic profile to previous research, and motivations for testing were also consistent with those previously described. This study also demonstrated that the best predictors for one's testing decision are familiarity with GINA and/or HIPAA and higher education level. While only a subset of survey participants were offered anonymous testing, survey participants found the option to be of high importance. Lastly, there were mixed preferences, both in favor and not, for the [genetic] counseling component of the current predictive testing protocol.

CHAPTER I

INTRODUCTION

Background

Huntington's Disease (HD) is a slowly progressive neurodegenerative disease that currently does not have a known cure. HD is inherited in an autosomal dominant fashion, which means that there is a 50% chance that an affected individual can pass on the HD causing mutation to their offspring (Andersson et al., 2013). Affecting about 4-8 in 100,000 individuals in most European countries (Harper, 1992), HD is classically characterized by a triad of symptoms: motor manifestations, psychiatric changes and cognitive decline. While mood and cognitive changes often precede motor findings in what is often considered a "prodromal phase", a clinical diagnosis of HD requires the presence of motor features, such as chorea, found on examination (Walker, 2007).

HD is a trinucleotide repeat disorder caused by a CAG expansion of the Huntingtin gene located on chromosome 4p16.3. This leads to polyglutamine fragments that then aggregate in the cytoplasm and nucleus causing a toxic gain of function (Trottier et al., 1995). A normal allele codes for 26 or fewer CAG repeats, whereas, an expanded allele codes for CAG repeats of 40 and above. HD is a fully penetrant disease and therefore individuals with a CAG repeat of 40 and above will develop symptoms of HD during his or her lifetime. Expansions within the 27-35 range are considered "intermediate alleles" and symptoms typically do not occur.

Alternatively, expansions within the 36-39 range are referred to as "reduced penetrance alleles" and symptoms may or may not occur. Individuals with CAG expansions within intermediate or reduced penetrance ranges have alleles that are considered mutable, in that they are meiotically unstable, and may result in an expanded CAG repeat for their offspring. This is particularly true for affected males, as CAG repeats are more likely to expand when passed on through men leading to the possibility of anticipation (Semaka et al., 2013). Generally, the number of CAG repeats can be used to roughly predict the onset of disease. There is an inverse correlation between the number of CAG repeats and the approximate age of onset; the greater the number of CAG repeats, the earlier the age of onset (Walker, 2007, Lee et al., 2012). The median age of onset is 40 years (Braude et al., 1998).

Genetic testing programs for HD began in 1987 with linkage analysis and evolved into direct gene testing in 1993 (Braude et al., 1998). For individuals at risk for HD there are four options of genetic testing currently available: diagnostic or confirmatory testing, predictive or presymptomatic testing, prenatal testing, and preimplantation genetic diagnosis (PGD). While all tests can determine the number of CAG repeats present, the timing in which the testing can be administered is what differentiates these types of testing options. Confirmatory testing, in which an individual presents with clinical features suggestive of HD, is the most frequently encountered type of testing and is diagnostically supportive. Genetic testing is not required for diagnosis in the setting of a family history and clinical features suggestive of HD; however, patients will often undergo confirmatory genetic testing

to support the diagnosis and aid in genetic counseling issues. Alternatively, individuals at risk for HD, through a positive family history, may present prior to the onset of symptoms seeking predictive or presymptomatic testing in order to determine their gene status. This type of testing is currently available to individuals 18 years or older and there has been a standardized predictive testing protocol established to guide testing centers (MacLeod et al., 2013). The last two options of testing, prenatal testing and PGD, are reproductive testing options and are infrequently sought out or requested by individuals at risk. Prenatal testing can be done during a pregnancy through invasive procedures such as a chorionic villus sampling or amniocentesis, which results in genetic testing of the fetus. Alternatively, PGD is done prior to a pregnancy in order to select embryos without the HD mutation, and pregnancy can then be achieved via *in vitro* fertilization (MacLeod et al., 2013). While each of these testing options present emotional challenges for individuals and their families, predictive testing has been the focus of much attention in HD research literature, specifically regarding the reasons for testing and the psychological implications of discovering one's gene status prior to the development of symptoms.

The predictive testing protocol typically involves several clinic visits, including at least one genetic counseling visit and one psychological/psychiatric assessment. Due to the varying protocols across testing sites, the exact number of visits can vary depending on where testing is taking place (MacLeod et al., 2013). Typically, information about the genetics and the clinical spectrum of HD is provided during these visits. Medical providers explore individuals' motivations for testing

followed by a discussion regarding the possible consequences of test results. Patients may also be informed of genetic privacy laws, such as the Health Information Portability and Accountability Act (HIPAA) and/or the Genetic Information Non-Discrimination Act (GINA) during these visits. Additionally, while most centers administer testing through insurance, some centers may offer anonymous testing with an out-of-pocket cost. This option offers individuals a layer of protection from the stigma associated with such a devastating disease and associated fears of discrimination. While test results should be delivered in person, per suggested protocol, patients have the option to cancel or delay the testing process at any time (Bernhardt et al., 2009).

Due to the sensitivity of genetic information, it is important for patients to understand HIPAA and GINA. Of note, HIPAA aims to do the following: (1) protect the privacy of patients' health information, (2) provide for the physical and electronic security of Protected Health Information (PHI), and (3) specify the right of patients to approve access/use of their information (Robles, 2015). Additionally, in 2008, GINA was signed into federal law and is designed to (1) forbid employers from denying employment, promotions, or health insurance coverage to people when genetic tests show they have a predisposition to an inherited disease, and (2) prevent health insurance companies from using genetic test information to determine an applicant's risk profile. GINA, does not, however, apply to life, disability, and long-term care insurance. Similarly, GINA does not protect individuals in the military and veterans seeking care through the Veterans Affair or Indian Health Service (Green et al.,

2015). Lastly, these proposed health provisions are not in effect once the individual has symptoms of the inherited disease.

Prior to direct mutation testing becoming clinically available, it was reported that between 40-60% of at-risk individuals expressed interest in pursuing predictive testing (Bernhardt et al., 2009). However, the actual uptake was lower than expected since becoming clinically available. Currently, it is estimated that only 5-20% of individuals at-risk for HD undergo predictive testing (Evers-Kiebooms and Decruyenaere, 1998). Due to the emotional and psychological implications of predictive testing, it is not surprising that only a small percentage of at-risk individuals choose seek this information. Testing uptake appears to be influenced by demographics, personality, and personal coping style (Evers-Kiebooms and Decruyenaere, 1998). Collectively, it appears that those who pursue testing tend to be young adult females, which is thought to be due to a female's greater willingness to face difficult health concerns and because of their significant involvement in reproduction and their mothering role (Meiser and Dunn, 2000). Previous studies have shown that individuals may forgo testing due to the lack of treatment and cure for HD, they are simply happier not knowing, they have concerns for their family or spouse, implications for life insurance, and/or the testing process itself can be a barrier for some as well (Evers-Kiebooms and Decruyenaere, 1998). Therefore, there are both emotional and logistical concerns that can either reaffirm and dissuade testing decisions.

Understandably, at-risk individuals test for many different reasons, and the decision to do so is a deeply personal one. Research has shown that individuals claim the following motivations for testing: uncertainty in life, family planning, and the need to inform their children about their risk. Other motivations include the need to plan relationships, finances, educational goals and career choices (Decruyenaere et al., 2004). Even though predictive testing has been available for over two decades, research has shown that motivations for testing appear to have remained fairly consistent.

Regardless of the reason for pursuing testing, the task of deciding to proceed with predictive testing is a complex and emotional process. The process of deciding to test usually occurs over time and in response to life events (Taylor, 2005). While the decision to test should be a personal decision free of coercion, some individuals may feel obligated to test for their children in order to clarify their risk. They may even feel that it is the "right thing to do," even if they do not want to learn of that information for themselves (Smith et al., 2013). For others, the experience of living at-risk can be enough of a deciding factor to pursue or forgo predictive testing. Once someone's gene status is discovered, it is not possible to un-know that information, and this type of certainty, assuming a positive result, can destroy any hope for an HD-free life. However, some individuals prefer the certainty of knowing, as it can help them move forward with their lives now knowing what the future actually holds. It is of note that those who do not test in order to preserve hope are not in denial and it is

by no means a passive decision; they are aware of what HD entails and simply choose not to let their hope subside (Quaid et al., 2008).

Specific Aims and Purpose of the Study

The University of California (UC) Davis Huntington's Disease Center of Excellence has provided care for the HD community since 1997. At this facility, patients are offered anonymous predictive genetic testing in which they come to the clinic under a pseudonym for genetic counseling and testing. Over the years there has been an observed shift in individuals seeking testing with a noticeable increase in the number patients coming to the clinic for predictive testing and who are of a younger generation. However, it is unclear if this is true for the rest of the at-risk population as well. With this in mind, it was important to reach out to the HD community to learn more about their attitudes and beliefs regarding predictive genetic testing in order to provide an updated understanding of who is testing and why.

The key purposes of this study are:

1. *To re-evaluate the patient population to determine who is currently seeking predictive testing, either anonymously or under their true identity.*
2. *To re-analyze these patients' demographic information to better characterize the current at-risk patient population.*

3. *To better understand their attitudes, beliefs and concerns regarding predictive testing, as well as genetic law literacy, in order to evaluate the shifting views with time.*

CHAPTER II

METHODS

This study is based on an anonymous survey that was initially offered at the 29th Annual Huntington's Disease Society of America (HDSA) convention. The convention took place on June 20th-22nd, 2014 in Louisville, Kentucky. The survey was also subsequently made available via the following:

1. Links to the survey on the UC Davis Medical Center, Center of Excellence HD Clinic website and Facebook page.
2. Postcards with the online survey link at local support group meetings and in the UC Davis Medical Center, Center of Excellence HD Clinic.
3. At regional HD conferences, both on paper and online, similar to the 2014 HDSA convention in Louisville, Kentucky.
4. With the permission of HDSA, a link to the online survey was made available on their website.

The inclusion criteria for participants in this study included:

1. Asymptomatic individuals at risk for HD who have *not* undergone predictive genetic testing.
2. Asymptomatic individuals at risk for HD who *have* undergone predictive genetic testing.
3. Individuals who are 16 years of age and above.

The exclusion criteria for participants in this study included:

1. Individuals who are 15 years of age and below.

Note that adults who may be unable to consent, pregnant women, and prisoners may be included in this research, however they were not specifically targeted.

Survey participants who were offered either a paper survey or a link to the survey via Survey Monkey® had 90 days to complete it. Individuals who completed the paper survey were given the option to return the completed survey in person or via mail addressed to the HDSA center at the UC Davis Medical Center. The survey was estimated to take approximately 10-15 minutes, and included both multiple choice and open-ended questions. The number of questions asked depended on if the survey participant had undergone predictive testing or not. For those who had not undergone predictive testing, there were 31 questions, whereas for those who did undergo predictive testing, there were 52 questions. A copy of the survey is included in the Appendix.

The survey was done on a voluntary basis and subjects were allowed to end the survey at any time. There was no identifying information obtained from survey participants. There was minimal risk to subjects and there was no direct benefit to the subjects. The survey participants were not compensated for their participation. Although the results of this study will not be shared with the participants directly, the data has been presented to the HD

community at the Huntington Study Group annual meeting and the plan is to submit these findings for publication.

All study data is stored and was analyzed on a password-protected computer. Descriptive statistics (percentages, means, standard deviations) were used for all sociodemographic data. Statistical analyses for the quantitative components of this survey include Fisher's exact test, Kendall's tau b, and a multivariate log linear analysis. For all analyses, a critical *p*-value of <.05 was used. Statisticians at UC Davis and California State University (CSU), Stanislaus helped conduct and review all statistical analyses. Data from the survey was exported into Microsoft Excel documents and was analyzed in both Microsoft Excel and Statistical Package for the Social Sciences (SPSS).

Open-ended questions with written responses were also organized into themes based responses based on their conceptual similarities via the content analysis method. After each theme and sub-theme was identified and named by the principal investigator, the co-principal investigator then reviewed the themes and sub-themes as well. All coding was done manually and organized in Microsoft Excel documents.

The University International Review Board (IRB) via CSU Stanislaus approved this thesis, #1516-029, on September 10, 2015. IRB approval via UC Davis was also obtained in 2014 (Protocol #612727).

CHAPTER III

RESULTS AND DISCUSSION

Demographics

There were a total of 284 survey respondents. Of these, 108 (38%) had completed the predictive testing process, whereas, 176 (62%) had not been tested. Of those who had not tested, 149 (52%) had decidedly not tested compared to 27 (10%) who remained undecided on whether or not they want to test. Of those who tested (n= 108), the average age was 36 years old (SD= 11), 86 (80%) were female, and additionally, 106 (98%) reported Caucasian ancestry. Of those who have not undergone predictive testing, (n= 176), the average age was 35 years old (SD= 12), 150 (85%) were female, and 168 (99%) reported Caucasian ancestry. Of note, 6 participants were 18 years or younger. Select demographics of this sample population are consistent with Bernhardt et al.'s 2009 paper; the sample population is predominantly young and female. Furthermore, in this study, of those who tested, 74% are employed, 62% are in a relationship, and 59% have children. Not surprisingly, a significant number of those who tested have health insurance (98%) and 69% have life insurance. However, of those who tested, 72% do not have disability insurance, and 78% do not have long-term care insurance. A summary of the survey participants' testing decisions and demographic information can be found in Figure 1 and Table 1, respectively, and their geographic regions can be found in Figure 2.

Figure 1
Summary of survey participants' testing decision

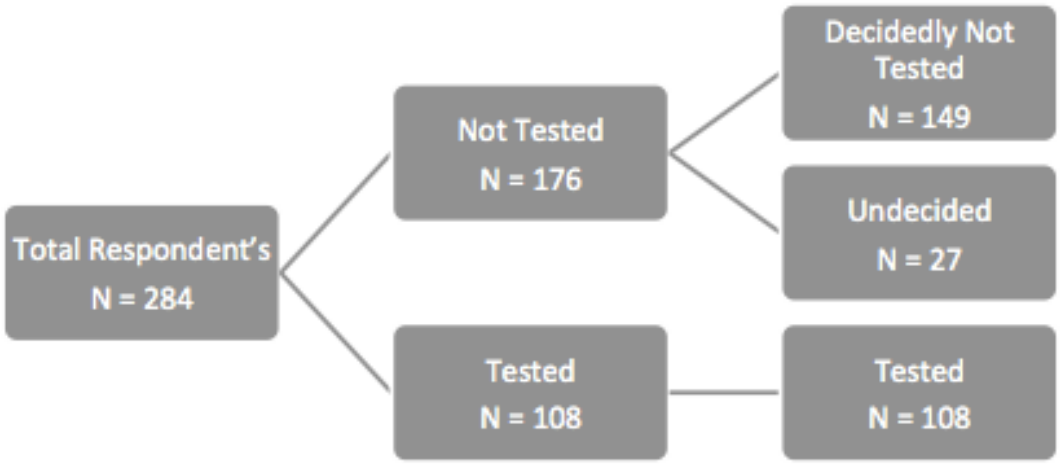


Table 1

Social and Demographic Characteristics of Survey Participants

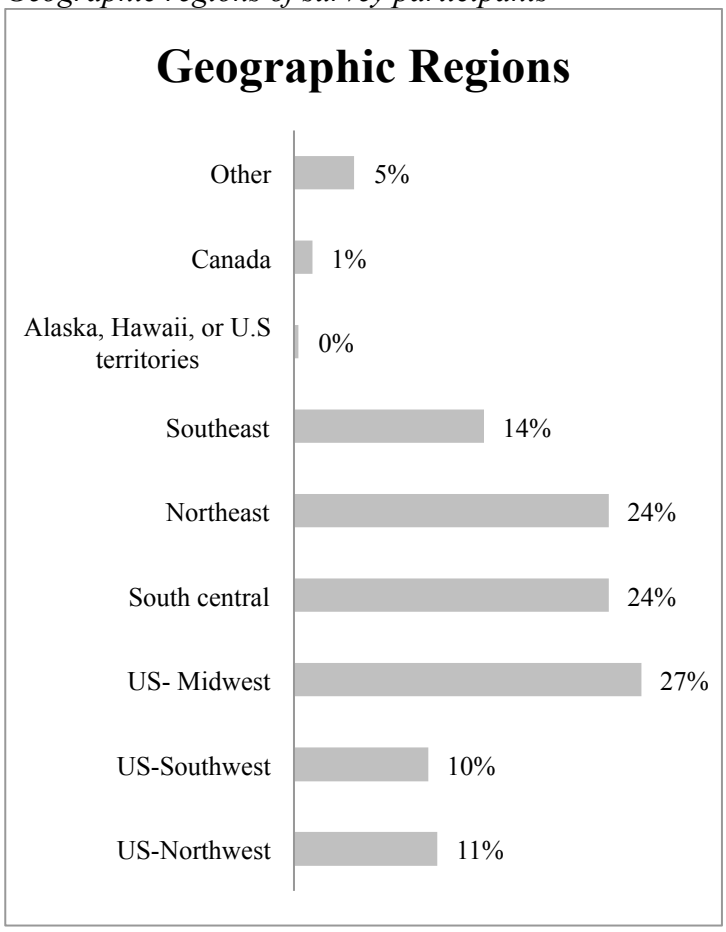
Demographics	*Yes, Tested*	*No, Not Tested**	*p-Value*
Total number (n)	*108*	*176*	
Age, y, mean (SD)	*36 (11)*	*35 (12)*	
Female, n, (%)	*86 (80)*	*150 (85)*	*0.324*
Caucasian, n (%)	*106 (98)*	*168 (99)*	*0.32*
Employment, n (%)			*0.42*
Student	*6 (6)*	*5 (3)*	
Working	*80 (74)*	*119 (69)*	
Unemployed or disability	*22 (20)*	*48 (28)*	
Marital status			*0.29*
*Single***	*39 (36)*	*79 (45)*	
Married/With Partner	*67 (62)*	*93 (53)*	
Widowed	*2 (2)*	*2 (1)*	
Children, n (%)			*0.31*
Yes	*64 (59)*	*93 (53)*	
No	*44 (41)*	*82 (47)*	
Affected parent, n (%)			***0.02***
Mother	*56 (56)*	*81 (57)*	
Father	*44 (44)*	*60 (43)*	
Insurance status, n (%)			
Health insurance			*0.56*
Yes	*98 (91)*	*156 (89)*	
No	*10 (9)*	*20 (11)*	
Life insurance			***0.0003***
Yes	*74 (69)*	*82 (47)*	
No	*34 (31)*	*94 (53)*	
Disability			*0.16*
Yes	*30 (38)*	*36 (20)*	
No	*78 (72)*	*140 (80)*	
Long term care insurance			***0.002***
Yes	*24 (22)*	*16 (9)*	
No	*84 (78)*	*160 (91)*	

**"No, not tested" includes individuals who have actively decided not to test, and those who are currently undecided*

***Single also includes those who are separated or divorced*

Bold p-values are significant

Figure 2
Geographic regions of survey participants



Factors for the testing decision – Gender

Fisher's exact test was performed to determine whether or not there is a significant association between gender and the testing decision. Although the majority of the survey participants were female, the testing rate for females was slightly lower than for men, 40.8% and 48.9%, respectively. However, while Meiser and Dunn reported in 2000 that those who test are more likely to be female, in this study, gender was not a statistically significant variable as to one's decision to test or not (p = .324).

Factors for the the testing decision – Education

Survey participants' completed level of education was obtained. Education was classified into four categories, (1) no college (including less than a 12th grade education), (2), some college (including an Associates' degree), (3) Bachelor's degree, and (4) Graduate degree. Thirty four percent (*n*= 37) of those who tested had completed some college, 29% (*n*= 32) had a Bachelor's degree, 23% (*n*= 25) had a Graduate degree, 12% (*n*= 13) have no college experience, and one participant reported an unknown level of education. The same trend was observed for survey participants who did not test. Forty percent (*n*= 61) had completed some college, 22% (*n*= 34) had a Bachelor's degree, 18% (*n*= 27) had a Graduate degree, and 18% (*n*= 27) had no college experience. Due to education being an ordinal variable, Kendall's tau-b was performed to determine whether or not there is a significant association between one's level of education and their decision to test or not. In support of what Meiser and Dunn stated in 2000, this analysis demonstrated that an individual is more likely to test with additional education (Kendall's tau-b = 0.11, *p* = .054). The testing rate for those with no college was 32.5%, 37.8% with some college experience, 48.5% with a Bachelor's degree, and 41.8% with a graduate degree. Additionally, Fisher's exact test was performed to determine if there is an association between having any type of degree and not having a degree and one's testing decision. The analysis was of borderline significance; the testing rate was 48.3% for survey participants with degrees and 36.2% for those without degrees (Fisher's exact

test, $p = .057$). A summary of education level and one's testing decision can be found in Table 2.

Table 2
Testing Decision and Level of Education

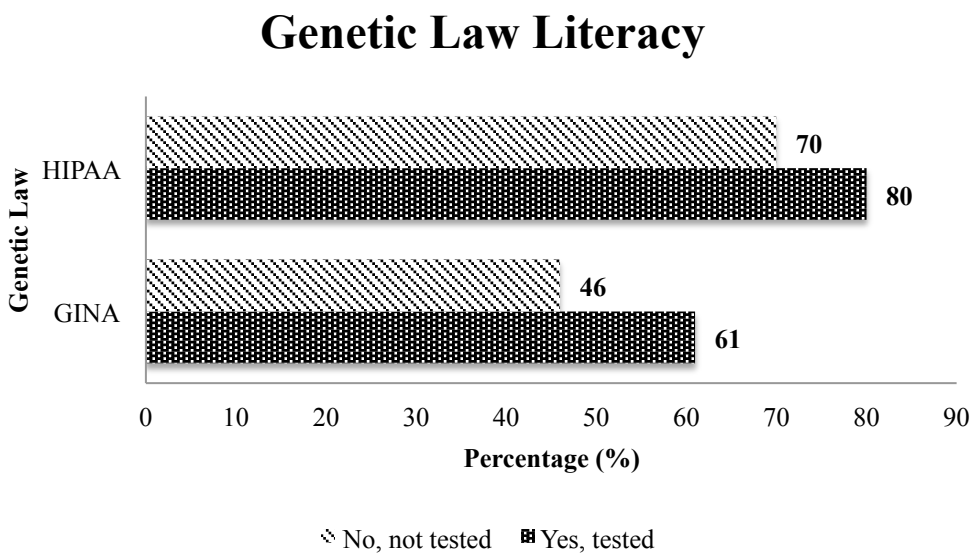
Education Level	*Yes, Tested*	*No, Not Tested**
No college, n (%)	*13 (12)*	*27 (18)*
Some college, n (%)	*37 (34)*	*61 (40)*
Bachelor's degree, n (%)	*32 (29)*	*34 (22)*
Graduate degree, n (%)	*25 (23)*	*27 (18)*
Unknown, n (%)	*1 (.009)*	*0 (0)*

**"No, Not tested" includes individuals who have actively decided not to test, and those who are currently undecided*

Factors for the the testing decision – Genetic law literacy

To assess survey participants' genetic law literacy, they were asked if they were familiar with HIPAA and GINA. They were given the following answer choices: yes, no, unsure, or other. For statistical analyses, "unsure" answer choices were combined with "no", and "other" was not used, as these were reported only one to two times per testing group. Of those who tested, more individuals were familiar with HIPAA than they were with GINA. Eighty percent ($n = 86$) were familiar with HIPAA, whereas 20% ($n = 22$) were not. Additionally, 61% ($n = 65$) were familiar with GINA, whereas 39% ($n = 42$) were not. For individuals who had not tested, 70% ($n = 103$) were familiar with HIPAA, whereas 30% ($n = 45$) were not. Lastly, 46% ($n = 67$) were familiar with GINA, whereas 54% ($n = 80$) were not. A summary of genetic law literacy and one's testing decision can be found in Figure 3.

Figure 3
Summary of genetic law literacy familiarity



**"No, Not tested" includes individuals who have actively decided not to test, and those who are currently undecided*
"Yes, Tested", GINA, *n*= 1 reported "other"
"No, Not Tested", HIPAA, *n*= 1 reported "other"
"No, Not Tested", GINA, *n*= 2 reported "other"

Fisher's exact test was performed to see if there is an association between familiarity with genetic privacy laws and one's testing decision. This analysis demonstrated borderline significance regarding knowledge of HIPAA. The testing rate for survey participants who are familiar with HIPAA was 45.5%, while it was only 30.6% for survey participants who were not familiar or of unsure about HIPAA (Fisher's exact test, $p = .074$). Conversely, although fewer individuals were familiar with GINA than with HIPAA, knowledge of GINA proved to be of significance. The testing rate for survey participants who were familiar with GINA was 49.2%, while it was only 33.0% for survey participants who were not familiar or unsure about GINA (Fisher's exact test, $p = .016$). Lastly, Fisher's exact test also demonstrated that there

is a strong correlation between knowledge of HIPAA and knowledge of GINA (Fisher's exact test, $p < .001$). This suggests that someone who is familiar with one genetic law is likely to be familiar with the other.

Multivariate Analysis – Genetic law literacy, education and testing decision

To determine the relationship between multiple categorical variables, such as genetic law literacy and education level, a log linear analysis was performed. Although a correlation between knowledge of HIPAA and knowledge of GINA has been demonstrated as shown above, a log linear analysis demonstrated that there is no interaction between an individuals' familiarity with HIPAA verses their familiarity with GINA (likelihood ratio = 0.002, df = 1, $p = .967$). While the data has shown that if someone is more likely to test if they are familiar with GINA, the likelihood of them testing is unaffected if they are also familiar with HIPAA (likelihood ratio = 3.543, df = 2, $p = .170$). Similarly, if an individual is familiar with HIPAA, they are no more likely to test if they are also familiar with GINA (likelihood ratio = 1.531, df = 2, $p = .465$). This suggests that one's testing decision is affected by knowledge of HIPAA or GINA, but not necessarily both (although previous analyses indicate only familiarity with GINA is significant).

As noted previously, the data suggests that both genetic law literacy and education are predictors for pursuing predictive testing. As familiarity with GINA proved significant, whereas familiarity with HIPAA did not, a log linear analysis regarding education and familiarity with GINA was also performed. Due to this

specific analysis' requirements and limitations, education was treated as a nominal variable. Additionally, although there were originally four categories of education, for this particular analysis the two lowest and two highest levels of education were combined, since the biggest difference in testing appears to be due to either having a degree or not. As this is considered an exploratory method of analysis, the findings described as follows are tentative.

The log linear analysis demonstrated that there is no interaction between education and GINA familiarity (likelihood ratio = 0.066, df = 1, p = .797). Because there is no evidence for an interaction, if it is assumed that testing likelihood varies with GINA familiarity, there is no need to assume it also varies with education (Likelihood ratio = 4.773, df = 2, p = .092). Similarly, if you assume that the testing likelihood varies with education, there is no need to assume it also varies with GINA familiarity (Likelihood ratio = 3.379, df = 2, p = .185). This is true because GINA familiarity is correlated with education (Fisher's exact test, p = .032), however, there is no interaction.

In summary, the data suggests that people who are familiar with HIPAA also tend to be familiar with GINA. Additionally, people who are more highly educated are also more likely to be familiar with GINA and HIPAA. Therefore in order to predict one's likelihood of pursuing predictive testing, assessing their familiarity with HIPAA, GINA, or their education level, would be sufficient predictors. Contrary to previous literature, in this study, gender does not appear to be a predictor. A

summary of all statistical analyses can be found in Table 3, and a summary of testing rates per variable can be found in Table 4.

Table 3
Summary of Statistical Analyses

Statistical Analysis	Variables	*P*-value*
Fisher's exact test	Gender, testing decision	.324
Kendall's tau b	Education, testing decision	***.054***
Fisher's exact test	HIPAA familiarity, testing decision	.074
Fisher's exact test	GINA familiarity, testing decision	***.016***
Fisher's exact test	HIPAA and GINA familiarity	***<.001***
Log linear analysis	HIPAA and GINA familiarity	.967
Log linear analysis	GINA familiarity, influence of HIPAA familiarity on testing decision	.170
Log linear analysis	HIPAA familiarity, influence of GINA familiarity on testing decision	.465
Fisher's exact test	Degree or no degree, testing decision	***.057***
Log linear analysis	Education, GINA familiarity	.797
Log linear analysis	Education, influence of GINA familiarity on testing decision	.185
Log linear analysis	GINA familiarity, influence of education on testing decision	.092

**Where p <.05 is significant*
Bold and italicized p-values are either significant or borderline significant

Table 4
Summary of testing rate per variable

Variables	Testing rate (%)	P-value*
HIPAA familiarity vs. testing decision	45.5% - Yes, familiar 30.6% - No, not familiar	.074
GINA familiarity vs. testing decision	49.2% - Yes, familiar 33.0% - No, not familiar	*.016*
Gender vs. testing decision	40.8% - Female 48.9% - Male	.324
Education vs. testing decision	32.5% - No college 37.8% - Some college 48.5% - Bachelor's degree 41.8% - Graduate degree	.054

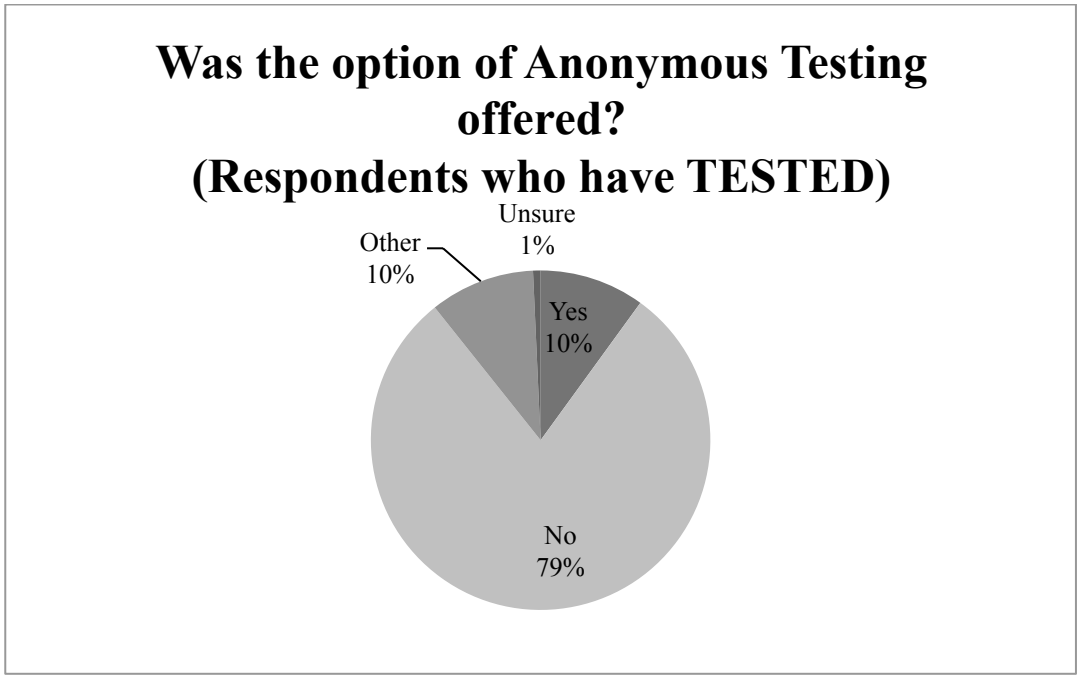
**Where p <.05 is significant*

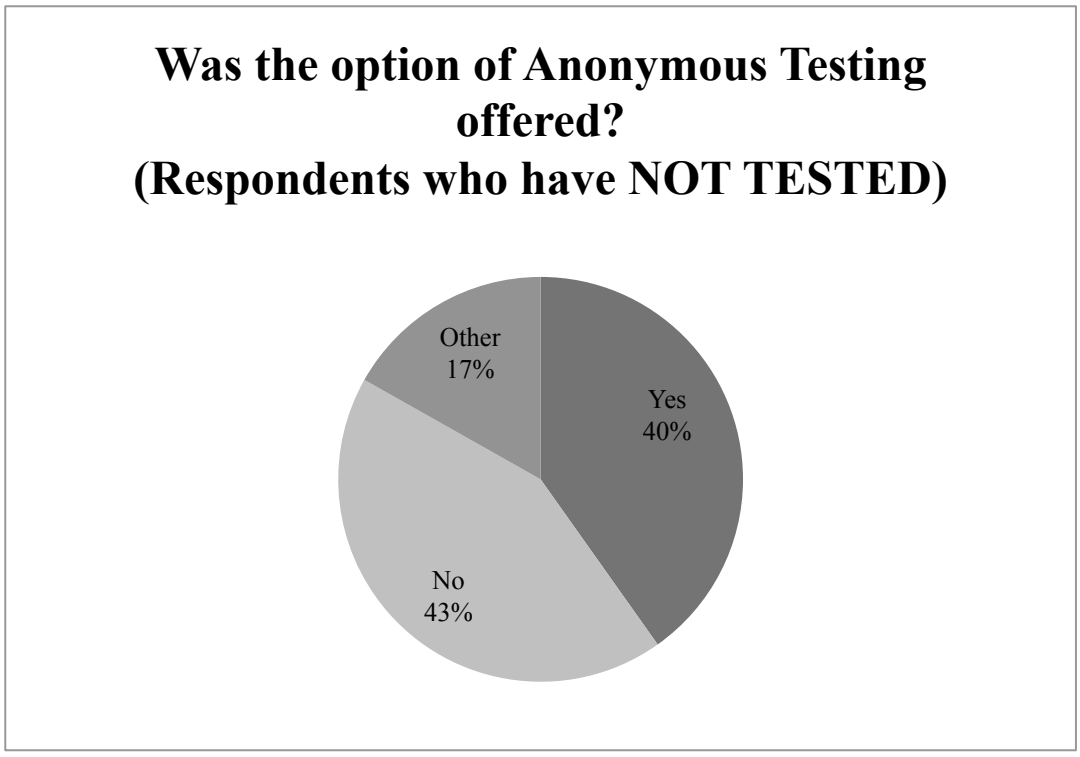
Opinions about and values of anonymous testing

Survey participants were asked if anonymous testing was made available to them, and if it had not been, then would that have changed their testing decision if it had been an option. Of those who pursued predictive testing, only 10% (*n*=14) had been offered anonymous testing, 79% (*n*=111) were not, 10% (*n*=14) reported "other" and 1% (*n*=1) was unsure. For those who ultimately did not undergo predictive testing, 40% (*n*=43) were offered anonymous testing, 43% (*n*=46) were not, and 17% (*n*=18) reported "other". A summary of these findings can be found in Figure 4.

Figure 4

Anonymous testing offering to those tested verses those not tested



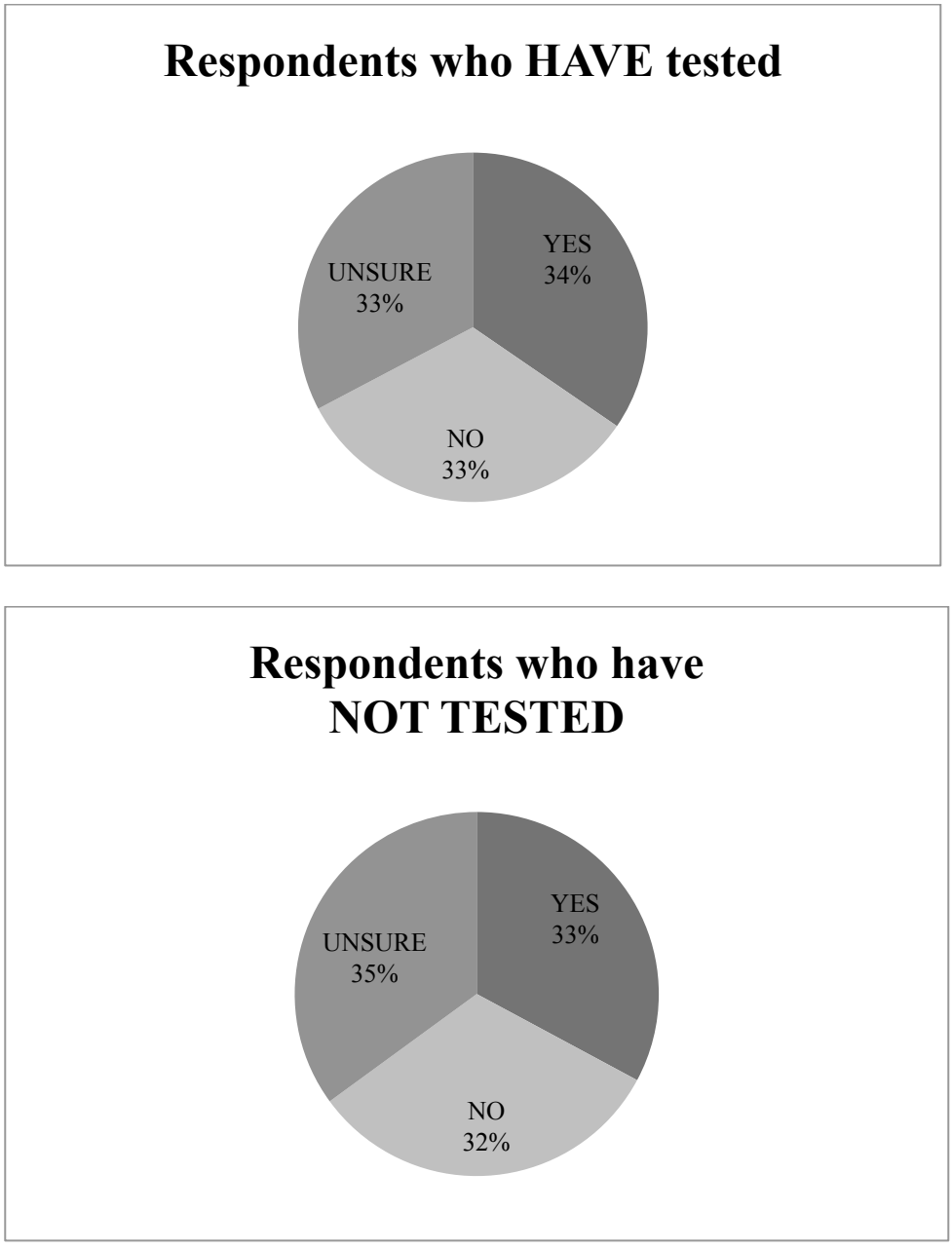


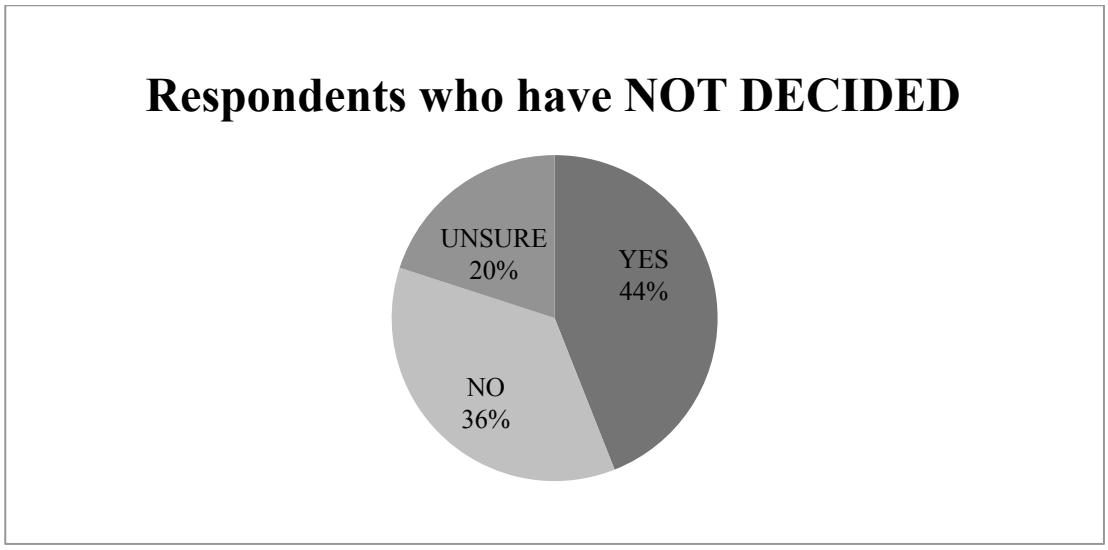
Survey participants were then asked if the option of anonymous testing would have changed their final decision regarding testing. Of those who tested, 35% (*n*=56) stated it would have changed their decision, 33% (*n*=53) reported it would not have changed their decision, and 33% (*n*=53) were unsure. The trend was similar to those who have decided not to test. Thirty-three percent (*n*=45) stated it would have changed their decision, 32% (*n*=44) reported it would not have changed their decision, and 35% (*n*=48) were unsure. However, for those who were still undecided about whether or not to test, a slightly greater number of 44% (*n*=11) stated the availability of anonymous testing would have changed their decision, 36% (*n*=9) said it would not, and 20% (*n*=5) were still unsure. A summary of these findings can be found in Figure 5.

Following the question above, survey participants were then asked to explain their answer choice. The following themes were identified: (1) testing anonymously would negate insurance concerns, (2) more knowledge about what anonymous testing is needed, (3) still would not pursue testing even if anonymous testing was offered, and (4) no perceived difference/benefit between anonymous testing and non-anonymous testing. Of note, some individuals did not appear to understand what anonymous testing entailed, with some stating that they thought it meant that the results would not be disclosed to anyone, including him or herself.

Figure 5

Summary of the influence of anonymous testing availability on testing decision





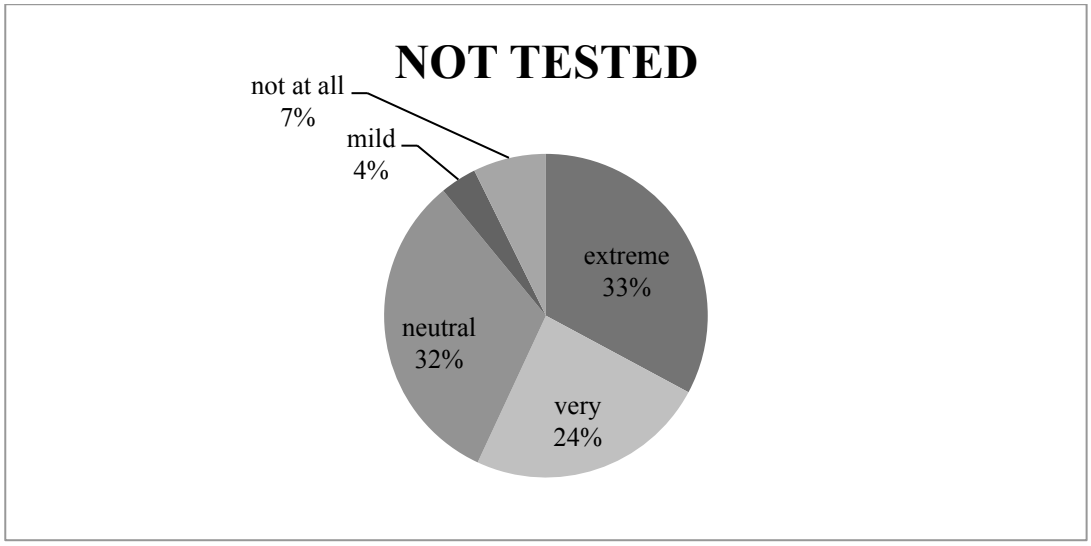
Additionally, survey participants were asked how important the option of anonymous testing was to them. The answer choices were (1) extremely important, (2) very important, (3) neutral, (4) mildly important, and (5) not at all important. Of those who tested, 40% (*n*=42) found the option of anonymous testing extremely important, 21% (*n*=22) did not find it important at all, 20% (*n*=21) found it to be of neutral importance, 12% (*n*=13) found it very important, and 8% (*n*=8) found it mildly important. For those who have not yet tested, 33% (*n*=45) found it extremely important, 7% (*n*=10) did not find it important at all, 32% (*n*=44) found it to be of neutral importance, 24% (*n*=33) found it very important, and 4% (*n*=5) found it mildly important. Lastly, of those who have not yet decided to test or not, 35% (*n*=9) found it very important, 31% (*n*=8) found it to be neutral importance, 15% (*n*=4) found it extremely important, 15% (*n*=4) also did not find it important at all, and only 4% (*n*=1) found it to be mildly important. These results highlight, that across each testing group, approximately 50% of individuals felt the option of anonymous testing

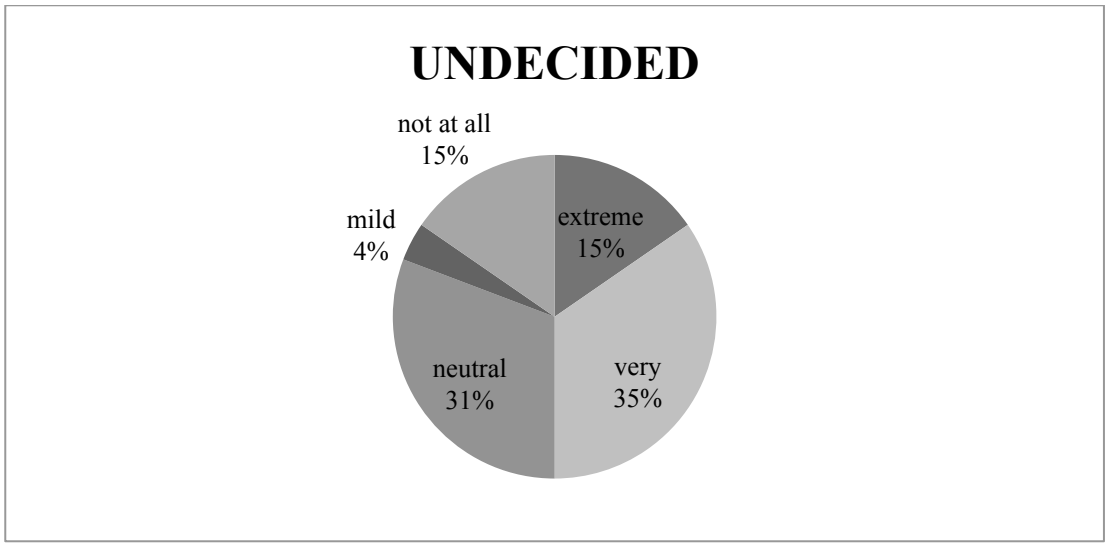
was either extremely or very important. A summary of these findings can be found in Figure 6.

Lastly, individuals who pursued predictive testing were also asked why they chose the identification they chose (true identity or anonymous testing). The following themes were identified: (1) overall concern, (2) overall unawareness, and (3) lack of necessity [for anonymous testing]. Sub-themes for each theme were also identified, and are presented in Figure 7.

Figure 6

Importance of the availability of anonymous testing per testing decision




UNDECIDED
not at all
15%
extreme
15%
mild
4%
neutral
31%
very
35%

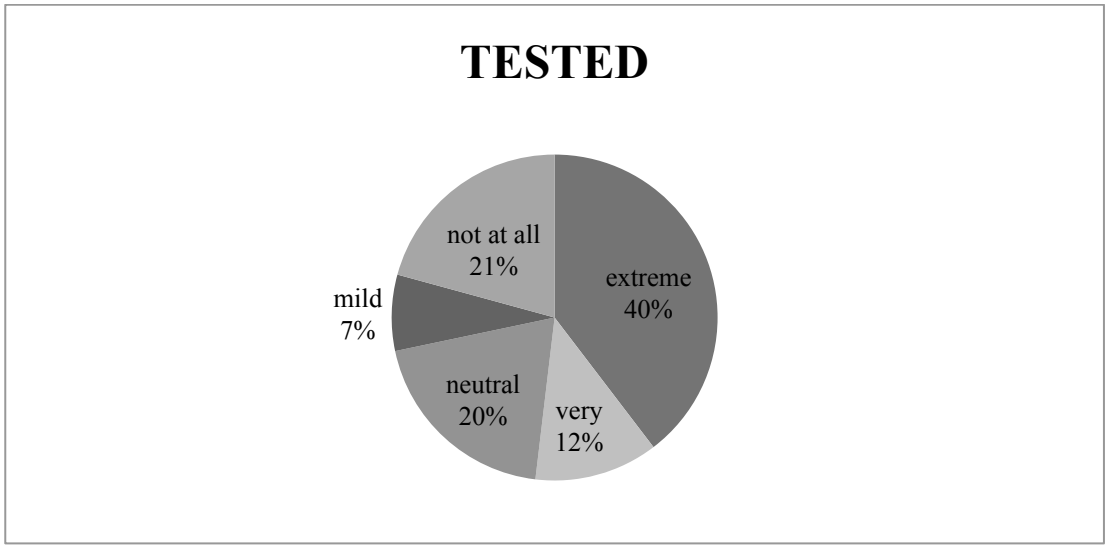
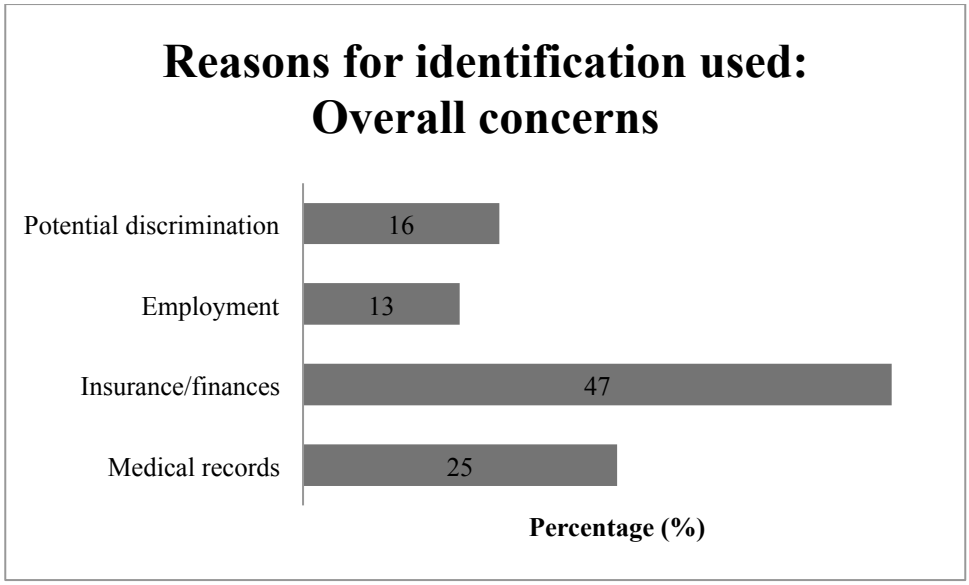
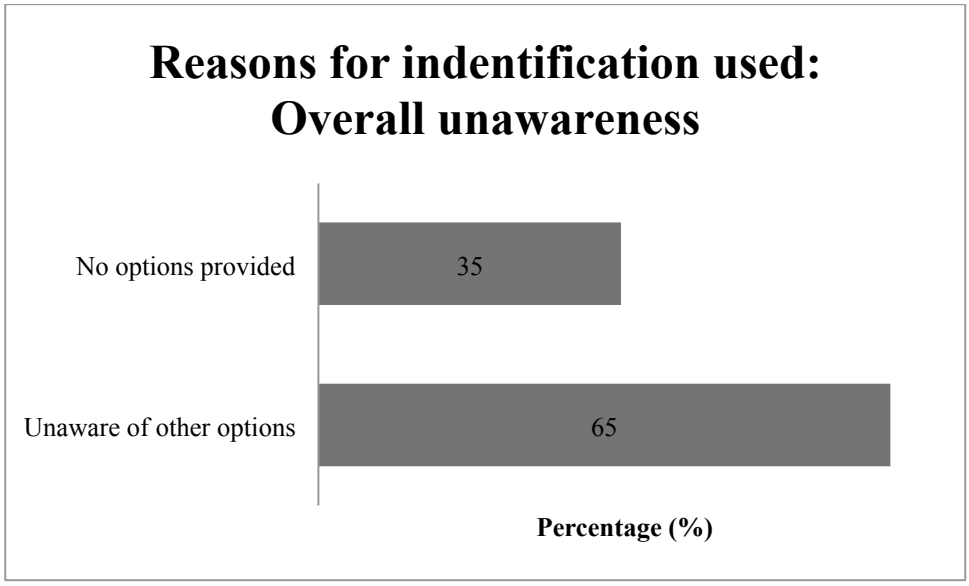
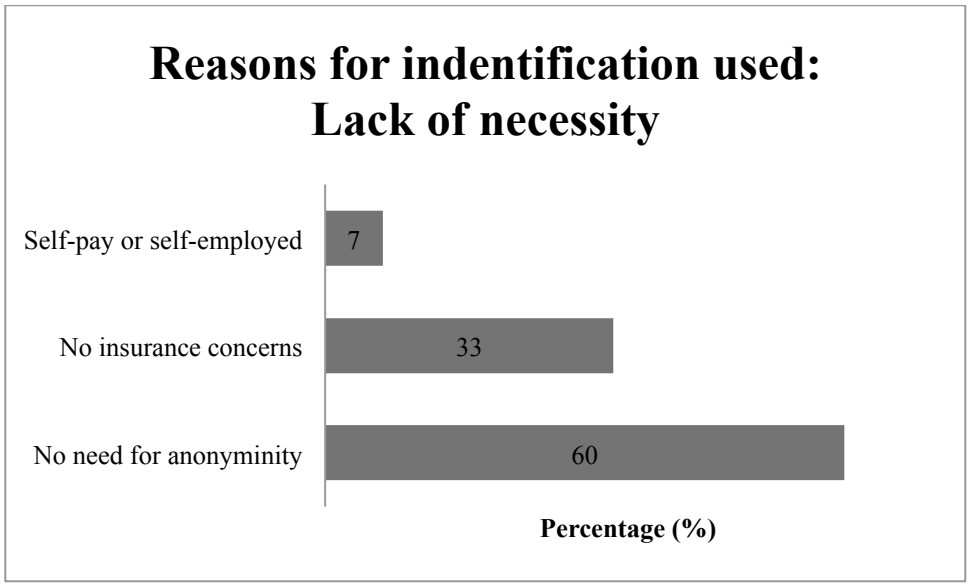

TESTED
not at all
21%
extreme
40%
mild
7%
neutral
20%
very
12%

Figure 7

Summary of identification used for predictive testing







It appears that a significant number of individuals continue to have concerns regarding employment, insurance and finances, privacy and the potential of genetic discrimination. Part of this may be due to the fact that only 10% of the testing population were offered anonymous testing. This would suggest that these individuals would have likely benefited from the option of anonymous testing and potentially from an explanation and discussion of both genetic laws, HIPAA and GINA. However, as these individuals still pursued predictive testing regardless of testing anonymously, it suggests that the benefit (learning their gene status) outweighed the perceived cost (potential discrimination), thus highlighting the high value of genetic information to at-risk individuals.

Additionally, as shown in Figure 5, 65% of individuals were unaware of other testing identification options. However, 60% claimed no need for anonymity. These responses demonstrate that each individual considering testing brings his or her own

motivations, concerns and needs that should be explored further to ensure that they are receiving the best care throughout the testing process.

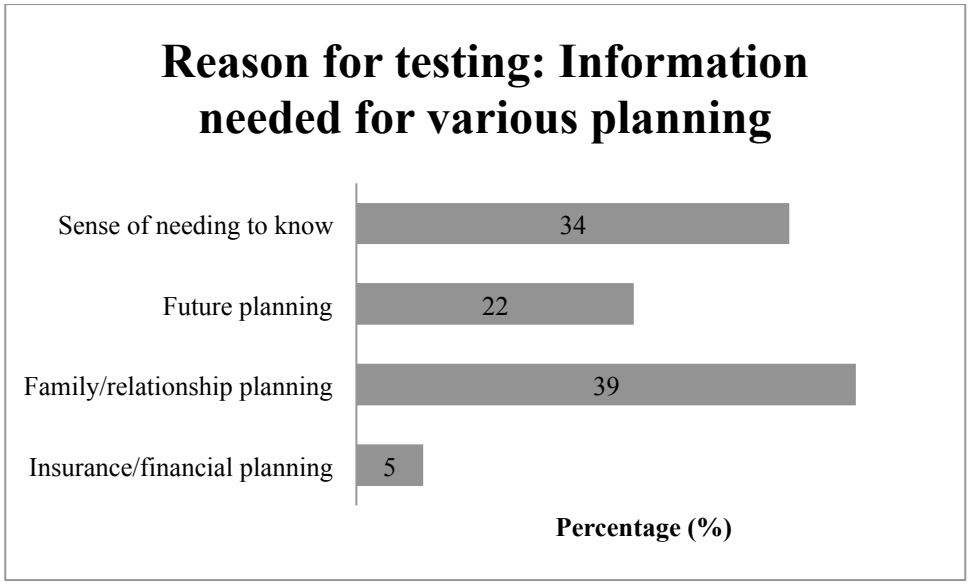
In summary, the data suggests that individuals believe anonymous testing would negate [insurance] concerns, despite the implementation of GINA, and that more individuals would potentially pursue testing if the option of anonymous testing was made available to them. However, there is some suggestion that some individuals are uncertain about what anonymous testing actually is. This would imply that medical providers offering predictive testing need to clarify what anonymous testing entails, if available at that center, so patients can make informed decisions. Testing centers may also wish to consider offering anonymous testing if they are not already, as it appears to be of high importance to at-risk individuals, especially those who have not yet decided if they want to test or not. While only 10% of those who did ultimately test were offered anonymous testing, 40% of them found the option to be extremely important.

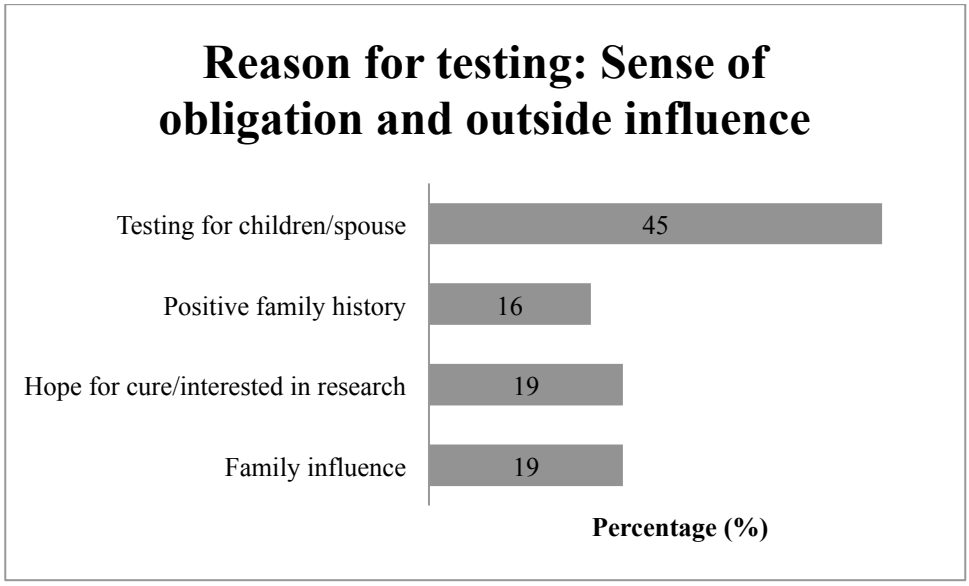
Reasons for and for not pursing predictive testing

Open-ended questions were asked to those who tested and those who did not test to better understand their reasons for doing so. For those who did pursue predictive testing, the following themes were identified when asked why, (1) information needed for various planning, and (2) sense of obligation and outside influence. Sub-themes for each theme were also identified, and are presented in Figure 8.

Figure 8

Summary of reasons for pursuing predictive testing





As expected from previous research (Codori et al. 1994, Decruyenaere et al., 2004), planning for various aspects in one's life was a central motivation in the

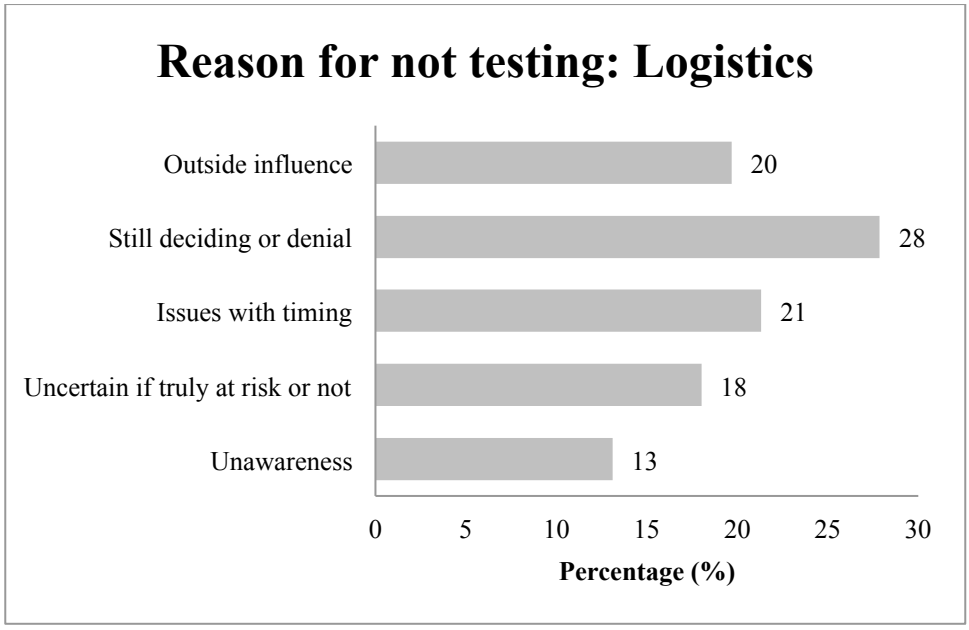
decision making process. Regarding testing motivations, 39% reported planning for one's family and romantic relationships, followed by 22% that reported future planning, whereas only 5% reported insurance and/or financial planning. Additionally, 34% indicated simply "needing to know." This demonstrates that although some individuals use their gene status to help plan for various parts in their lives, others seek knowledge driven by need to know for their own peace of mind.

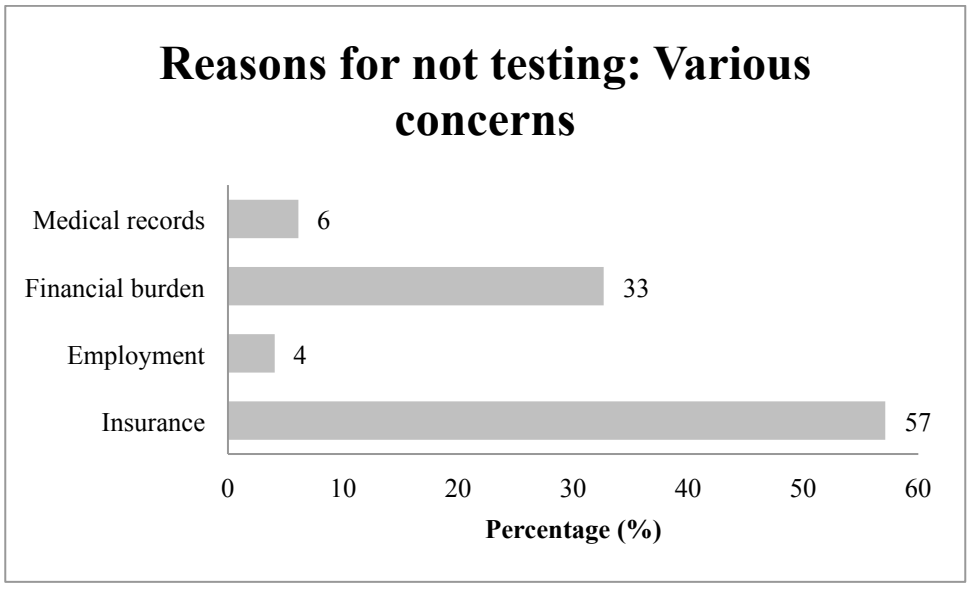
Other individuals are motivated to test out of a sense of obligation to their family members. This has also been previously reported (Smith et al., 2013). This study shows that 45% of individuals reported that they were either testing for their children or their spouse because they felt a moral obligation to do so. Additionally, 19% felt influenced by other family members to test. Of note, one individual reported that they were influenced by their family to test so that it would help them better understand what their personal role is in their family, likely to help clarify if they would either need to be a caretaker or have a caretaker themselves. About 19% of individuals were influenced to test by the HD community as a whole. Lastly, a hope for a cure and the desire to participate in research were also motivating factors.

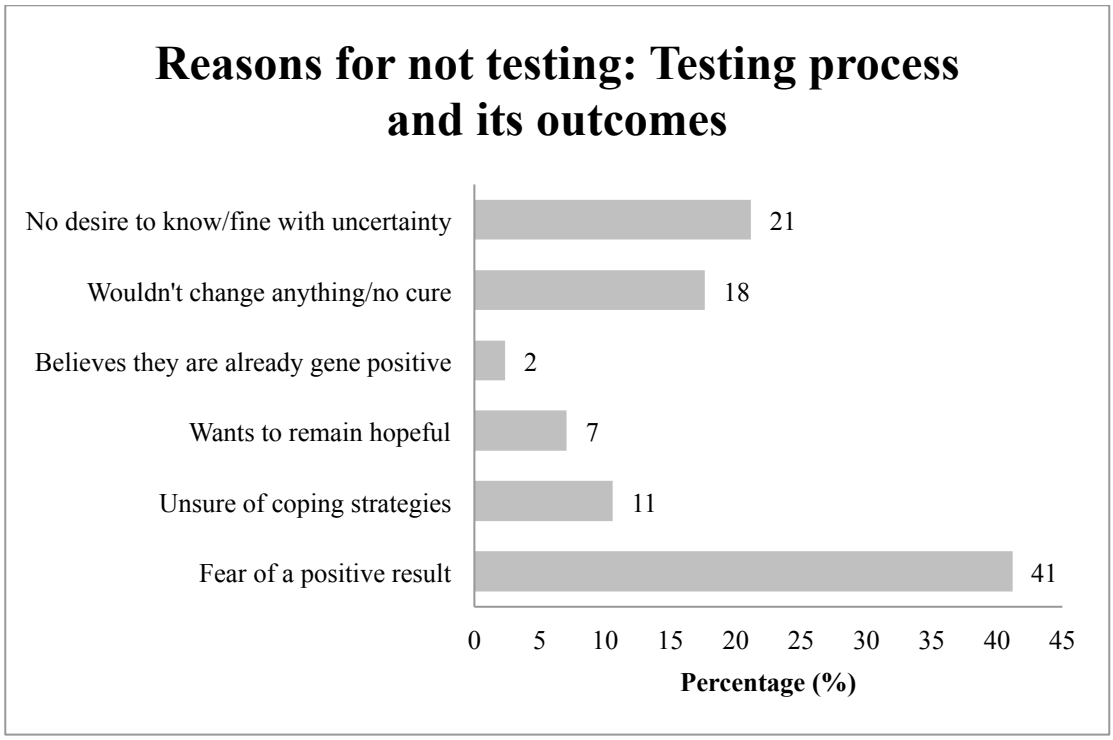
For those who decided not to pursue predictive testing, the following themes were identified when asked why, (1) various concerns, (2) logistics, and (3) the testing process and its outcomes. Sub-themes for each theme were also identified, and are presented in Figure 9.

Figure 9

Summary of reasons for not pursuing predictive testing







It is interesting to note that there is still a significant degree of concern regarding insurance, employment, and privacy despite HIPAA and the implementation of GINA in 2008. Employment concerns included both the fear of losing an existing job and the fear of employment biases. Certainly the creation of GINA was meant to alleviate majority of these concerns (namely health insurability and employment), yet these concerns are still evident. This would suggest that either patients who chose not to test were either unaware of HIPAA and/or GINA, were aware and did not understand its protections, or were aware but still had lingering concerns or mistrust of the law, their insurance provider and/or employers.

Additionally, it appears that various logistical aspects of testing are also barriers for testing for some individuals, which is consistent with previous literature

(Evers-Kiebooms and Decruyenaere, 1998). As shown in Figure 8, 20% of individuals expressed feeling pressured to test either by their physician or family. Other individuals were still in the process of deciding and/or were in denial. This highlights the weight of the decision to test, both personally and within a family. This also reiterates that the decision to test, as previous literature has suggested (Taylor, 2005), is not an easy and quick decision. There also appears to be issues with timing, including the lack of free time to test given the number of visits required, being too young an age to test, and it simply not being the right time in their lives to test. This demonstrates an understanding of the magnitude of information that this test can bring.

It is interesting to note that 18% of individuals remain uncertain if they are truly at risk for HD or not. It is possible that there is a known family history however, their parent might not yet have demonstrated any clinical symptoms and/or they themselves have not opted for confirmatory or predictive testing. It is also possible that the individual no longer has contact with the affected relative and does not know with certainty if they were diagnosed with HD, or even that their parent passed away before HD manifested. When it is unclear if testing for HD is even indicated, these uncertainties can undoubtedly complicate the testing process and decision of whether or not to test. Lastly, there also appears to be some general unawareness of the testing process. Figure 8 shows that 13% of individuals reported that they were either unaware of where to go to obtain testing, or were even unaware that testing for HD was even possible.

For those who did not test due to the overall testing process and its outcomes, 41% stated that they feared a positive result compared to 21% who stated that they were comfortable with the uncertainty of not knowing and therefore had no desire to know. This exemplifies that for some individuals fear can be crippling and uncertainty can be comforting, while for others fear can be a motivator and uncertainty can be daunting. Some individuals also had a "gut feeling" prior to testing and believed they were either positive or negative, which could be either motivating or deterring regarding their testing decision. Lastly, 18% stated that testing would not change anything for them because the results would not affect their future decisions or there is currently no known cure.

Opinions regarding the overall testing process

Survey participants who had completed the predictive testing process were asked about their experience in regards to the frequency of visits, how satisfied they were with the overall process and what follow-up care they received. When asked about the frequency of visits, 77% (n=81) of individuals stated that the number of visits was adequate, 15% (n=15) thought there were too few visits, 6% (n=6) reported "other", and only 2% (n=2) reported there were too many visits. While most "other" comments were due to the individual simply not remembering the number of visits, or not caring about the number of visits required, select "other" explanations of interest are quoted below:

> *"I was fully aware of HD and what it entailed, so I was not required to have any appts prior to my testing."*

"I was traveling a long distance so, they permitted me to do less pre-visits."

While majority of the survey participants who tested stated that the number of visits required was adequate, it would appear that various testing centers have different requirements. In regards to how satisfied survey participants were with the overall testing process, 60% (*n*=64) of the individuals was very satisfied, 31% (*n*=33) were somewhat satisfied, 8% (*n*=9) reported "neutral", and 1% (*n*=1) was somewhat dissatisfied. No one reported that they were very dissatisfied. A summary of these findings can be found in Figure 10.

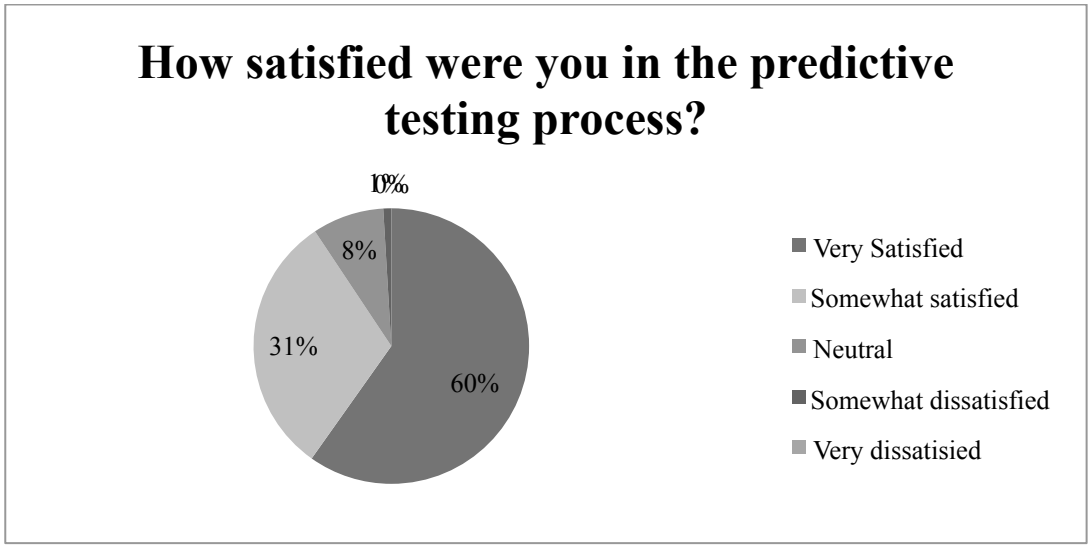
Lastly, the majority of survey participants reported that no follow-up was received. However, other answers included a phone call, letters, a neurology appointment, a genetic counseling/psychiatrist/psychologist visit, and that follow-up was offered but not accepted. These answers further highlight the differing clinical approaches used during the testing process. This data demonstrates the difference in testing protocols across various centers, and suggests that follow-up may only given on a case-by-case basis, as needed with a specific provider. Below are two examples of the striking differences between one's opinions of the testing process:

> *"I think counseling and support is necessary to deal with the news. My sibling, 2 years after the test results elected to stop eating. Death occurred in 62 days from starvation."*

> *"I am not up on any of the newest testing protocols. I was very offended before when I was told I had to jump through all sorts of hoops to learn I carried the HD gene. If that hasn't changed, then I have no interest in being tested."*

Figure 10

Satisfaction with the predictive testing process



When asked about any issues encountered during the testing process that they felt could be done differently or better, 66% of the responses reflected on the logistics of the process and the specific care that was or was not provided. Regarding the logistics of testing, some individuals felt that they needed more follow-up care and the option of pre-test and post-test counseling. This suggests that patients may need more assistance regarding the decision making process, and more opportunities to process the results afterwards with an experienced healthcare professional.

Some individuals also reported that they wish there was more encouragement about receiving [genetic/psychological] counseling. This suggests that some patients may need an further support to pursue counseling or therapy, or that there may even be a stigma with receiving this type of care. Conversely, other individuals did not

want genetic counseling at all and wanted to encounter fewer providers during the testing process. The spectrum of opinions regarding the testing process highlights the varied needs of individuals seeking testing and necessitates an individualized approach to patient care throughout this process. While predictive testing protocols exist to guide healthcare providers through patient education and potential implications of testing, those seeking testing need to be treated as individuals, and may thus require an individualized approach in order to broaden the accessibility of testing.

With regards to the care that was ultimately received during the testing process, respondents shared a wish that their healthcare provider were more knowledgeable about HD. Additionally, individuals asked that their healthcare provider was more empathetic to their current situation and that they be non-directive throughout their decision making process. This could suggest that not all individuals are interacting with genetic counselors for pre-test counseling, as genetic counselors are typically trained to be non-directive (Pencarinha et al., 1992), although this certainly does not ensure that a genetic counselor would counsel this way. For example, general healthcare providers who may not be familiar with HD and/or the motivations for testing, may default to a paternalistic approach that may not be as appreciated in this arena, as other medical recommendations might be. These specific requests support the use of medical centers that specialize in caring for HD patients. These centers often offer a multidisciplinary care approach to patient care and they

are experienced with the predictive testing process. It is here that patients can be offered the appropriate counseling, education, and support.

Regarding the logistics of the testing process, at-risk individuals desire a cheaper test with a shorter turn around time, and more convenience with scheduling the many appointments required. These suggestions highlight the sense of urgency at-risk patients have about learning their gene status; they want to be seen right away and get results as soon as possible. It appears that at-risk individuals would like their results to be more easily obtained and more accessible to them as much as possible. Noteworthy quotes from these open-ended responses can be found in Table 5.

Table 5

Noteworthy quotes regarding the testing process

"Post follow up is important with a therapist or counselor since the results take years to sink in."
"I think the protocol of advance counseling is very important. Structure and a supportive process is important to blunt the shock- even if you are negative. I could have used more counseling to prepare me for the family issues that unfolded from the mixed results among siblings."
"I believe everyone needs to go about the testing process differently. Everyone has different needs and views so the genetic counseling process should be flexible to who they are working with."
"People need to be treated better this is already such an emotional situation."

CHAPTER IV

CONCLUSION

Summary of Findings

The purpose of this study was to re-evaluate the at-risk testing population for HD to better understand who is testing and what their attitudes, beliefs, and concerns about predictive testing are. Additionally, the study also explored the at-risk testing populations' genetic law literacy, and how that may or may not influence their testing decisions.

Consistent with previous research (Trembath et al., 2006, Dufrasne et al., 2011, and Sizer et al., 2012), this study revealed that those who pursued predictive testing are on average 36 years old and 98% reported Caucasian ancestry. However, this study was 80% female compared to previously reports ranging from 57.6-66.7% (Dufrasne et al., 2011, Sizer et al., 2012, and Trembath et al., 2006).

Reasons for and against testing were also found to be consistent with previous literature (Codori et al. 1994, Decruyenaere et al., 2004). For those who tested, the following themes were identified: (1) information needed for various planning and (2) sense of obligation and outside influence. For those who did not test, the following themes were identified: (1) various concerns (for example financial and employment concerns), (2) logistics, and (3) the testing process and its outcomes.

Regarding the testing process, this study also showed that only 10% of those who tested were offered anonymous testing, however, 40% found the option to be

extremely important. Of note, about a third of those who tested, did not test, and are still undecided all find the option of anonymous testing influential of their overall decision to test. This study also shows that those who test would prefer easier scheduling, although the number of visits required does not appear to be that strong of a deterrent, faster turn around time for results, and a cheaper cost of testing. Despite these requests, majority of those who tested were reportedly very satisfied with their testing process.

Statistical analyses were performed to determine various associations between one's demographics and genetic law literacy and its relationship to their testing decision. This study showed that the higher the education level, the more likely they are to test, which was also suggested by Meiser and Dunn in 2000. Additionally, individuals who are more familiar with GINA are more likely to test, while familiarity with HIPAA is of borderline significance. While previous literature has shown that females are more likely to test (Bernhardt et al., 2009), and even though this study's population was predominantly female, statistical analyses showed that gender does not contribute to the likelihood of testing. The study also demonstrated that those who are familiar with HIPAA are also familiar with GINA, and those with higher educations are more likely to be familiar with genetic laws such as HIPAA and GINA. Therefore, according to this study, the best predictors for one's testing decision are their familiarity with GINA and/or HIPAA, and their education level.

Implications for Practice and Profession

Survey participants were asked to provide their feedback regarding the testing process and there was a mixture of those who favored pre-test and post-test counseling, and those who strongly opposed it. As it would be difficult to determine who would most benefit from such counseling, or who is in favor or opposed to such counseling based on a referral alone, perhaps the testing protocol could be better tailored to each patient. Genetic counseling could serve as an initial screen, among other things, to assess the patient's reason for and understanding of predictive testing, as well as what their current knowledge of HD is. From there, the genetic counselor can determine the adequate amount of counseling needed and can tailor their agenda based on the patient, and/or suggest additional counseling either with themselves or other healthcare providers only as needed. In other words, the genetic counselor can assess whether or not the patient should see, for example, a psychiatrist or psychologist, and the patient would not be required to see them if the genetic counselor does not think it is necessary.

Survey participants were also frustrated with the lack of knowledge of HD from various providers. It might be helpful for non-HD specialists, for example primary care physicians, to be required to make appropriate referrals to either neurologists or HD clinics when a patient requests testing instead of simply ordering the test for them. This would ensure that 1) the provider overseeing their care is knowledgeable in what the patient is being tested for, and 2) appropriate pre-test and follow-up care, e.g. counseling, is offered and/or available to the patient, as needed.

This would be particularly helpful for patients who would otherwise be ashamed in seeking out post-test counseling on their own, or even for those who do not know what post-test resources are available to them.

Lastly, many survey participants also disliked the lengthy turn around time for genetic testing results, which can be at a minimum approximately two weeks. While this reflects the sense of urgency patients have when testing, perhaps better clarification and explanation of the turn around time and testing process should be explained at some point prior to administering testing. While in certain circumstances test results can be expedited, turn around times are often dependent upon the lab and out of the healthcare provider's control, and the patient should be aware of this prior to testing in order to ensure overall satisfaction of the testing process.

CHAPTER V

LIMITATIONS AND FUTURE STUDIES

Limitations

This study had several limitations. While the sample size was large enough for sufficient statistical analyses, an n=284 is still small for generalizability for the entire at-risk population. Therefore larger sample sizes in future studies may help to even further characterize the at-risk population as a whole. There was also some missing data, in that some participants left some questions blank, and some questions were also answered incorrectly (e.g. their response did not address the question) which were not included in the analysis.

Additionally, for question number 29 on the paper survey, the goal was to have survey participants clarify their response for question number 28, but the question incorrectly referred to question number 27. Some participants realized what was being asked and answered correctly, however some participants clarified their response to the incorrect (although indicated) question. These responses were therefore not used. However, this question was later corrected in the online survey, and only a total of 10 responses were recorded via the paper survey with the error. This type of error illustrates the innate limitations of online surveys in that clarification is not readily available. Another innate error of online surveys and open-ended questions is that such questions are open for interpretation and answers may not necessarily address the intended question if clarification is unavailable.

Future Studies

In this study, only those who completed testing were asked about their opinions of the testing process. In future studies it would be insightful to learn the opinions of those who have yet to completed testing, or those who have chosen not to be tested, about the testing process. This type of information may be useful in determining any barriers to the testing process for those who do not test, and to determine if there are any predictors for not testing that are innate to the current testing process/protocol. More exploration of not just who did not test, but those are still in the process of deciding to test, could also provide useful feedback regarding the current testing protocol. Additionally, since a number of survey participants were disappointed with the lack of knowledge of HD from their healthcare provider, a study exploring the attitudes, beliefs and logistical outcomes of non-HD providers ordering HD predictive (or confirmatory) testing for their patients would be insightful.

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APPENDIX

APPENDIX A

HUNTINGTON'S DISEASE PREDICTIVE TESTING SURVEY

Are you or were you an individual at-risk for Huntington's disease and faced with a decision about undergoing genetic testing? If yes, we invite you to please take a moment to read our introduction letter and consider taking our anonymous survey. Please note that persons must be 16 and older to participate. Thank you.

Introduction:

We are reaching out to individuals who are considering or have already undergone predictive testing for Huntington's disease to complete a brief survey. This information will help us understand your opinion about anonymous predictive testing. In anonymous testing, the person is assigned a pseudonym such as "John Doe" or a numeric-letter code in order to protect their identity. We would like to know the following:

- *For those who have not undergone predictive testing, how would the option of anonymous testing influence your decision to test?*
- *For those who have already undergone predictive testing, were you offered the option of anonymous testing? If so, how important was that option to you?*

This survey includes questions about your age and background, your family history, your decision to undergo testing or not, and your experiences if you have tested. You may complete the survey on paper or by using the following link to an on-line version: http://www.surveymonkey.com/s/HDSA2014UCD1. *The survey will take 10-15 minutes to complete.*

We thank you for your participation and time.

Sincerely,
Alexandra Duffy, DO
Vicki Wheelock, MD
HDSA Center of Excellence at UC Davis

Questionnaire

1. **Are you age 16 years or older?**
   - ☐ Yes
   - ☐ No

   If you answered YES, please proceed to Question 2.

   If you answered NO, you are not eligible to complete this survey. Thank you for your interest in participation.

2. **If you are either age 16 or 17, you must have permission from a parent or legal guardian to participate in this survey. Do you have permission from a parent or legal guardian to participate in this survey?**
   - ☐ Yes
   - ☐ No

   If you answered YES, please proceed to Question 3.

   If you answered NO, you are not eligible to complete this survey. Thank you for your interest in participation.

3. **What is your age in years? __ __**

4. **Please indicate the geographic region in which you currently reside:**

United States regions:
- ☐ Northwest
- ☐ Southwest
- ☐ Midwest
- ☐ South central
- ☐ Northeast
- ☐ Southeast
- ☐ Alaska, Hawaii, or US territories

- ☐ Canada

☐ Other (please specify): _____

5. **What is your gender?**

☐ Male
☐ Female
☐ Other

6. **Race ("X" those with which you identify):**

☐ American Indian or Alaska Native
☐ Asian
☐ Black or African-American
☐ Native Hawaiian or Other Pacific Islander
☐ White
☐ Other

7. **Highest Level of Education:**

☐ <9th grade
☐ 9th to 12th grade, no diploma
☐ High school graduate, no degree
☐ Some college, no degree
☐ Associate degree
☐ Bachelor's degree
☐ Graduate or professional degree

8. **Employment:**

☐ Student
☐ Full-time work
☐ Part-time work
☐ Unemployed
☐ Applied for OR receiving disability benefits

9. **Marital status:**

☐ Single – Never married
☐ Single – Separated or Divorced
☐ Married
☐ Widowed
☐ With partner

10. **Please indicate the number of biological children you have: __ __**

11. **Do you plan to have more children?**

- ☐ Yes
- ☐ No
- ☐ Undetermined

12. **Is your mother affected by HD?**

- ☐ Yes
- ☐ No
- ☐ Unknown

13. **If yes, mother's age of symptom onset (in years): __ __**

14. **Is your father affected by HD?**

- ☐ Yes
- ☐ No
- ☐ Unknown

15. **If yes, father's age of symptom onset (in years): __ __**

16. **Is your grandmother on your affected parent's side affected by HD?**

- ☐ Yes
- ☐ No
- ☐ Unknown

17. **If your grandfather on your affected parent's side affected by HD?**

- ☐ Yes
- ☐ No
- ☐ Unknown

18. **How many siblings do you have? __ __**

19. **How many siblings do you have who are affected by HD? __ __**

20. **How many children do you have who are affected by HD? __ __**

21. **Insurance status: Which of the following types of insurance do you have? (You may choose multiple answers)**

- ☐ Health insurance

- ☐ Life insurance
- ☐ Disability insurance
- ☐ Long term care insurance
- ☐ Other (please specify): _____

22. **When applying for insurance, did any questions or items specifically mention Huntington's disease?**

- ☐ Yes
- ☐ No
- ☐ Unsure
- ☐ Not applicable

23. **Are you familiar with the Health Insurance Portability and Accountability Act (HIPAA) which protects health information under a set of national standards?**

- ☐ Yes
- ☐ No
- ☐ Unsure
- ☐ Other (please elaborate): _____

24. **Are you familiar with the Genetic Information Nondiscrimination Act of 2008 (GINA) which is a federal law that protects all Americans and prohibits discrimination in health coverage and employment based on genetic information?**

- ☐ Yes
- ☐ No
- ☐ Unsure
- ☐ Other (please elaborate): _____

25. **Testing status: Have you completed predictive testing for HD?**

- ☐ Yes
- ☐ No
- ☐ Not decided yet

If you have not completed the predictive testing process please answer questions 25 – 30 to complete your survey.

If you have completed the predictive testing process please skip to question 31 and continue from there.

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26. **If NO or not decided, please explain why.**

27. Were you offered anonymous predictive testing?

☐ Yes
☐ No
☐ Unsure
☐ Other (please specify): _____

28. Would the option to undergo anonymous predictive testing affect your decision about whether or not to test?

☐ Yes
☐ No
☐ Unsure

***29.* Please explain the reason for your answer (Q28).**

***30.* How important would the option of testing anonymously be to you if you were given the option to do so?**

☐ Extremely important
☐ Very important
☐ Neutral
☐ Mildly important
☐ Not at all important

***31.* For those who have not completed the predictive testing process, if you have any additional thoughts or suggestions that you wish to share with us, please include them here:**

Thank you for participating in our survey.

The remaining questions are only for those who have completed the predictive testing process.

32. **If your answer to Question 24 is YES (you complete the testing process), what were your personal reasons for seeking predictive genetic testing?**

33. **Please indicate the year hat you completed testing. _ _ _ _**

34. **Please select the health care providers who were involved in your predictive testing. (You may select one or more responses).**

- ☐ Geneticist
- ☐ Genetic counselor
- ☐ Neurologist
- ☐ Psychiatrist or mental health provider
- ☐ Social worker
- ☐ Nurse or nurse practitioner
- ☐ Primary care physician
- ☐ Other

35. **If you chose to have someone accompany you to your predictive testing visits, who was it?**

- ☐ Parent
- ☐ Sibling
- ☐ Other family member
- ☐ Friend
- ☐ Spouse
- ☐ Partner
- ☐ Other

36. **Please select the answer that best describes how the costs for predictive testing were paid.**

- ☐ Private payment (cash/check)
- ☐ Private health insurance
- ☐ Government-sponsored health insurance
- ☐ Disability or worker's compensation insurance
- ☐ Other (please specify): _____

37. **How important was the cost of predictive testing in making your decision to proceed?**

- ☐ Extremely important

- ☐ Very important
- ☐ Neutral
- ☐ Slightly important
- ☐ Not at all important

38. **Please indicate whether you were given the option of testing anonymously.**

- ☐ Yes
- ☐ No
- ☐ Unsure
- ☐ Other (please elaborate): _____

39. **Please indicate how you identified yourself to the genetic testing program for predictive testing.**

- ☐ True identity
- ☐ Anonymously
- ☐ Other (please elaborate): _____

40. **Please discuss your reasons for the identification you used.**

41. **How important would the option of testing anonymously be to you if you were given the option to do so?**

- ☐ Extremely important
- ☐ Very important
- ☐ Neutral
- ☐ Mildly important
- ☐ Not at all important

42. **How many in-person visits did you attend during the predictive testing process?**

__ __

43. **How do you feel about the number of in-person visits?**

- ☐ Too many
- ☐ Adequate
- ☐ Too few
- ☐ Other (please specify): _____

44. **What type of follow-up did you receive after completion of your predictive testing?**

45. **How satisfied were you with the predictive testing process?**

- ☐ Very satisfied
- ☐ Somewhat satisfied
- ☐ Neutral
- ☐ Somewhat dissatisfied
- ☐ Very dissatisfied

46. **How confident are you that your privacy was maintained during the process?**

- ☐ Very confident
- ☐ Somewhat confident
- ☐ Neutral
- ☐ Somewhat unsure
- ☐ Not at all confident

47. **Do you feel that there were any inadvertent disclosures about your identity?**

- ☐ Yes
- ☐ No
- ☐ Unsure
- ☐ Other (please specify): _____

48. **Which issues, if any, did you encounter during your predictive testing experience that you think might be done differently or better?**

49. **What was your gene test result?**

- ☐ Expanded (Positive Test)
- ☐ Not expanded (Normal Test)
- ☐ Intermediate test results
- ☐ Reduced penetrance
- ☐ Other (please specify): _____

Note: In most laboratories, HD genetic testing interpretation is as follows. Please note that CAG repeat length interpretation may vary slightly from one diagnostic laboratory to another.

CAG Repeat Length	Interpretation
< 27	Normal/negative
27 – 35	Normal but potentially unstable, "intermediate"
36 – 39	Reduced penetrance
> 39	Expanded/positive

50. **For those with expanded, intermediate or reduced penetrance, please specify number of CAG repeats on the expanded gene if known: __ __**

51. **How do you feel that your genetic test results affected you?**

52. **If you have any additional thoughts or suggestions that you wish to share with us, please include here.**

Thank you for your time and effort. We appreciate your participation.