3-2013

Direct-to-Consumer Genetic Testing: A Survey of Consuming Perceptions

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DIRECT–TO–CONSUMER GENETIC TESTING: A SURVEY OF CONSUMER PERCEPTIONS

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Masters of Science
Science, Technology and Public Policy
Thesis Submitted in Fulfillment of the
Graduation Requirements for the

College of Liberal Arts/Public Policy Program at
ROCHESTER INSTITUTE OF TECHNOLOGY
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March 2013

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ACKNOWLEDGMENTS

I would like to thank the following individuals and entities for their support in this research.

- Professors Deborah Blizzard, Ronil Hira, Javier Espinosa for agreeing to serve on my Thesis committee, their review of my work, and for their continued support throughout this process
- The RIT Faculty and staff for their thoughtful responses to the survey, and for their valuable time
- The Rochester Institute of Technology (RIT) Institutional Review Board (IRB) for their timely review and approval of this work
- My parents, Keith and Maureen Blankley, and my fiancé Kiersten Shinrock for their continued support and guidance

ABSTRACT

INTRODUCTION AND LITERATURE

New technology presents difficulties for policy makers in that it is impossible to determine every subsequent impact of a novel technology when it is introduced to society. Direct-to-consumer (DTC) genetic testing exemplifies this problem while presenting the additional complication of having an impact on both individual and public health. Little research has been done on what consumer perceptions of information presented to them are. Some studies have demonstrated a need for further work and expert consensus has identified issues with advertisements but no research has been done on consumer perceptions.

METHODOLOGY

A cohort of faculty and staff at the Rochester Institute of Technology (RIT) were presented with a screenshot of a website from a leader in the DTC genetic testing industry and asked about their perceptions of the genetic tests presented on that page. The survey was distributed via email and presented using RIT Clipboard software.

RESULTS AND DISCUSSION

103 responses to the survey were received and analyzed. There was a wide range in the answers provided to questions but several themes emerged upon analysis. This population was significantly more educated than the general U.S. population. Many respondents indicated some form of knowledge in science and or technology, either through formal education or work experience. The responses indicated a significant lack of understanding of the information presented by the company. Some respondents demonstrated a misunderstanding of the basic concepts underlying the information presented and a failure to correctly interpret the advertisement. These results indicate a potential need for policies regarding the structure, content and interpretation of these advertisements. Further research should focus on establishing similar results for other genetic tests and DTC genetic testing companies as well as developing methodologies to assess retention of information and economic and political acceptance of potential regulation.

INTRODUCTION

In any field, new technology presents substantial difficulties to policy makers. When the public's health can be impacted by a new technology, those difficulties can become real and immediate threats to public well-being. As medical technology continues to advance, policy needs to be continually updated to deal with the new benefits and possible drawbacks to each development. Within this arena, genetic testing has become an issue which has advanced to a point where there may be need for government oversight. Among the promises of genetic testing are better health outcomes for patients due to improved clinical treatment. The eventual goal of utilizing this information is to improve patients' quality of life and to increase longevity. However, the benefits and costs of genetic testing are difficult to determine and many factors complicate the clinical treatment of patients. Direct-to-consumer (DTC) genetic testing presents an additional problem in that a large amount of complex information is made available to the consumer without the interpretational assistance of a trained professional. There are concerns regarding multiple aspects of these tests and there is little oversight from the federal government of the tests and the surrounding issues. This lack of oversight may eventually lead to poorer outcomes for patients and an increased burden on the healthcare system.

Genetic testing is a process by which a specific mutation or mutations are identified in the genome of an individual. In a clinical setting these tests should lead to a diagnosis of a specific disorder or influence a treatment decision. There are two key indicators of whether a genetic test is useful. Analytic validity is the ability of the test to accurately identify the specific mutation for which it was designed. Clinical validity means that the mutation being tested for has an impact on the how the patient's treatment and can positively influence a health outcome.

A genetic test which demonstrates both of these qualities is that for Hereditary Non-polyposis Colorectal Cancer (HNPCC) gene. The presence of this gene indicates an approximately 80% chance of developing colorectal cancer. The increased risk is

demonstrative of the tests analytic validity. The identification of a family history and subsequent testing for and discovery of this gene alter the course of screening for carriers. Normally an individual is recommended for screening every ten years, beginning at age 40 for colorectal cancer. However, if a patient is identified as having the HNPCC mutation, they should begin screening between ages 20 to 25 and continue to be screened every one to three years. This change in screening demonstrates clinical validity. The added genetic knowledge is beneficial because it increases screening procedures which may find a tumor at an earlier stage, increasing the likelihood of a positive outcome for the patient. (Rex, Johnson, Lieberman, Burt, & Sonnenberg, 2000). Unfortunately the number of tests which have been shown to demonstrate both of these traits is a very small percentage of the hundreds of commercially available tests. If a test has neither analytic nor clinical validity it is not generally recommended by the majority of health and scientific communities.

The use of genetic testing in the clinical setting is a fairly recent development. The process typically involves a physician ordering a test in order to confirm a diagnosis or to determine a specific treatment. These tests are performed by different laboratories, usually outside of a physicians practice. Each physician or hospital has purview to decide which company he or she uses to complete genetic testing for their patients. Typically, a physician only orders tests which have the ability to positively impact physical health outcomes for an individual. Due to the variations in human reaction and experience the mental health impacts (such as stress and anxiety due to a misinterpreted test result) are even more complex than the physical implications and are outside the scope of this work. The physician then reviews the results and implications with the patient and then the two make decisions together based on all available clinical information, including genetic test results.

Having a physician aid in decision making is ideal for the patient because it provides an opportunity for someone with applicable knowledge to help the patient understand information and make informed decisions. However, the advent of direct-to-consumer (DTC) advertising presents a dilemma to each person considering genetic testing. Due to its much longer history, DTC advertising is most often discussed in the context of

prescription drugs. However there has been a recent and dramatic increase in the number of genetic testing laboratories using this method to attract customers. There are many protections from DTC advertising of prescription drugs by numerous agencies of the federal government. These protections limit how companies can represent and market their products. However, the federal government has not implemented similar regulations on genetic testing advertisements. In addition, the potential consumer of a prescription drug is protected by the fact that a physician can act as a gatekeeper because a doctor is still required to write a prescription for a particular medication. In many states patients can order genetic tests directly from companies and receive results without any physician or genetic counseling guidance. Economic theory suggests that the lack of a gatekeeper results in an incentive for firms to create supplier induced demand. This would create an overproduction of genetic test (Folland, Goodman, & Stano, 2007). Firms can accomplish this through aggressive marketing and advertising and this is arguably what we see in the current genetic testing market.

The Centers for Medicare and Medicaid Services (CMS) the Food and Drug Administration (FDA) and the Federal Trade Commission (FTC) are the oversight bodies for laboratory developed tests (LDT's) such as genetic testing. The Clinical Laboratory Improvement Act (CLIA) is the law which grants CMS the ability to conduct periodic "proficiency testing." This act was designed to "strengthen federal oversight of clinical laboratories to assure that the tests results are accurate and reliable." It was based on congressional hearings which determined that lab tests had a large impact on clinical outcomes (Javitt, Stanley, & Hudson, 2004).

A significant gap in this act is that it lacks specificity as to which tests to regulate and how to regulate them. It also contains no provisions regarding *the advertisement* of any medical device or procedure, including genetic testing. Over time the FTC and the FDA have stated that they have joint purview over the advertising of LDT's. However, neither has taken the initiative to regulate the advertising of genetic tests. Both have cited limited resources as the reason why genetic testing advertisements are not regulated similarly to prescription drugs (Gollust, Hull, & Wilfond, 2002). The only action taken by the FTC to date is a warning statement issued in 2006 that consumers

should be wary of claims made by testing companies and should enlist the help of a qualified physician in deciding whether to order genetic tests and interpreting the results. The FDA has only begun to take action and has started to warn companies and held open forums relating their concerns and allowing for input from parties with vested interests regarding advertising practices (Pollack, FDA Faults Companies on Unapproved Genetic Tests, 2010).

Although not cited by the government agencies as a barrier to regulation, it is also the case that the link between genetic testing and poor health outcomes is difficult to establish. The connection between prescription drugs and negative health outcomes has been more easily identified, placing more pressure on the government to regulate those ads. In the case of genetic testing there are several logical steps between the advertisement and the health outcome. First, the patient must decide to take the test, the patient must then interpret the test results, and then the patient must make a decision whether or not to consult others regarding the test results. Finally the patient must make a decision regarding his or her health. That decision will have an impact on the patient's health and may be positive or negative or neutral depending on a multitude of factors. Figure 1 demonstrates these steps graphically. They can choose to include or disregard the genetic information in that decision. As a final note, the patient must also decide whether to tell his or her biological family about the test results, which may have implications for their health as well. These complex steps make the justification more complex and thus remove pressure on the federal government to establish regulations.

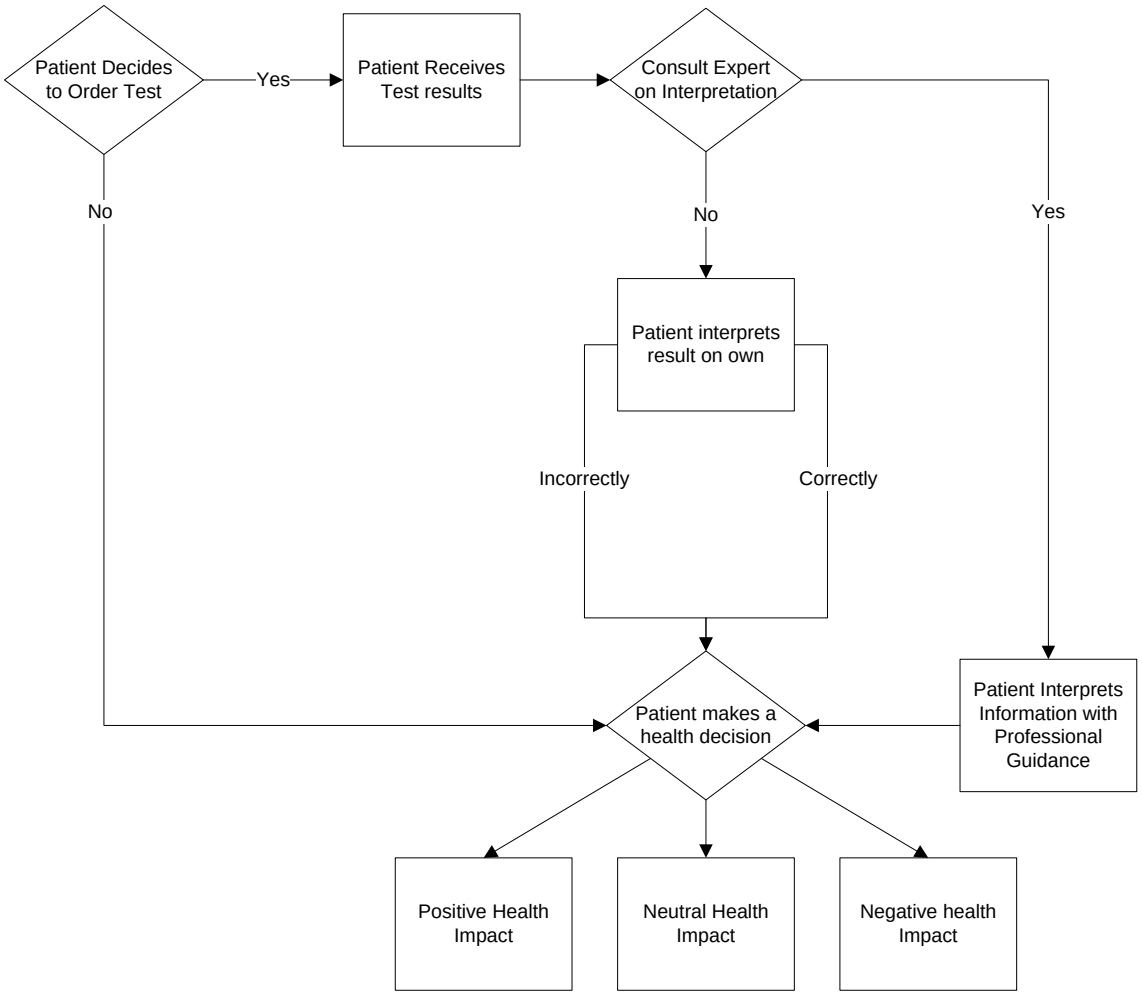

Figure 1. Flowchart of patient decision making steps which may involve a genetic test.

The lack of federal oversight has forced individual states to legislate and enforce individual regulations. This has left the country with a patchwork of laws with varying strictness. Some states have remained entirely silent on the issue. Others require that patients meet with physicians for any genetic testing which has clinical applications. Still other states have completely banned any DTC advertising and only physicians may order these tests. Because the internet is such a widely utilized resource, both for companies and persons seeking genetic tests, it is difficult to determine who is being impacted by a given advertisement. It is entirely plausible that a patient in one state with strict regulations will visit the site of a company in a state with no regulations and obtain genetic testing without a physician's guidance, rendering the state law moot.

A typical scenario where the lack of regulation would negatively impact health outcomes might play out as follows. A woman with a family history of breast cancer is interested in seeing if she has an increased risk for cancer. She searches the internet for genetic testing sites and although she lives in New York, a state which strictly prohibits DTC advertising she is able to access *23 and Me*'s website. The company's advertisement states "BRCA1 and BRCA2 mutations account for most (though not all) cases of inherited breast cancer in women (23 and Me, 2011)." Relieved that there is a test which can assuage her fears she purchases the test. She is mailed a test kit and sends her sample back to the laboratory. In a week she receives the results, she is negative for both BRCA-1 and BRCA-2, meaning that she is not at increased risk for hereditary breast cancer. Because she has not spoken with a physician she takes this to mean that she will not get breast cancer. However, while this result does decrease her risk of hereditary breast cancer it does not account for the large percentage of cases which are not hereditary. In fact, having either BRCA-1 or BRCA-2 increases the lifetime risk of a woman developing breast cancer from 12% to only 60% (National Cancer Institute, 2005). The woman in this scenario still has a 12% lifetime risk of developing breast cancer. *23 and Me* does present information to this effect on the page but it is unclear whether consumers are able to draw the complex connection. If she misunderstands it, this information reduces the likelihood that she will engage in preventative behavior (mammograms and self-exams). Five years later she notices a lump. By the time her cancer is diagnosed it has metastasized throughout her body and in addition to the personal costs and pain associated with therapy her medical costs skyrocket. This anecdote provides one example of how the misinterpretation of a test result could lead to poor health outcomes for the patient.

This scenario illustrates how poor information and the lack of regulation could have a devastating impact on a patient. Although the company's website was technically accurate, it did not provide all the information the patient needed to make an informed decision about her healthcare. Because the regulations were different in each state, the state law which was supposed protect her was rendered useless by the interstate nature of the internet. A federal regulation or interstate pact may be required to protect the health of both individuals and the population. Regulating this type of advertising on a

national level would help to prevent this and similar issues from occurring due to inconsistencies in regulation.

There is a precedent for national regulation in light of inconsistent state laws regarding a medical topic. The issue of abortion has remained contentious for decades but prior to the 1973 ruling of the Supreme Court in Roe v. Wade individual states had differing regulations on whether women were allowed to get abortions. The result of this was that women in states with restrictive abortion laws were forced to either travel to a state with fewer restrictions or to obtain the abortion illegally. This decision could result in significant pain, sterilization, and even death. By setting a national standard, the Supreme Court was able to make a medical procedure available to all women regardless of which state they resided in (Pence, 2011). The case with DTC genetic testing is similar in that requiring individuals to consult a physician would be equivalent to providing individuals with access to a service consistently on a national level.

While a link has been established between poor analytic validity and poor health outcomes, the link between incomplete or misleading advertising is less proven. Beyond that, no one has examined whether the advertisements themselves are misleading to consumers. There have been a few studies which have established that advertisements may contain incorrect information, but no one has determined whether patients can understand information presented accurately in advertisement, or whether the misleading advertisements have an effect on actual patient decisions. Anecdotal evidence suggests that this may be the case but there have been no systematic studies of how consumers process the highly technical information presented in genetic testing DTC advertisements (McBride, Wade, & Kaphingst, 2010).

A New York Times article examined the extent to which direct to consumer genetic testing was occurring in March of 2010. The industry's leader, *23 and Me* was noted to have tested 35,000 customers. Two other companies, Navigenics and Decode Genetics have provided services to 20,000 and 10,000 customers respectively. The article itself argues that the market is relatively small. However, the numbers tell a different story. According to a filing with the Securities and Exchange Commission in December 2009

23 and Me raised $27.8M. This indicates that the size of this problem is not insignificant and while these companies may begin to struggle (as the article contends), this could cause them to utilize even more aggressive advertising, further inflating the demand and potentially causing more harm to misled consumers (Pollack, Consumers Slow to Embrace the Age of Genetics, 2010).

Another news article in the Washington Post compared DTC genetic test marketing to the way Apple aggressively marketed the iPod and how other high tech companies market their products (Cruikshank, 2010). What this article fails to note is the health impact from a genetic test is still unproven and it is unclear whether aggressive marketing practices are appropriate for products which have a primarily medical purpose. There are also questions regarding whether consumers can comprehend material presented in these types of advertisements. While both examples demonstrate the power of consumerism, when the products being marketed may have impacts on health it is even more important for consumers to understand what they are buying.

Within the past several years genetic testing has become more publicized in media outlets. Highlighting this increase in attention is a series of articles by Greg Pollack in the New York Times (of which the articles noted previously are a part) which has followed the issue closely (Pollack, FDA Faults Companies on Unapproved Genetic Tests, 2010). As this issue gains more attention it is likely that more people will be interested in obtaining these tests, whether or not they have clinical value. Pollack's articles focus on government action against DTC companies and one could argue that they are a call for action. As a result of the increased public attention and other factors in 2010 there was a dramatic increase in action by the FDA on looking into DTC genetic tests (Singer, 2010). However, no finalized regulations have been established and there is a call for more research to be done examining what the potential impacts of this technology could be and whether it should be regulated. One aspect of this research is examining how consumers process and comprehend the information presented in DTC genetic test marketing.

BACKGROUND AND LITERATURE

There are still significant questions as to whether these tests and their advertisement should be regulated, yet few studies have examined the impact of advertisements on clinical outcomes. There are many editorial and opinion pieces published regarding this issue, but there is little research to support them. The studies that have been conducted focus primarily on the actual validity of the genetic tests. Research focused on the advertisements themselves use expert opinions of the factual information in the advertisements but do not account for the general public's perception of the issue.

The DTC advertising of prescription drugs is closely related to that of genetic testing. Prescription medication advertisements are strictly regulated therefore more research has been done regarding the impact of DTC advertising of prescription drugs than genetic tests. One study found that ads for prescription drugs are insufficient for consumers to make informed decisions (Kanfar, Louden, & Sircar-Ramsewak, 2007). This research can be applied to genetic testing advertisements as well. Because of the complexities involved with genetic testing and the lack of regulations, the confusion is likely to be even greater with consumers of genetic tests.

Many companies have defended their DTC policies by stating that consumers are not utilizing the information for clinical reasons. This theory was shown to be questionable by a study which determined that 78% of consumers who would have a genetic test would seek their physicians help in interpreting the results (Caufield, Ries, Ray, Shuman, & Wilson, 2009). This indicates that the primary reason for patients to purchase genetic testing is that they believe knowing their genetic structure will have some positive impact on their health and their lives. Whether or not this belief is an accurate one is something that is still not proven.

An article published in the *Journal of the American Medical Association* (JAMA) examined several advertisements for their accurate portrayal of a test and whether the tests themselves had both clinical and analytic validity. Their research found that each

of the advertisements had at least one of three problematic aspects. The first is that the advertisements themselves were misleading. The second is that the information being advertised is complex, a fact which cannot be accurately portrayed in the abbreviated communications. Lastly there is a large amount of contention over whether the tests being advertised have substantial clinical utility (Gollust, Hull, & Wilfond, 2002).

In order to determine the clinical validity of genetic tests for cancer Marchionni et al. examined three separate tests. They found that the three varied in their ability to both accurately analyze the genetic information and provide valuable clinical data. Of the tests studied, *Oncotype Dx,* had the best clinical utility, but even its clinical utility was insufficient to be considered definitive. Further, the authors determined that more population genomic research was required in order to prove clinical utility (Marchionni & Marinopoulos, 2008). Although this was not mentioned in the article, all three tests were available to the public and were advertised as pharmaco-genetic indicators.

A report from the conservative bioethical think tank, The Hastings Center, identified one print advertisement for BRCA-1 and BRCA-2 genes and systematically reviewed it for its merits and problems. They determined that the ad overstated the test's ability to predict and misrepresented what the test actually did. This determination was made by a group of experts in the field (Hull & Prasad, 2001). While this study does present valuable information, it only establishes a problem for one advertisement and does not consider how the public itself would view the advertisement. The authors assume that because the public has a limited knowledge of the tests that they would be misled. There has been no research to date regarding whether this assumption is accurate.

In a study published late in 2010, Rahm reviewed the findings of a focus group of Kaiser Permanente Health Plan members regarding their thoughts on genetic testing. The group considered both genetic information presented in advertisements and test results. The conclusion of the author was that there was a large amount of individual variation with regards to opinions on genetic testing and potential impacts of test results on individual decision making. This study demonstrates that there are many factors which

may influence how patients comprehend and process information presented in these advertisements (Rahm, 2010).

Another study by McBride et al. examined the impact of genetic test results on consumer decision making. The authors note that there has been no established research demonstrating harm to patients as a result of receiving DTC genetic tests. The authors recommend accelerated research including long-term responses to information, optimal support decision making and looking at primary care provider competency as it relates to genetic testing. The implications of this paper are that it solidifies the need for future research on this topic and the social impacts of DTC genetic tests, specifically the link between DTC genetic tests and clinical outcomes needs to be established (McBride, Wade, & Kaphingst, 2010).

In addition to these studies, several editorials and opinion pieces have theorized that the general public does not have the knowledge necessary to make informed decisions without the help of a physician. A statement from the Genetics and Public Policy center claims that, "consumers are vulnerable to being misled by advertisements and lack the knowledge to make appropriate decisions about whether to get tested or how to interpret the results (Williams, 2008)." This and similar theories have not been examined systematically and we are uncertain whether the public does have the understanding required to properly interpret the test results.

Along with the peer-reviewed literature, and in conjunction with the public interest in this topic, there have been two hearings held before various House of Representative subcommittees by the Government Accounting Office (GAO). In 2006 the GAO testified that advertisement claims were often vague and at times blatantly false and that they contributed little to patient health awareness (Government Accounting Office, 2006). More recently the GAO presented a report which stated that the tests themselves are misleading and that they are being marketed inappropriately to consumers. The GAO sent DNA samples to five different companies and often received conflicting information from those companies as to genetic risks (Government Accounting Office, 2010).

METHODOLOGY

In order to assess how consumers understand these advertisements a survey was utilized which measured their scientific background, comprehension of the information presented and key demographic questions. A survey was selected as the research instrument in order to gather a wide range of information on a relatively large cohort at a specific moment in time.

The instrument can be found in Appendix A and contains questions in three sections: demographics, interpretation of the advertisement, and scientific background. The participants were presented with information from 23 and Me's website regarding their genetic tests for alcohol dependency and were asked a number of questions on the material presented. Questions included multiple choice, "Yes / No" free text and likert responses.

COHORT

The faculty and staff at the Rochester Institute of Technology (RIT) were chosen as a cohort for several reasons. There is likely to be a mix of education both in terms of highest level achieved as well as field of study. Students were not included because of their limited age range and a decreased likelihood of purchasing the test in the near future. Including a wider range of ages may provide information regarding differences in age groups and comprehension. This cohort was also selected because they are more likely to be interested in obtaining the genetic tests and more likely to have the means to purchase the tests. Lastly, they represent a convenience sample due to time and resource constraints.

There are potential limitations associated with this cohort. There is likely to be a bias toward persons with a high amount of technical skill because of the nature of the university being sampled. With a history steeped in technology and a faculty and curriculum that support technological education there is significant cultural importance placed on that subject. The result of this is that it is likely that individuals who have an

interest in technology and science are likely to be drawn to this university. In addition, because the survey was distributed by email, the sample is limited to faculty or staff with an RIT email account, potentially excluding less educated members as they would be less likely to have an email address that is checked frequently. There may also be a self-selection bias as persons who have a background in biological or medical sciences may be more apt to respond to the survey. As a result of these biases the results may not be able to be generalized to the U.S. population. However, if this cohort still demonstrates a lack of understanding of the advertisements, it is unlikely that the general public will be able to interpret them.

INSTRUMENT

The RIT Institutional Review Board (IRB) input and approval were obtained during survey development and prior to distribution. The survey was distributed to faculty and staff at RIT via an email containing a link to an RIT clipboard survey and a brief description of the research and its intent. Distributing the survey in this manner created a straightforward way for participants to respond and ensured that the majority of the faculty and staff receive the email. The limitations of this method are that it limits persons without consistent email access from participating and therefore may skew the results towards faculty and away from staff. Additionally, a survey limits the ability for follow up discussion based on participant responses.

Participants were presented with a screenshot taken from 23 and Me's website. Specifically the entire webpage on alcohol dependency was presented unaltered as it was available in February of 2011. They were asked to take several minutes to review the screenshot and familiarize themselves with the information. There was no time limit placed on the review period since respondents were able to return to the material during the survey (although this was not suggested or mentioned in the instructions). The respondents were asked to begin answering questions once they felt comfortable with the material.

This company was selected because it is currently the largest distributor of DTC genetic tests in the U.S (Pollack, Consumers Slow to Embrace the Age of Genetics, 2010).

Alcohol dependence was selected to be the condition and test because it is not gender dependent, is relatively well-known by the public, and has both genetic and environmental factors which contribute to its clinical presentation. 23 and Me identifies their test as a "disease risk" test and also as a "preliminary research report." The number of tests noted as preliminary is 72%. The majority of tests are also classified as "disease risk" (23 and Me, 2011). Therefore alcohol dependency is representative of the majority of tests made available by this company

The first section of the instrument asked for demographic information from the respondent. Because genetic tests vary in reliability depending on ethnicity and sex it was important to have the participants self-identify both of those factors. In addition, participants were asked to identify their level of education and area of study (if applicable) as well as their current occupational field. Both of these measures were collected as potentially important predictors of comprehension.

The second section of questions attempted to establish what participants were able to glean from the information presented to them. They were asked to respond to several questions on both the content of the advertisements and what their impressions of the test itself are, as well as whether they might take action based on the results of the test. Questions included "Yes/No", free text, Likert scales and multiple choice. The type of answer was dependent on the question being asked and whether the potential answers could be easily categorized. In some instances it was important to allow participants the ability to react in depth to the question. For other questions, scales and categorized answers allowed for easier analysis and created a homogenous data set.

Lastly, respondents were asked several questions to measure the depth their scientific background and literacy. These questions were primarily an effort to establish whether the assumption that persons at RIT may be more technically savvy is correct. These questions also helped to identify possible correlations between scientific understanding and advertisement comprehension. Without better understanding how participants understand the background material it would be impossible to interpret their responses to

the comprehension questions. For a listing of all questions and the purpose behind each particular question please see Table 1.

TABLE 1. A list of each question and reasoning behind why it was asked. When applicable, the reason for the question being asked in a particular way is noted

QUESTION	PURPOSE	ANTICIPATED RESPONSE
1) What is your age?	Identifies age-rage of sample for normalization purposes Helps control for the topic, as alcohol is pervasive on college campuses	Varied, but with a distribution centering around between the ages of 30 and 50
2) What is your sex? • Male • Female • Other	Identifies sex of sample for normalization purposes	50% male and 50% female
3) Education (please select one) • Some High-school • Graduated High-school • Attended some college • Associate's degree • Bachelor's degree	Identifies education of sample to compare to U.S. population Provides insight into whether a correlation may exist between education level and knowledge of topic	The majority of individuals will have a at least a bachelor's degree,
4) If you have received a college education (associate's or more) please indicate your major	Further clarifies question 3 for analysis	A high percentage of liberal arts, science and engineering respondents
5) What is your highest terminal degree?	Further clarifies question 3 and 4 for analysis	Many advanced degrees
6) What is your current occupation?		Administrators / Faculty
7) Ethnicity • African America • Asian / Pacific Islander • Caucasian • Latin / South American • Native American • Other	The genetic test presented only applies to a certain ethnicity, ethnic background of sample may have impacted responses	A large percentage of Caucasians
8) What are your impressions of alcohol dependency and genetics?	Asked before analysis questions to attempt to obtain a baseline of participants' understanding of topic	Varied by individual
9) How many studies are described in this advertisement?	Attempt to determine whether the *structure* of the website information was clear	3
10) If you had a family member who had a dependency on alcohol would you be interested in getting a genetic test for yourself? • Yes • No	This question was developed to determine the participants interest level in alcohol dependency	Varied by individual

11) If you had a blood relative who had a dependency on alcohol would you be interested in getting a genetic test for yourself? • Yes • No	Attempt at determining whether participants would vary their response based on a hereditary vs. familial proximity to the topic	Varied by individual
12) What is the relationship between marker RS27072 and alcohol dependency?	Attempt to determine whether the *content* of the website information was clear	Having a particular allele lowers the odds of having withdrawal seizures
13) How closely are genes and alcohol dependency related (Likert)? • Not closely At All • Not Very Closely • Somewhat Closely • Fairly Closely • Very Closely	What were the participants' overall impressions of the website and how closely the genetics and alcohol dependency were related? A likert scale was utilized to simplify for coding purposes	Varied by individual
14) Does ethnicity have an impact on this relationship? • Yes • No	Did the participants notice the ethnicity qualifier present on the website?	Yes
15) If so, how?	Clarifying question 14. If the participant guessed correctly, A free text response allows them to explain their response	Studies only included European and Asian ancestries, results may not apply to other ethnicities
16) How interested are you in this condition (Likert)? • Very Uninterested • Not Interested • Neutral • Somewhat Interested • Very Interested	An attempt to control for formal education. If someone was very interested in the topic they may be more educated than would be typical	Varied by individual
17) Approximately how many people's genetics were studied to determine the relationship between marker RS18001197 and alcohol dependency? • 10 • 100 • 1000 • 10000	Attempt to determine whether the *content* of the website information was clear	10,000
18) What would you do if you received a test result that indicated you had this gene? • Nothing • Seek more information on the condition on the internet • Talk to my doctor about the risks • Call the company to speak to a genetic counselor	A look forward at what the potential "end-user" acceptance of the policy would be if they were required to utilize a physician as a gatekeeper	Varied by individual

19) Where did you receive the majority of the information? • Charts • Graphs • Numbers • Text • Photos	This question was aimed at obtaining which piece of the website the participants found most helpful.	Varied by individual
20) What is an established research report?	An attempt at determining what the general consensus was regarding this language as it is not defined by any traditional convention, yet is used by 23 and Me.	There is no official scientific definition for this term
21) What journals did the research stem from?	This question's intent was to determine whether individuals could determine the journal names based on their abbreviations	Archives of General Psychiatry, Molecular Psychiatry, and Alcoholism: Clinical & Experimental Research
22) What is a SNP?	Because this acronym is used in the website but not defined, this was asked to determine what the general knowledge of the term was	Single Nucleotide Polymorphism
23) Susie requested that this analysis be done for her. The result stated that she had an increased risk for alcohol dependency. The letter from the lab said that her test was ____. • Positive • Negative	Because there are differences between the social and clinical connotations of positive and negative, this was asked to see whether participants could separate those meanings.	Positive

ANALYSIS

The analysis of the survey results included both basic statistical and interpretative qualitative analyses. Interpretive analysis was used on any answers which were provided via free text. In order to obtain the most information from this limited sample it was important to use qualitative methods such as interpretive analyses to better understand the respondent's mindset. By using basic statistical analyses to establish areas of interest and furthering that work with interpretive analyses I was able to develop an adequate picture of whether this sample of consumers understand the advertisements and whether the ads have a potential impact on their future decision making if they were to obtain the test.

Free text was analyzed with either interpretive analysis or a coding scheme, depending on the information. Therefore the coding scheme was developed after data based on the

data which was obtained. Using this scheme, themes were developed and categorized to both help describe the sample and identify trends.

Simple statistical analyses looking at correlations between all the variables were developed in order to examine if any relationships may exist. The relationship between education and comprehension were examined (as measured through several questions) were as well as between ethnicity, age and gender and potential decision making. The apparent gaps in understanding were especially important to review, given sample's scientific background.

RESULTS

103 responses to the survey were received. All responses were included in the analysis. While it was clear that some persons were unable to see the actual advertisement it was decided that their response to the questions was as valuable as the ones who did not complete the survey. There was an issue with one particular browser type and the ability to view the screen shot. The responses they provided to the scientific background and demographic questions should not have been impacted by the advertisement. Further we can assume that they chose not to answer questions pertaining to the advertisement when they could not see it. Therefore no response was eliminated from the analysis. Where noted there were questions left unanswered by certain respondents.

Demographically the sample was relatively diverse. 54% of the respondents were female and 46% were male. The average age was 47.8 years, with a standard deviation of 15 years indicating that 68% of the respondents were between the ages of 32 and 62. Of those that responded to the question "What is your highest terminal degree," 68% noted that they had achieved a master's degree or higher (n = 60/88). Fields of education were relatively diverse as well with the majority being liberal arts disciplines as well as a significant number being business, science and engineering (See Table 1). Ethnically the vast majority (92%) identified as Caucasian.

TABLE 2. Break-out of demographics by discipline. Note that there is a wide distribution of disciplines identified by the respondents.

Discipline	Count	Percentage
Engineering / Computer Science	20	19%
Liberal Arts / Fine Arts	35	34%
Science	15	15%
Unclear / No Answer	19	18%
Total	103	100%

The next section of the survey focused on the respondent's impressions of the material which was presented to them. The first question allowed free form response to "What are your impressions of alcohol dependency and genetics?" Upon coding of the responses, 50 persons acknowledged some form of relationship, 11 stated that they

believed there was no relationship and 23 noted that both genetics and environment could have an impact on alcohol dependency.

The next two questions focused on determining whether respondents distinguished between family member and blood relatives in terms of relations. When asked if they would be interested in getting a genetic test for themselves if a blood relative or family member had a dependency on alcohol the majority in both cases (66% and 65% respectively) stated that they would not be interested in the test, however 8 respondents (8%) changed their response in some way.

When asked what the relationship between RS27072 and alcohol dependency was there were a wide variety of responses. 24 (18%) individuals stated that they did not know or left the answer blank. 61 (58%) respondents identified "withdrawal seizures" or "symptoms" in their response. 18 respondents identified that there was some relationship but only noted a general correlation to alcohol dependency.

TABLE 3. Categorization of answers to how respondents viewed the relationship between marker RS27072 and alcohol dependency.

Response Category	Total Responses (N)	Percentage (%)
Did not know / Blank	24	23
Noted relationship but no identification of withdrawal seizure correlation	18	17
Noted "withdrawal seizures" or "withdrawal symptoms"	61	59

When asked how closely genes and alcohol were related the majority (52%) stated "somewhat related." The rest of the responses appear to follow a normal distribution in terms of their interpretation of the directness of this relationship. This is demonstrated in Figure 2.

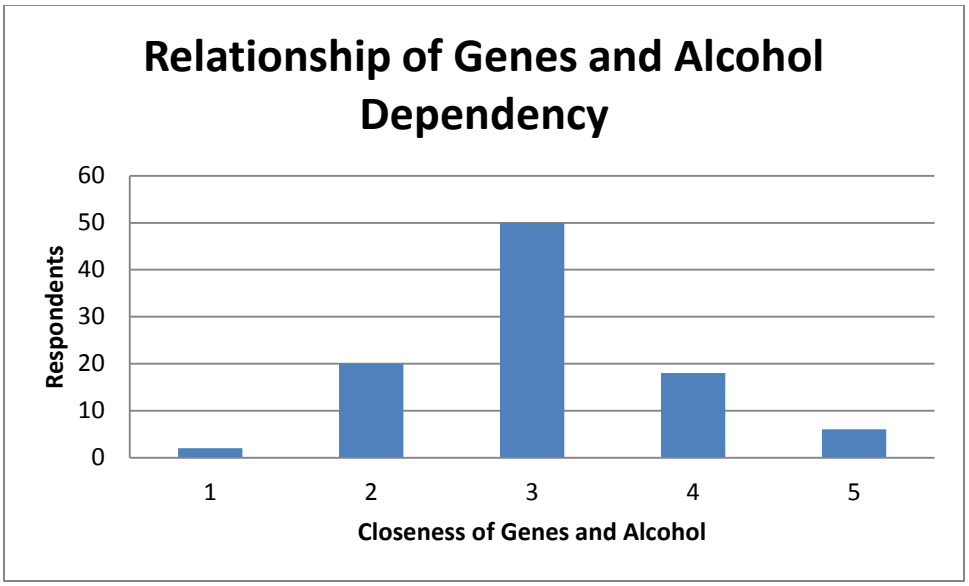

FIGURE 1. Results of the asking survey respondents to define the relationship between alcohol dependency and genetics on a likert scale. Raw counts are presented here to demonstrate the normal distribution of the responses.

Because the advertisement noted the applicable heredities for these tests, the respondents were asked whether ethnicity had an impact on the relationship between genetics and alcohol dependency. 49% answered that it did not, 45% answered that it did and 5 respondents did not answer. When asked to expand upon that relationship, if there was one, respondents answered from varying perspectives. Some listed general thoughts regarding ethnicity, others noted the ethnicities from the advertisement and still others listed that they were unclear as to the exact nature of the relationship.

When asked how many people were studied in order to determine the relationship between a specific marker and alcohol dependency 64% chose the correct answer of 10,000 people, while 18% chose incorrectly and 19% simply did not answer.

Respondents were asked to answer on a Likert scale how interested they were in the condition. 43% chose the "somewhat interested" option with the others selecting neutral, not interested very uninterested and very interested 24%, 15% 9% and 6% of the time respectively. Two respondents did not answer. (See Figure 2).

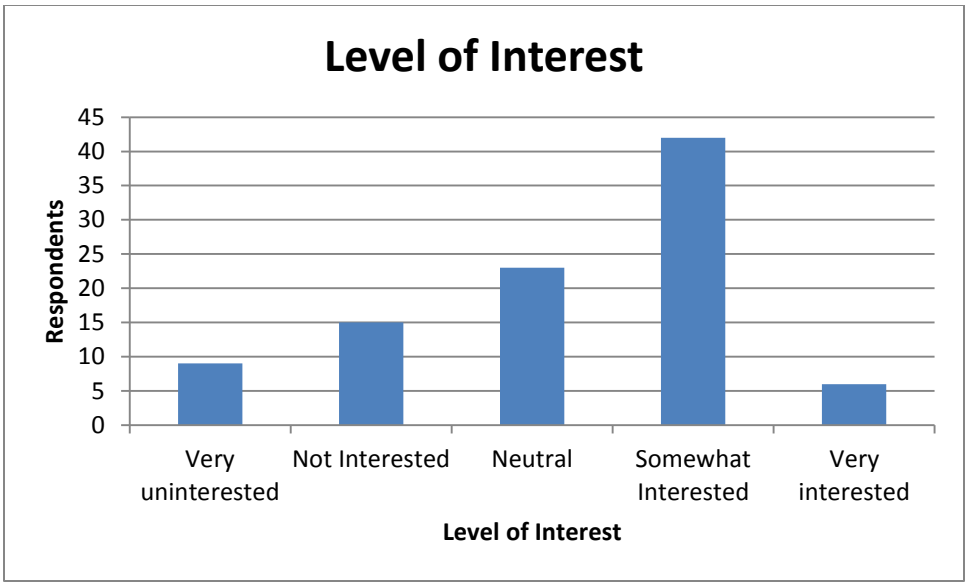

FIGURE 2. Graph demonstrating the level of interest in alcohol dependency noted by respondents who answered (6 participants did not provide a response). As predicted the majority of individuals were somewhat interested or neutral toward the topic.

When asked where they received the majority of the information the vast majority responded 'text' (80%). The remaining noted that they received information from charts, numbers and graphs in descending order.

The respondents were presented with a question which allowed multiple responses to what their action plan would be if they received a test result indicating that they had "this gene." 49 respondents stated they would go to their doctor, 31 would do nothing, 21 would do further investigation on the internet and 6 would seek out a genetic counselor provided by the company doing the testing.

The last segment of questions focused on determining what the basic scientific knowledge of the sample was. Because the company uses the term "established research report" in their description of the tests, persons were asked what that term meant. 21 responded with some variation of "I don't know" or "not sure." 23 did not answer the question. Of those that did answer 26 stated peer review as part of the criteria of a research report. Three individuals noted that "established research report" has no significant meaning.

The respondents were also asked what journals the research was published in. 34 individuals responded by copying the abbreviations listed in the advertisement, 8 respondents wrote out the full titles of the journals. Two respondents made note of the fact that the titles were only presented in abbreviated form.

Individuals were asked what a "SNP" was. Only 12 individuals responded with accurate answers, of these 4 noted that they used an internet source to research the information before responding. The remaining respondents did not answer the question, stated they didn't know the answer or responded inaccurately.

When asked to determine whether a test result was positive or negative for a hypothetical person, 7% of respondents responded inaccurately and 8% chose not to answer the question.

DISCUSSION

The survey was distributed online in order to limit the study sample to persons who might have the opportunity to interact with the material presented in the survey. This limitation was enforced in order to target the survey to a more representative sample of potential utilizers of these tests. Including persons who do not use the internet would have added limited value to the survey responses. In addition, targeting people who have a general understanding of the internet allows the researcher to make certain assumptions when asking questions, allowing for more specific information gathering. For example, it was possible to assume that the respondents would be able to understand how to navigate the survey and respond accurately to the questions. It is likely that individuals familiar with the internet would have taken online surveys previously and would therefore be able to navigate and answer questions with little direction in the content of the survey. From a policy perspective, only surveying those who might interact with the test allows us to understand the opinions of individuals who might interact with the advertisements and tests, thereby allowing us to gauge the perception of those who might be impacted by a regulation.

The population the study sample represented was that of RIT faculty and staff. This population was well represented with an almost even distribution of men and women. The majority of respondents had at least a Bachelor's degree, indicating a higher level of education than is typical for the U.S. citizen. In 2010 29.9% of US citizens had obtained a minimum of a 4 year degree while 84.5% of this sample noted that they had obtained at least that level of formal education (US Census Bureau). This significant difference implies that the overall the individuals studied were more likely than the average U.S. citizen to have some formal education in a scientific discipline. There was a large proportion of respondents with careers in liberal arts, science and engineering. For those disciplines we can assume a higher likelihood that these individuals have received some form of formal training which would assist in their understanding of the material presented. Therefore we can infer a relative familiarity with the material. By

acknowledging this familiarity it is possible to make better inferences to the survey responses that follow.

The demographics of this population were sought for several reasons. Due to the value placed on technology at the university (Rochester Institute of Technology, 2012), the faculty and staff at RIT are more likely to be technologically inclined and therefore potentially more likely to encounter and subsequently purchase DTC genetic tests. Additionally there is a higher likelihood of an above-average education level. However, there is still a large proportion of individuals with limited educations. It may be that those individuals are the most at risk in this environment as they have access and interest in these services but are lacking the formal education that may be required to better interpret both the advertisements and the tests themselves. Due to the nature of the university there may be some pressure for these individuals to have a greater understanding of scientific material and they may be reaching out to unqualified sources for answers, the result of which could be a misunderstanding of the material. For these reasons, this selection of individuals makes RIT an excellent sample set for this research.

Because it was assumed that a typical RIT employee would have a higher education level than the average internet user it is likely that they would have a greater chance of understanding the information presented. By using this strategy, if the individuals with a relatively high education level cannot understand the information then it is likely that those with less education will struggle as much, if not more so. By selecting for more highly educated individuals, there is an increased likelihood that the results are as or more meaningful for persons with lower levels of education. The level of scientific literacy is likely to be higher in those individuals studied and therefore results may be applicable to individuals with lower levels of literacy.

The respondents' answers to the interpretation questions were enlightening. Their impressions of alcohol and genetics were coded in order to glean further information. Because the majority of respondents noted that there was some sort of relationship we can infer that, in general, this population believes that this condition is at least somewhat

inheritable. One respondent was confident enough to respond with exact percentages. They noted: "Variability (38-64%) of predisposition related to genetic factors (Science, Post-Baccalaureate)" While research has shown that alcohol dependency is very directly tied to environment (Theall, et al., 2009), only 23 respondents independently noted the environment in their response. The more typical response was similar to "Genetics do matter, but are not the only important factor (Liberal Arts / Fine Arts, Post-Baccalaureate)" This indicates that the individuals responding were acknowledging that something else has an impact on alcohol dependency but were unwilling to take the next step and indicate that the environment has an impact. This could mean that when examining the advertisement persons tend to relate only to the information presented to them and not consider what they likely know to be true in a different context. It would be interesting to test this hypothesis with a focus group study which provided two sets of participants with different sets of information and determined how they limited themselves to what was presented and / or utilized their own experience to answer questions.

As with many types of social research, the instrument may have had an impact on individual responses. Further exploration utilizing different instruments and methodologies would be useful to confirm this research. Because only one test from one company was utilized it is unclear what impact different tests and different advertisements would have on the results. In addition, by presenting the survey online and allowing respondents to refer back to the materials this study may be an over-estimate of respondents' actual retention and more of a review of their strict comprehension.

This indicates that consumers may require more information than what is presented in the results of the studies by a certain company. While the advertisement does give some background it is general descriptions of the condition. Perhaps companies should be required to give a more comprehensive scientific literature review in these types of advertisements. It is clear that not all respondents have a clear and comprehensive understanding of all of the factors which influence alcoholism.

Alcoholism was chosen as the test profile of choice because of the numerous social issues surrounding it. Even when compared to other substance abuse disorders, alcohol dependency is more highly stigmatized. Persons with alcohol dependencies are more likely to be held responsible for their addictions and provoke more negative emotions than other chemical dependencies (Schomerusl, Lucht, Holzinger, Matschinger, Carta, & Angermeyer, 2010). Because of its prevalence, it represents a type of disorder where persons are likely enough to be familiar with it but unlikely to have extensive knowledge of the topic. There are biases that result from additional knowledge of the subject. One respondent noted that they had experience with Native Americans and as a result believed there to be a genetic influence. While some of this previous knowledge exists in the survey results, utilizing this particular test instead of something as familiar as breast cancer likely limited that impact.

Although it was limited, it is important to note that racial and other biases do exist in the population (as noted previously) and that they might be influencing decision making regarding genetic testing. People's inherent knowledge may be a key factor and force a decision regardless of the information presented in an advertisement or website. Future research should focus on identifying what portion of potential users have already decided to utilize DTC genetic testing before doing online research.

In order to determine whether respondents identified a difference between family and blood relatives receiving test results (and the meaning of each) two questions were asked to that effect. While the percentage of persons responding that they would not be interested remained similar, 8 individuals changed their response. This indicates that for some people having a blood relative receive a positive result is different than having a family member receive one, and that these people may place a higher value on the genetic component.

In general respondents indicated that they thought genes and alcohol were somewhat related. The rest of the responses fell in a normal distribution around that response. While this indicates a broad understanding of the relationship, it is clear by responses to following questions that the details of the relationship are not entirely clear to this

population. Although the information was made readily available as they answered the survey, a large number of participants were unable to articulate the relationship between one of the genetic markers included in the test and alcoholism. As a result of this gap, individuals may believe that they have a greater understanding than they actually do. Due to the publicity that genetics have received over the past two decades it is possible that individuals have a false perception of expertise when it comes to all genetic tests as a result of being informed through lay media which, in general, is unable to convey the full complexities of an issue.

Developing safeguards against this type of mentality may be another place for implementation. Perhaps individuals who receive certain tests should be quizzed on the impacts of those tests and the overall diseases and their relationships to genetics. Informed consent is a concept which has been common to U.S. medical practice for over 50 years (Salas). Engaging patients in a program to ensure informed consent may assist in the incorporation of correct information in to their decision making processes.

In considering informed consent, we must first examine the language utilized in discussions between doctors and patients. The language which is used by doctors and scientists is one with unique rules and subtleties which may not effectively communicate the risk to potential research subjects or patients. Without even knowing it, researchers make assumptions about the knowledge base of the research participant. Even when materials are utilized to measure understanding, such as questionnaires the participant may respond correctly but still not understand the impact of the risk or the full meaning of the question. For example, if a doctor were to tell a patient that the risk from a drug could include a blood clot and the patient answers on the quiz that blood clots are a potential risk; the doctor has not truly assessed the participant's understanding of the risk. Because the doctor knows that blood clots can cause heart attacks and stroke he assumes that the patient understands this as well, but it is possible that this may not be the case at all. When this information is presented in an advertisement it forces the discussion to become unidirectional and removes the ability of the patient to question what is presented.

With a topic such as genetics, science is continually moving forward and in some cases even the individuals presenting information may not fully understand that information. Therefore a question arises as to how it can be possible to inform patients of all the risks. One example of this in the medical community is that of Viagra. Scientists at Pfizer originally developed the drug to treat hypertension. However, during clinical trials they discovered that the drug was much more efficient at aiding individuals with erectile dysfunction (Boolel, Alan, & Ballard, 1996). This "side-effect" had not been considered by the scientists until it was reported by participants and was not likely part of their informed consent briefing. While this side-effect was not extraordinarily harmful it does demonstrate the point that science is ever evolving and that it is unlikely that researchers will even be able to communicate all the potential risks of a new procedure or drug because they themselves are still trying to determine what those are. Currently, genetic testing falls into this category, but because the link between the tests and outcomes is less clear, it is even more likely that the individuals advertising for the test are unaware of all of the potential implications of a positive or negative result.

While complete informed consent may be a lofty goal, the idea of ensuring equality of information should be carried into the realm of DTC genetic testing. While the down-stream implications are not well established, it is clear that when there is a potential for negative consequences, patients need to be as well informed as is possible, both of the scientific possibilities before the test is administered and of what the results truly mean.

The results of this study indicate that there may be a disparity of information in terms of ensuring that consumers have the same understanding of the tests as those who market those tests. In a perfect economy, one would expect a true equality of information between producers and consumers; however it is clear that in this instance there is a better understanding of the product by the producer than the consumer. It is unclear what the final impact this disparity of information might have and future research should focus on determining the extent of the disparity and what impacts, if any it has on patient decision making and subsequent outcomes.

49% of respondents indicated that ethnicity had no impact on the relationship between genetics and alcohol dependency. Given that the studies were ethnicity dependent and that this was noted in the advertisement this response is somewhat startling. The number of people who did not either understand or missed this notation may indicate that it is not presented clearly in the advertisement. While some individuals may take the time to look at the studies referenced by the advertisement it may be unlikely that most would take the time to do so. If an individual trusts an advertisement citing scientific literature to be accurate and does not understand the true meaning of the literature itself it is likely that they will not understand whether the information applies to them. If this mistake is not corrected, the individual may purchase a test which is of no value to them, or worse, they could apply misleading test results to themselves and make decisions based on false information.

There was also confusion when respondents were asked how many people were studied to identify a relationship presented in the advertisement. While 64% did choose the right answer, more than one-third chose incorrectly or did not answer the question at all. Again this may be a result of the information not being presented clearly or persons not being able to interpret it in the way it was intended. Statistically, study size has a large impact on the conclusions that can be drawn from a given research project. The inability of consumers to identify this critical component of the projects is concerning. Further, the results of this study may indicate that the underlying concept of statistical significance is not apparent to this population. Future studies should examine the ability of consumers to both understand why study size is important and whether they are able to readily identify the size of a particular research sample.

These two results indicate that there may be a base set of information that needs to be presented in every one of these advertisements which creates a common understanding of certain terms. Ethnicity is referred to in the advertisement but nowhere is it directly stated which ethnicities were studied to get these results. The same is true of the sample sizes. While the ad presents the number of people in text and as a diagram it is clear that a significant portion of this population was unable to glean this information. It may be beneficial to design and implement a set of conventions for genetic testing advertisement

similar to that of the conventions the pharmaceutical industry must follow when advertising to the general public (US Food and Drug Administration, 2011).

Whether or not there was significant interest in the condition may have had an impact on the results. Excessive interest may lead to a group of individuals with extensive knowledge of the disease and its genetic relationships. Conversely, a bias toward individuals with a lack of interest may indicate a lack of understanding which may result in no knowledge of the disease. Both of these situations are less than desirable if we are attempting to use this population as an analog to the target consumers of these tests.

When asked to identify their interest on a likert scale the majority noted "somewhat interested" with a normal distribution of responses on either side. Therefore it appears that a moderate amount of interest existed in this population. This is what would be expected and therefore helps to normalize this population. There is likely no skewing of the data as a result of excessive or no interest in the condition.

In order to see what respondents' actions might be if they did receive a positive result for this test they were asked what their next step would be. The majority indicated that they would talk to their physician. As noted previously, physicians act as gatekeepers in many instances and serve as a guide for most individuals as they navigate health care. Having an informed and independent party available throughout a process has been demonstrated to have better outcomes. From a policy perspective this is important because it identifies that most people want a physician involved in this discussion and therefore mandating that option or a similar one may be palatable to a majority of people.

A secondary individual responsible for assisting in decision making is often made available in the form of a health care advocate. These advocates can assist patients in making complex clinical decisions as well as aiding in navigating the complex insurance and legal issues inherent in the U.S. healthcare system. In times of emotional distress and when the individual is incapable of making decisions, oftentimes an advocate will speak to the person's wishes, as related from previous decisions when there is no

immediate threat. Typically the individual has clinical training and a good working knowledge of the healthcare system. Although debated, nurses have taken on this role and have been used in a structured model as patient advocates (Hewitt, 2002).

While not entirely applicable, this model may contain certain concepts which would be applicable in the DTC testing arena. For example, it may be preferable to require individuals have a consult with a specially trained social worker or other third party before making any decision based on genetic test results, similar to the idea of a patient advocate. With the advent of genetic testing for clinical purposes a new profession has been developed to act in this role. Genetic counselors now exist and are well organized as a profession with a national professional society (National Society of Genetic Counselors, 2012). Perhaps these professionals should be made available at no cost to the consumer to be utilized as independent resources for persons who decide to purchase DTC testing. Patients could be notified by DTC genetic testing companies before or after purchasing to ensure that those who are interested in genetic tests have an additional source of information and a bidirectional information flow not provided by the advertisements.

23 and Me utilizes a term "established research report." After numerous search attempts on their website it was unclear how they defined this. In order to determine how other might interpret this statement, respondents were asked what they thought it meant. There was wide variability in the answers but a major theme in responses was that established reports were peer reviewed. Most answers were similar to that of one respondent who stated, "A work of scientific research that has been subjected to peer-review within a discipline (Engineering / Computer Science, Post-Baccalaureate.)"

Interestingly, three individuals noted that there was no real significant meaning to this title. One respondent noted "I am not familiar with this terminology. And, I must say, I have been in the business of scientific research for a long time; but I don't attach any particular significance to the modifier 'established' in this context. (Engineering / Computer Science, Post-Baccalaureate.)" Indeed, an internet search for the term "established research report" returns first 23 and Me's website and no definitions of the

term from any organization outside of 23 and Me. Again it is clear that certain terms need to be defined as industry standards in order to ensure that all parties have an equal understanding of the information.

When asked what journals contained the studies reported in the majority of persons simply copied the abbreviations verbatim from the advertisement. A few made attempts at guessing the abbreviations and some even attributed their response to an internet search. One respondent even noted that "Would've b[e]en n[i]ce if they had included the full titles of each (Unknown, Baccalaureate)." Given this, it is unclear how reliable internet sources utilized by these individuals will be. While the persons who noted an internet search did answer correctly this may not be true in all cases. Having individuals rely on third parties to get their information is not a reliable and consistent way to provide that information. In addition, it is unclear how individuals would be able to access the primary literature on the subject and whether the abbreviations presented were clear enough to help users find the journals.

Only 12 individuals correctly identified a "SNP." Of those 12, 4 noted using an internet search. One individual went so far as to say: "According to the web definition, it is a Single Nucleotide Polymorphism; I don't know the science behind it but it is a DNA sequence variation that was used in the cited studies (Liberal Arts / Fine Arts, Post Baccalaureate)." This individual admits to not understanding the science but was able to provide an adequate definition of the term. This ability to recite definitions with no substance behind the understanding of those definitions is indicative of a lack of true understanding of the material. Further research should be completed in order to determine whether individuals who fall into this category of understanding actually believe that their knowledge is sufficient to make decisions.

Because the term SNP is used in the advertisement it would be prudent for consumers to understand the meaning of that term, however it is clear that the vast majority of this sample did not know that answer. It is standard convention to define acronyms whenever they are used in literature. It seems clear that given the importance of this

information it would be important to define acronyms such as SNP in a clear and readily identifiable manner.

7% of respondents failed to understand the fundamental difference between a positive and negative test result. This is a concern not just in this arena but also for medical reports in general. It also indicates that persons need results explained by someone who is able to ably communicate the meaning of test results. From a policy perspective this reinforces the idea that people may need a health professional to help them navigate these results. As a gatekeeper a physician may be able to help explain these results in an individual way which makes sure that every person understands what their result means specifically to them. If it is impractical for a physician with sufficient training and education regarding genetic testing to serve this purpose, it may be beneficial to utilize genetic counselor's to provide these services to patients.

Being "positive" and "negative" have different connotations in medical language and popular culture. In medicine being 'positive' means that a patient has a certain factor, trait, or pathogen. It is not necessarily a good or beneficial trait. Perhaps the best example of this would be in reference to HIV. Being HIV positive indicates that you have live virus particles in your body. This is, contrary to popular consensus, not a 'positive' or good state. The 'positive' test result counter-intuitively identifies a negative state. Over time the confusion regarding positive and negative in context has lessened but as we can see with genetic testing there may still be a significant portion of the population that confuses the popular understanding of 'positive' with the medical definition.

Overall, the results of this study indicate that there is a lack of understanding of the material presented in this manner. Attributing a cause for this is difficult but given that several authors have published concerns regarding this practice and the fact that the majority of respondents were well educated in related fields it is likely that there is a disconnect between what information is presented how it is interpreted. The result of this disconnect is a dangerous situation wherein individuals firmly believe they understand what a result means but are mistaken. This may lead them down a path of

unnecessary treatment and difficulties which may be preventable with better information.

From a policy standpoint it appears that individuals would support requiring a physician or other qualified professional to interpret results with a patient. In addition it may be necessary to regulate these advertisements in the same way prescription drug advertisements are regulated. While there are concerns that the actual information is useful, perhaps a less invasive approach would be to make sure that the information is comprehendible by a wide majority of consumers and require an unbiased professional be available for interpretation questions. This could be accomplished through a regulatory commission setting standards on what and how items can be presented or an independent third party assigned to review information with patients who receive these tests.

Precedent has been set for regulation in the pharmaceutical industry. Companies are required to give information regarding side-effects to consumers in their advertisements. One could argue that mental anguish can cause significant health issues on par with the numerous side effects that pharmaceutical companies are required to list (i.e. high blood pressure). While there is limited literature linking genetic tests to negative physical impact there is currently evidence and debate regarding this issue for a similar screening and non-invasive test.

Currently, a debate exists regarding prostate cancer screening utilizing prostate specific antigen (PSA) and the potential mental anguish it causes. Once a high PSA is found in a man he often proceeds to biopsies and further treatment, even when that treatment may not be considered beneficial. For 'peace of mind' many men elect to have surgery or radiation therapy when it may be in his best interest to not actively treat the cancer (Denenberg, Melhado, & Steiner, 2006). Genetic tests may lead to similar thinking. There have been instances where women have elected to have bi-lateral mastectomies when no cancer has been found but a genetic test has indicated that they are at a higher risk for breast cancer (Smith & Issacs, 2011). The test is not 100% predictive; therefore the elective mastectomies may be for naught. There are obviously dangers associated

with any surgery and choosing elective surgery without proper indication has the risk of leading to negative outcomes for the patient. Merely the risk of having a future potential health impact can lead some individuals to take unwarranted action. By ensuring that patients understand their genetic information completely it may be possible to limit some of these unnecessary and costly medical procedures. Because of the complexity of this information and the indirect quality of the side-effects it may be beneficial to have an independent third party review this issue with patients and help them decide whether or not to be tested for each trait they wish to test for.

While studies showing direct links between testing and poorer health outcomes are rare because of the multiple steps involved, the logical steps are clear. This research demonstrates and a clear disconnect between this population's understanding of genetics and clinical information. Further research needs to be done in order to verify and expand these results to other populations but this preliminary research suggests that there is clearly disconnect between what is presented and what consumers perceive.

Due to the personal nature of the subject, the medical implications and the continuing expansion of genetic knowledge the question of regulating people's genetics is a sensitive issue. That considered, there are various instances of local and national governments intervening on behalf of consumers, especially when health and welfare of individuals is involved. The primary example of this is the food and drug administration, which at a federal level protects consumers from false claims and misleading advertising from medical equipment and pharmaceutical companies (Food and Drug Administration, 2011). The findings presented here certainly support some type of intervention. It is clear that while consumers may believe they understand these complex relationships, a large percentage do not.

Because these tests are still primarily in their infancy, their usability in a clinical setting is unclear. We need to be certain that consumers understand the information presented. If someone chooses to obtain this information at a fair price then they deserve to fully understand it. The fact that this issue applies in a market where a person's health is at stake also raises several moral questions beyond this basic economic tenant. While not

addressed here, this should also be a consideration for government intervention. In cases where the market has failed to provide equality of information it is the government's role to ensure that all parties understand the transaction they are about to make. This study indicates that this relationship may exist and while further research needs to be completed it appears that there may be a need for intervention. Because the tests may have direct impacts on an individual's health the need is more immediate.

There are several limitations to this study which should be improved upon in further research. Because of the nature of the survey tool, respondents were able to look at the advertisement while answering questions. The result of this is that the answers may not have been a good indicator of the actual retention of information, but rather simple interpretation of what was available. Therefore the answers are not a good measure of retention. Future research should prevent respondents from viewing the form after a certain time in order to measure this. However, if individuals are unable to answer questions while they still have the information in front of them the likelihood of retention is diminished.

The nature of the survey itself is likely to have had an impact on the results. Allowing individuals to be able to review the advertisement while they answered questions does prevent us from understanding how much information was retained. One respondent did note that in a response saying "Just FYI as a respondent, I found myself going back to the advertisement after reading each question and doing a scavenger hunt for the answers to your questions, looking for information that I would not have necessarily looked for on my own. If you want to measure what people comprehend from advertisements, perhaps instruct the respondent to spend five minutes reading the advertisement then switching to a new page to answer comprehension questions… (Unknown, Post-Baccalaureate)." I would agree with this respondent; however what the survey does tell us is how much information people can garner while they still have the advertisement at their disposal. One could assume that if a respondent could not answer accurately when they still had the advertisement, the respondent would be less likely to answer correctly based on recollection.

Because respondents were allowed to take the survey on their own, there may be other impacts to the results. As previously noted, several respondents admitted to using an internet search to help them find answers. It is also possible that respondents had help from colleagues or other persons near them at the time they took the survey. The decision was made not to limit time or location of the survey in order to provide privacy and security to respondents. However, by not monitoring their activity it is possible that the answers provided are not an accurate reflection of each individual's true comprehension.

This sample was skewed toward highly educated individuals. As a result this may not be an accurate measure of the general population. However, if it is clear that a group of highly educated individuals has difficulty understanding the information it is likely that less educated groups may have similar if not more exaggerated difficulties.

In order to group answers coding exercises were completed on several questions. By codifying answers there is a certain resolution lost in analysis. Every effort was made to mention unique or specific answers in the discussion but it is likely that some specific knowledge is not accounted for here. All answers to free text questions (blinded and randomized) are presented in Appendix B.

The content of the survey instrument itself could be improved for future research. The wording of several questions and their order may be improved upon to glean different responses. Only one test was presented to the respondents and one test is certainly not indicative of all genetic testing. However, the test was chosen with care as a representative of a disorder with which many people are at least somewhat familiar and one that may have genetic and environmental components.

CONCLUSION

Necessarily, policy consistently lags behind technological innovations. It is impossible to determine the unintended consequences of a given technology before its widespread adaptation. There are often both positive and negative impacts of any new technology and it is unlikely that DTC genetic testing will be any different. That said, given the potential health impacts at both individually and at a population level, it is critical that we attempt to anticipate and mitigate any potential negative consequences of this technology. Ensuring consumers have the best possible knowledge is the first step in that process.

This research provides the first look at this issue and informs us that in a relatively well educated population there are discrepancies in the understanding of one particular advertisement. However there is still much work to be done in order to determine whether regulation is necessary at a national level. Outstanding issues include reviewing similar advertisements and websites with consumers, establishing the retention of the information by consumers and determining what political and economic tolerance may be for including gatekeepers in the process of obtaining genetic information. Further studies should focus on these topics to help determine if policies are required for these tests.

Genetic testing is currently being adopted by individuals but it may be that those individuals do not understand the implications of this new technology. It is clear that further research is needed to better understand how individuals interpret genetic testing advertisement. However, based on the information presented here, it appears reasonable that some policy measures be put in place to assist consumers in processing this information. Just as with prescription drugs it may be important to regulate not only what information is presented to the public, but also how that information is presented.

Currently, the FDA is moving towards some form of regulation in this arena. Letters have been sent to major manufacturers requesting changes be made to their advertising practices but to date, no serious interventions have been undertaken. Information similar to what is presented here has the opportunity to inform those decisions in a meaningful way. While this research is limited in scope, it does indicate that there is reason to move forward with larger and more complex studies in a similar manner. National policy measures requiring communication with a licensed professional may be the most palatable and effective step to regulating these tests.

At least in this population, there is a disconnect between an individual's perceived expertise and their true understanding of the information presented in this advertisement. Because this population has a higher average education than the general population we can presume that this trend may prove true or indeed worsen in a broader sample. In general it appears that this population believes they have a better understanding of the information than they actually do. In addition a definitive line needs to be drawn in terms of what patients actually need to know and at what level they need to understand this information. Given the major decisions that can be made based on the results of these tests, a fundamental misunderstanding of their meaning may be devastating to a patient's health in the long-term.

Whether or not government regulation is required in this arena is dependent upon a multitude of factors, including consumer understanding, consumer decision making, and health outcomes as a result of those decisions. Any break in this chain could result in a devastating impact to individual and public health. This research focuses on the first link in that chain and indicates that there is a need for further work. Without adequate patient understanding the other two links are necessarily impacted as misinformed decisions can be quite dangerous especially when it comes to a person's health. This research indicates that there is a good possibility that individuals who purchase these tests without further guidance are currently misinformed and would benefit from an increased regulatory structure requiring clear communication and personalized intervention from knowledgeable health care professionals.

WORKS CITED

23 and Me. (2011, February 21). Alcohol Dependence - Sample Report. New York, NY.

Boolel, M., Alan, M., & Ballard, S. (1996). Sildenafil; an orally active type 5 cyclic GMP-specific phosphodioesterase inhibitor for the treatment of penile erectile dysfunction. *International Journal of Impotency Research*, 47-52.

Caufield, T., Ries, N., Ray, P., Shuman, C., & Wilson, B. (2009, August 5). Direct-to-Consumer Genetic Testing: good, bad or benign? *Clinical Genetics*, pp. 1-5.

Cruikshank, J. (2010, 11 13). How they sell you what you don't understand. *The Washington Post*.

Denenberg, T., Melhado, T., & Steiner, J. (2006). Patient treatment preferences in localized porstate carcinoma: The influence of emotion, misconception and anecdote. *Cancer*, 620-30.

Folland, S., Goodman, A., & Stano, M. (2007). *The Economics of Health and Health Care*. Upper Saddle River, New Jersey: Pearson Education.

Gollust, S., Hull, S., & Wilfond, B. (2002). Limitations of Direct-to-Consumer Advertising for Clinical Genetic Testing. *Journal of the American Medical Association*, 1762-1767.

Government Accounting Office. (2006). Testimony before the Special Committee on Aging. Washington DC.

Government Accounting Office. (2010). Testimony before the Subcommittee on Oversight and Investigations. Washington DC.

Hewitt, J. (2002). A critical review of the arguments debating the role of the nurse advocate. *Journal of Advanced Nursing*, 439-45.

Hull, S., & Prasad, P. (2001, May). Reading Between the Lines: Direct-to-Consumer Advertising of Genetic Testing. *Hastings Center Report*, pp. 33-35.

Javitt, G., Stanley, E., & Hudson, K. (2004). Direct-To-Consumer Genetic Tests, Government Oversight, and the First Amendment: What the government can (and can't) do to Protect the Public's Health. *Oklahoma Law Review*, 251-324.

Kanfar, N., Louden, D., & Sircar-Ramsewak, F. (2007, March). FDA Direct-to-consumer advertising for prescription drugs: what are consumer preferences and response tendencies? *Health Marketing Quarterly*.

Marchionni, L., & Marinopoulos, S. (2008, January). Impact of Gene Expression Profiling Tests on breast Cancer Outcomes. *Evidence Report and Technology Assessment*.

McBride, C. M., Wade, C. H., & Kaphingst, K. A. (2010, September). Consumers' views of Direct-to-Consumer Genetic Information. *Genomics and Human Genetics, 11*, 427-446.

National Cancer Institute. (2005). *Surveillance Epidemiology and End Results.* Retrieved January 18, 2012, from SEER Cancer Statistics Review: http://seer.cancer.gov/csr/1975_2005/results_merged/sect_04_breast.pdf

National Society of Genetic Counselors. (2012). *National Society of Genetic Counselors*. Retrieved 1 18, 2012, from National Society of Genetic Counselors: http://www.nsgc.org/

Pence, G. E. (2011). *Medical Ethics.* New York, NY: McGraw Hill.

Pollack, A. (2010, 19 March). Consumers Slow to Embrace the Age of Genetics. *The New York Times*, p. B1.

Pollack, A. (2010, June 11). FDA Faults Companies on Unapproved Genetic Tests. *The New York Times*, p. B2.

Pollack, A. (2010, May 12). Walgreens Delays Selling Personal Genetic Test kit. *The New York Times*, p. B5.

Rahm, A. (2010). Public Opinion of Family History and Genetic Testing for Common Diseases: Results from Focus Groups with Kiaser Permanente COlorod Members. *Clinical Medicine and Research*, 190-191.

Rex, D. K., Johnson, D. A., Lieberman, D. A., Burt, R. W., & Sonnenberg, A. (2000). Colorectal Cancer Prevention 2000: Screening Recommendations of the American College of Gastroenterology. *The American Journal of Gastroenterology*, 868-877.

Rochester Institute of Technology. (2012). *Programs Of Study*. Retrieved 1 17, 2012, from RIT - Rochester Institute of Technology: http://www.rit.edu/overview/programs-study

Salas, H. S. (n.d.). *Life & Health Library.* Retrieved November 22, 2011, from IRB: Ethics and Human Research: http://findarticles.com/p/articles/mi_6824/is_4_30/ai_n31874569/pg_8/

Schomerusl, G., Lucht, M., Holzinger, A., Matschinger, H., Carta, M., & Angermeyer, M. (2010). The Stigma of Alcohol Dependence Compared with Ohter Mental Disorders: A Review of Population Studies. *Alcohol and Alcoholism*, 1-8.

Singer, N. (2010, July 22). Federal "Sting' Slams Gene Tests. *The New York times Prescriptions Blog*.

Smith, K., & Issacs, C. (2011). BRCA Mutation Testing in Determining Breast Cancer Therapy. *Cancer*, 492-9.

Theall, K., Scribner, R., Cohen, D., Bluthenthal, R., Schonlau, M., Lynch, S., et al. (2009). The neighborhood alcohol environment and alcohol-related morbidity. *Alcohol*, 491-499.

US Census Bureau. (n.d.). *Education.* Retrieved November 22, 2011, from US Census Bureau: The 2012 Statistical Abstract: http://www.census.gov/compendia/statab/cats/education.html

US Food and Drug Administration. (2011, April 1). *CFR - Code of Feder Regulations Title 21*. Retrieved 21 November, 2011, from U.S. Food and Drug Administration: http://www.accessdata.fda.gov/scripts/cdrh/cfdocs/cfCFR/CFRSearch.cfm?CFRPart=202&showFR=1

Williams, S. (2008). *Direct-to-Consumer Genetic Testing: Empowering or Endangering the Public.* Genetics and Public Policy Center.

My Surveys | Help | Welcome, Albert Blankley (logout)

Direct-to-Consumer Genetic Testing

You are invited to join a research study which looks at Direct-to-consumer (DTC) genetic testing. In this research study, we are investigating how people understand information presented to consumers by DTC genetic testing companies.

You will be asked to complete a short survey, in which you will be presented with a screenshot from a website and asked questions about the information presented, as well as some demographic questions and some questions relating to your background in biological science. We think this will take you between 5 and 10 minutes.

You may stop participating at any time. If you decide to leave the study simply close the window in which the survey is presented.

CONFIDENTIALITY

Your name will not be used when data from this study are published. All survey results are anonymous and are not linked to a specific user. Every effort will be made to keep all personal information confidential. As the primary investigator, only Albert Blankley will have access to the data and there is no link between who responded to the survey and the specific answers you provide.

Participation in this study is voluntary. You have the right not to participate at all or to leave the study at any time. Deciding not to participate or choosing to leave the study will not result in any penalty or loss of benefits to which you are entitled.

If you have any questions or concerns please feel free to contact me at aab8038@rit.edu.

Alcohol Dependence - Genetic Testing - 23andMe

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Alcohol Dependence - Sample Report

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Example Data

About Alcohol Dependence

Alcohol dependence is a complex disorder that combines alcohol abuse – continued drinking despite negative consequences – with physical symptoms of tolerance, withdrawal and uncontrollable craving for alcohol. The heritability of alcohol dependence is estimated to be between 38% and 64%. This means genetic and environmental factors contribute varying amounts to risk for this condition, probably depending on the population in question. Environmental factors that may increase risk include a family history of alcohol or drug abuse, having peers who exhibit common alcohol abuse, being male, cultural background, psychiatric disorders such as anxiety or depression and drinking at an early age. Genes that may play a role in alcohol dependence include those that affect brain chemistry and the liver's ability to process alcohol.

Alcohol dependence in men

Journal	*Arch Gen Psychiatry*
Study Size	
Replications	None
Contrary Studies	None
Applicable Ethnicities	European
Marker	rs7590720

In this study, researchers compared 1,460 men with clinically-diagnosed alcohol dependence to 2,332 men without alcohol dependence, all of European ancestry. They found that each copy of a G at rs7590720 was associated with about 1.35 times higher odds of alcohol dependence.

Who	Genotype	What It Means
	GG	Moderately higher odds of alcohol dependence in men.
Greg Mendel (Dad)	AG	Slightly higher odds of alcohol dependence in men.
	AA	Typical odds of alcohol dependence in men.

Citations

Treutlein J et al. (2009) . "Genome-wide association study of alcohol dependence." *Arch Gen Psychiatry* 66(7):773-84.

Alcoholism

Journal	*Mol Psychiatry*
Study Size	
Replications	None
Contrary Studies	None
Applicable Ethnicities	European, Asian
Marker	rs1800497

Who	Genotype	What It Means
	AA	Slightly increased odds of alcoholism.
Greg Mendel (Dad)	AG	Slightly increased odds of alcoholism.

https://www.23andme.com/health/Alcohol-Dependence/[4/19/2011 8:36:07 AM]

This analysis combined data from a number of previous studies that looked at the correlation between rs1800497 and alcoholism in a total of more than 10,000 people. The results of those studies may have varied in part because alcoholism has multiple definitions that are similar to, but not identical to, alcohol dependence. But taken together, the combined studies indicated that each copy of A at rs1800497 increased a subject's odds of alcoholism by about 1.2 times. (Note: this SNP is commonly referred to in the literature as the DRD2 TaqIA polymorphism. Having a copy of the DRD2 TaqIA A1 allele is equivalent to having an A at rs1800497, while having the A2 allele is equivalent to having a G.)

GG	Typical odds of alcoholism.

Citations

Munafò MR, Matheson IJ, Flint J (2007) . "Association of the DRD2 gene Taq1A polymorphism and alcoholism: a meta-analysis of case-control studies and evidence of publication bias." *Mol Psychiatry* 12(5):454-61.

Alcohol withdrawal seizures

Journal	*Alcohol Clin Exp Res*
Study Size	
Replications	None
Contrary Studies	None
Applicable Ethnicities	European
Marker	rs27072

Withdrawal seizures are one indication of alcohol dependence, though their absence does not necessarily rule out that diagnosis. This study compared 60 alcohol-dependent subjects who had withdrawal seizures to 190 alcohol-dependent subjects who did not. The authors found that alcohol-dependent people with at least one T at rs27072 had about half the odds of experiencing withdrawal seizures compared to those with none.

Who	Genotype	What It Means
Greg Mendel (Dad)	CC	Typical odds of alcohol withdrawal seizures (if alcohol-dependent).
	CT	Moderately lower odds of alcohol withdrawal seizures (if alcohol-dependent).
	TT	Moderately lower odds of alcohol withdrawal seizures (if alcohol-dependent).

Citations

Le Strat Y et al. (2008) . "The 3' part of the dopamine transporter gene DAT1/SLC6A3 is associated with withdrawal seizures in patients with alcohol dependence." *Alcohol Clin Exp Res* 32(1):27-35.

Demographics

1. What is your age?

2. What is your sex?

- (•) Male
- ( ) Female
- ( ) Other

3. Education (please select one):

- (•) Some High-school
- ( ) Graduated High-school
- ( ) Attended some college
- ( ) Associate's degree
- ( ) Bachelor's degree

4. If you have a received a college education (associate's or more) please indicate your major

5. What is your highest terminal degree?

6. What is your current occupation?

7. Ethnicity

- (•) African American
- ( ) Asian / Pacific Islander
- ( ) Caucasian
- ( ) Latin / South American
- ( ) Native American
- ( ) Other:

Interpretation Questions

8. What are your impressions of alcohol dependency and genetics

9. How many studies are described by this advertisement?

10. If you had a family member who had a dependency on alcohol would you be interested in getting a genetic test for yourself?

- ⊙ Yes
- ○ No

11. If you had a blood relative who had a dependency on alcohol would you be interested in getting a genetic test for yourself?

- ⊙ Yes
- ○ No

12. What is the relationship between marker RS27072 and alcohol dependency?

13.

	Not Closely At All (1)	Not Very Closely (2)	Somewhat Closely (3)	Fairly Closely (4)	Very Closely (5)
How closely are genes and alcohol dependency related?	⊙	○	○	○	○

14. Does ethnicity have an impact on this relationship?

- ⊙ Yes
- ○ No

15. If so, how?

16.

	Very uninterested	Not Interested	Neutral	Somewhat Interested	Very interested
How interested are you in this condition?	⊙	○	○	○	○

17. Approximately how many people's genetics were studied to determine the relationship between marker RS18001197 and alcohol dependency?

- ⊙ 10
- ○ 100
- ○ 1000
- ○ 10000

18. What would you do if you received a test result that indicated you had this gene?

- ☑ Nothing
- ☑ Seek more information on the condition on the internet
- ☑ Talk to my doctor about my risks
- ☑ Call the company to speak to a genetic counselor

19. Where did you receive the majority of the information?

- ⊙ Charts
- ○ Graphs
- ○ Numbers
- ○ Text
- ○ Photos

Science Background

20. What is an established research report?

21. What journals did the research stem from?

22. What is a SNP?

23. Susie requested that this analysis be done for her. The result stated that she had an increased risk for alcohol dependency. The letter from the lab said that her test was _____.

- (•) Positive
- ( ) Negative