Personal Genome Tests: Consumers’ Motivations and Perspectives

Master’s Thesis

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Barbara Lerner, Advisor

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by
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And finally, I have to thank my family and husband: Without their love and support, I do not know how I would have made it to this point. I love you all!

Thank you!
ABSTRACT

Personal Genome Tests: Consumers’ Motivations and Perspectives

A thesis presented to the Department of Biological Sciences, Program in Genetic Counseling
Graduate School of Arts and Sciences
Brandeis University
Waltham, Massachusetts

By Erica Lynn Blouch

The future applications of genetic testing have expanded with the completion of the human genome project. Researchers have discovered genetic polymorphisms that predispose individuals to common diseases, such as diabetes, glaucoma, and prostate cancer. Recently, several companies have begun offering personal genome testing (PGT) for predisposition to common diseases. The aim of this study was to explore customers’ motivations for pursuing personal genome testing and their experiences regarding PGT, including their health behavior changes since receiving their results. We recruited PGT customers as study participants using a newsletter and email distribution provided by Navigenics, a commercial laboratory offering PGT services. Respondents were invited to complete an anonymous, online survey consisting of both multiple choice and open-ended questions. Of the 98 survey respondents, most (85.7%) said that the PGT process met their expectations and that the results were useful in making medical decisions (78.1%). Just over half of the respondents (54.8%) reported making behavioral changes after receiving their PGT results. Of these, lifestyle changes, such as diet and exercise, were the most often cited. The most common motivating factor for PGT was, “to define my chance of developing any common disease” (77.1%). Curiosity was the second most common motivating factor (67.5%). Individuals that reported having children
were more likely to be motivated by a wish to define risk to family than individuals who reported having no children. Our study population represents a small subset of PGT customers from a single laboratory. Despite hesitation in the medical and genetics community regarding the clinical utility of PGT, our findings suggest that many of these individuals have a positive perception of PGT. Future research to further explore the role of genetic counselors and physicians in the PGT process could provide additional insight into the consumer perspective.
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Introduction

The future of genetic testing has expanded with the completion of the human genome project. Previously, researchers and physicians in the field of genetics focused on rare diseases, such as cystic fibrosis or Huntington’s disease, which are caused by mutations in a single gene. In single-gene disorders, having a disease-causing mutation generally correlates with disease development. As a result, these mutations can be tracked in a family to see who will become symptomatic, and who will not.

Researchers have now discovered genetic variations that predispose an individual to common diseases, such as diabetes, glaucoma, macular degeneration, Crohn’s disease, and prostate cancer. (Barrett, et al., 2008; Haiman, et al., 2007; Maller, et al., 2006; Saxena, et al., 2007; Thorleifsson, et al., 2007) Having these polymorphisms does not mean an individual will definitely develop the disease, but rather, having the polymorphism only increases the individuals’ chances. The single-nucleotide polymorphisms (SNPs) that predispose to common diseases are fairly common in the general population, and their relative risk is sometimes quite small when compared to mutations that cause Mendelian genetic syndromes. (Altshuler, et al., 2008) As a result, these conditions are considered multifactorial, and environment or lifestyle will also play a role in disease development. Currently, the greater genetics community debates the utility of such information. (Altshuler, et al., 2008; McBride, et al., 2010; Scheunier, et al., 2008) However, the hope is that by knowing an individuals’ genetic predisposition for disease, and factoring in environment or lifestyle choices, medical professionals can further define a person’s risk for a common disease. If the patient is then considered to be at high-risk as compared to the general population, methods for disease
prevention can be discussed. This is one goal of personalized genomic medicine. (Ginsburg & Willard, 2009)

Companies offering PGT

In the past few years, several companies have begun offering personal genome testing (PGT) for predisposition to common diseases using SNP analysis. Navigenics and 23andme are currently the predominant players in the field of personalized genomics. Consumers can order PGT on their own via direct-to-consumer (DTC) testing or, in the case of Navigenics, PGT can be ordered through their doctor. In the general DTC model, the consumer orders testing from the company website. They are then sent a salivary DNA kit for home collection and mail the completed kit back to the company for analysis. After the SNP testing has been completed, the consumer logs on to the company website to view their individual risk analysis for the panel of common diseases. Depending upon which company is performing the testing, variations in this model are seen. For example, 23andme will not only test for common disease polymorphisms, but for “novelty traits” as well. These include earwax type and bitter taste perception. Another variation on this model is seen at Navigenics, which employs genetic counselors that are available to counsel, educate, and interpret test results to consumers via telecounseling. In addition, Navigenics partners with multiple healthcare organizations (for example: Cleveland Clinic, Scripps Genomic Medicine, MDVIP) to bring the personal genome test directly to consumers via their physician. Navigenics’ primary product, the Health Compass, currently costs $999 for the initial analysis of 28 common diseases (Table 1). In addition, in April 2010, Navigenics began to offer pharmacogenetic testing for response to 11 medications. The initial fee also includes consultations with their genetic counselors and updates for one year, as new research is available. After one year,
individuals can purchase an annual subscription for $199, which ensures that the updates and additional testing continue.

Table 1: List of common diseases on Navigenics Health Compass panel.

<table>
<thead>
<tr>
<th>Abdominal aneurysm</th>
<th>Crohn's disease</th>
<th>Lactose intolerance</th>
<th>Osteoarthritis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alzheimer's disease</td>
<td>Deep vein thrombosis</td>
<td>Lung cancer</td>
<td>Prostate cancer</td>
</tr>
<tr>
<td>Atrial fibrillation</td>
<td>Diabetes, type 2</td>
<td>Lupus</td>
<td>Psoriasis</td>
</tr>
<tr>
<td>Brain aneurysm</td>
<td>Glaucoma</td>
<td>Macular degeneration</td>
<td>Restless legs syndrome</td>
</tr>
<tr>
<td>Breast cancer</td>
<td>Graves' disease</td>
<td>Melanoma</td>
<td>Rheumatoid arthritis</td>
</tr>
<tr>
<td>Celiac disease</td>
<td>Heart attack</td>
<td>Multiple sclerosis</td>
<td>Sarcoidosis</td>
</tr>
<tr>
<td>Colon cancer</td>
<td>Hemochromatosis</td>
<td>Obesity</td>
<td>Stomach cancer, diffuse</td>
</tr>
</tbody>
</table>

Professional attitudes towards DTC genomic testing

In general, the concept of direct-to-consumer genomic testing has not received a lot of support from the genetics community. (Haga, et al., 2003; Hunter, et al., 2008) On the positive side, the analytic validity is considered to be high, meaning that errors are unlikely during the testing process. (Hudson, et al., 2007) On the other hand, both the clinical validity and clinical utility are often questioned. (Scheuner, et al., 2008) Clinical validity refers to the diagnostic or predictive value of the test results: What are the chances that this person will develop the disease? Clinical utility is an analysis of the benefits: Now that the risk information is known, what can be done? In these two respects, there is not yet enough data for many healthcare professionals to be convinced that personal genome testing in the DTC setting is a good idea.

In the last few years, the major professional organizations have released position statements in regards to direct-to-consumer testing. (Note: These do not relate to personal genomic testing when in the clinical setting.) A 2008 statement from the American College of Medical Genetics describes their minimum requirements for a proper genetic testing protocol: First, the process should involve a knowledgeable professional. Second, there
should be informed consent about the benefits and limitations of the testing. Third, the scientific evidence of the test should be clearly stated. And finally, testing should be performed in an accredited laboratory. (ACMG, 2008) Similarly, a 2007 release from the American Society for Human Genetics made three recommendations to its members and the genetics community regarding DTC testing. (Hudson, et al., 2007) First, companies that offer genetic testing DTC should promote transparency by disclosing pertinent information in an accessible format, including, the analytic validity of the tests, the clinical utility, the risks associated with testing, CLIA certification status, privacy policies, and evidence for any recommended interventions. The second recommendation from ASHG is to professional societies, requesting that they be aware of what tests are available DTC and educate their members appropriately. Thirdly, ASHG made a request to the appropriate governmental bodies for proper regulation of genetic testing. Also in 2007, the National Society of Genetic Counselors released a statement regarding DTC genetic testing and outlined a series of questions for consumers to consider before proceeding with DTC testing. (NSGC, 2007) The position statement encourages potential consumers to do thorough research on the companies’ policies and practices. In their opinion this should include assessing the manner by which the information is presented and asking if the testing company encourages test result discussion with their doctors or family members. Also, the consumer should understand the privacy policy and informed consent procedures of the company. In summary, these professional societies are concerned about both the medical implications and ethical standards of DTC testing.
Who gets genetic testing and why?

As this study will examine demographics and attitudes of current PGT consumers, it is necessary to understand what is known about genetic testing in other scenarios. A 2004 survey by the Genetics & Public Policy Center at Johns Hopkins University reported that 88.5% of US citizens have heard of genetic testing for reproductive purposes. (Kalfoglou, et al., 2004) Those who have heard of genetic testing were more likely to have at least some college education and to make more than $50,000 per year. A study published in 2009 found that individuals who would recommend genetic testing to a friend, used as a proxy for positive attitude toward genetic testing, were also likely to undergo genetic testing themselves. (Barnoy, et al., 2009)

It is also important to examine why patients decide to have genetic testing in both the traditional medical setting, as well as the DTC setting. In studies of genetic testing for cancer predisposition, the most common motivator for genetic testing is to gain information for one’s offspring. (Esplen, et al., 2007; Lynch, et al., 1997) These studies also report that personal medical management (increased screening, prophylactic surgery, etc.) is another major reason for genetic testing in the cancer setting. A survey of individuals interested in genetic testing for predisposition to bipolar disorder revealed slightly different results. (Meiser, et al., 2008) In this case, the main reason that survey participants were interested in genetic testing was for better definition of their own risk and for determining treatment options. In contrast to the cancer studies discussed above, the genetic risk to children was a lower priority in the study of testing for bipolar disorder. Similar results can be seen in studies of genetic testing for Alzheimer’s diseases, where common reasons for genetic
susceptibility testing included making the proper arrangements for the future and coping with the increased threat. (Roberts, et al., 2005)

Very little has been published regarding the interest in personalized genome testing or the actual uptake of genomic testing. Initial published reports examined nutrigenomic testing, which is available DTC. This testing involves genetic analysis to provide a personalized health and nutrition report, informing at-risk individuals of healthy food choices, while also marketing the company’s own brand of vitamins. In 2007, it was reported that 17% of the US population was aware of DTC nutrigenomic tests. (Goddard, et al., 2007) These persons were more likely to be younger than 55 years of age, to have more than a high school education, and to have heard of it in the media. Twenty-nine individuals (0.6%) reported using a DTC nutrigenomic test. A more recent study examined nutrigenomic testing when offered through a physician. (Stewart-Knox, et al., 2009) Thirty-nine percent of the 5000 respondents would have a genetic test for general interest while only 28% would have the test to follow a personalized diet. Persons over the age of 65 were more likely to answer in the affirmative for these questions. When asked about reason for undergoing nutrigenomic testing, ‘risk to children’ was not reported as a concern for this population.

A study published in 2009 attempted to characterize attitudes held by individuals that would undergo PGT when it was offered free-of-charge. (McBride, et al., 2009) The authors surveyed and offered PGT to approximately 2000 adults aged 25-40, who were members of a participating managed care organization. The PGT that was offered examined genetic variants involved in eight common diseases: type 2 diabetes, coronary heart disease, hypercholesterolemia, hypertension, osteoporosis, and lung, colon, and skin cancer. The authors reported that about 16% of the original population ultimately had their blood drawn
for testing. They found that individuals interested in PGT tended to feel confident that they would understand the results and the genetic information given to them. In addition, this population consisted of internet-users and persons who believed genetics were a part of understanding overall health. However, these individuals did not have an overly deterministic view of genetics and felt that they had behaviors that could be changed in order to lower risk.

In a survey of individuals in the Coriell Personalized Medicine Collaborative (CPMC), presented data shows that 97% of individuals feel learning about disease risk is a benefit of PGT. (Gordon, et al., 2009) Other benefits were using the results as motivation for behavior change (90%), knowing which medications to avoid (73%), and defining disease risk to children (49%).

*Behavior changes after genetic testing*

One of the arguments against the current PGT offerings is the value of the medical information and ability to make meaningful behavior changes to reduce disease risk. (McBride, et al., 2010) Some studies examine what individuals do with the information they receive from genetic test results. It is believed that in the case of highly-penetrant, single-gene disorders, individuals who test positive are likely to follow through with changes to medical management or increased screening. This can be seen in hereditary breast/ovarian cancer syndrome. (Peshkin, et al., 2002) In this study, it was found that in the year following a positive BRCA1/2 result disclosure, 74% of women aged 40 or older had a mammogram. However, studies regarding genetic predisposition to lung cancer and behavior changes have not been as straightforward. One study showed no difference in smoking cessation rates between individuals who were informed they had a high genetic predisposition (GSTM1-missing genotype) to lung cancer, versus those who had a low predisposition (GSTM1-
present genotype). (Sanderson, et al., 2009a) In fact, individuals who had the high-risk genotype felt they were less likely to change their lung cancer risk, even if they stopped smoking. Alternatively, a hypothetical study of genetic test results and the risk of developing obesity showed a different result. (Sanderson, et al., 2009b) The authors reported an increase in participants’ motivation to avoid obesity when individuals were given a hypothetical high-risk test result. A recent review article addressed the impact of genetic test results on behavior modification. (McBride, et al., 2010) Citing the studies above and others, the authors concluded that “personalized genetic information has its greatest impact on behavior when the disease risks are appreciable.”

**Study aim**

Currently, there is limited information in the medical literature regarding the consumer’s perspective of PGT. The aim of this study was to determine the attitudes of a personal genomics company’s customers towards PGT and their motivations for pursuing PGT. Specifically, we asked individuals why they decided to have this testing, their opinions about its clinical utility, whether it has led to any behavioral changes, and whether they are satisfied with the information that they received with their results. The data gathered in this exploratory study will better prepare the genetics community for meeting the needs of this population in the future, while also improving the quality of PGT services.
Methods

Study Design

This was a cross-sectional study of Navigenics customers that utilized a self-administered, online survey assessing the attitudes towards PGT and respondents’ motivations for pursuing this testing.

Participant Recruitment

All potential participants are customers of Navigenics and were given the same recruitment notice. (Appendix A) The recruitment notice introduced the aim of this research project, outlined the requirements for participation, and described the compensation offered for participation. Compensation was the opportunity to enter a raffle for one of two Navigenics subscriptions, which provide the individual with one-year of updated risk information as new research becomes available. The recruitment notice informed individuals that participation is anonymous and voluntary, and therefore, the decision to participate cannot affect the individual’s relationship with Navigenics. Distribution of this notice occurred in two ways as outlined below.

Many Navigenics customers have PGT ordered through their doctors’ office. A subset of these individuals received an electronic-mailing with the recruitment notice directly from one of the genetic counselors at Navigenics. The second method of recruitment was through the monthly Navigenics electronic-newsletter that is sent to every Navigenics customer. This newsletter contained a variety of articles from Navigenics, including a copy of the recruitment notice. It is possible that some potential participants were contacted via both recruitment methods.
Data Collection

All participants completed the same survey at surveymonkey.com regardless of how they were recruited. (Appendix B) The survey contained 47 questions, subdivided into 3 sections. Section I collected demographic information in order to compare this sample to both the general population and to previously published results regarding genetic testing. Section II of the survey asked participants about their opinions and attitudes towards genetics and healthcare. Section III asked participants questions that relate specifically to their PGT experiences and included closed- and open-ended questions. The survey was open for a total of 8 weeks. This study was approved by the Institutional Review Board of Brandeis University, Waltham, Massachusetts. (IRB Protocol # 10073)

Data Analysis

The data were downloaded into SPSS (PASW Statistics, v17.0, for Mac) for analysis. The frequency of responses for each closed-ended questions was calculated. Correlations were carried out using bivariate analysis methods including chi square tests and independent samples t-tests. Open-ended questions were analyzed using qualitative methods to identify themes.
Results

Response rate and demographics

The recruitment notice was sent to 2879 individuals via the direct email or newsletter forms. Of those, Navigenics was able to track that 1152 (40%) individuals opened the emails containing either the recruitment notice or the Navigenics newsletter. Of the 2879 individuals, 98 participants completed survey for a response rate of 3.4%.

After reviewing the data, it became apparent that certain respondents had not yet received their PGT results. Responses pertaining to thoughts or actions after reviewing PGT results were discarded for these individuals. However, demographics and responses regarding motivation for PGT were included in the data analysis, after determining that there was no statistical difference between these responses based on having received results.

Demographic data is summarized in Table 2. The greatest majority of the respondents are 50 years or older, white, well educated, and with an annual household income of greater than $200,000. The average number of biological children in this group was 1.44 per respondent. Just less than 35% of the population had no biological children. Three individuals reported being adopted, equaling 3.1% of the sample.

Interestingly, there was an excess of female respondents (female = 62.5%; male = 37.5%). This is statistically different from the general population of the United States ($X^2=5.169, p=.023$) and from the total Navigenics population ($X^2=8.087, p=.004$).
TABLE 2: Demographic characteristics of PGT survey participants.

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Frequency (N)</th>
<th>Percentage (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Female</td>
<td>60</td>
<td>62.5%</td>
</tr>
<tr>
<td>Male</td>
<td>36</td>
<td>37.5%</td>
</tr>
<tr>
<td>Age (yrs)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>18-29</td>
<td>1</td>
<td>1%</td>
</tr>
<tr>
<td>30-39</td>
<td>8</td>
<td>8.2%</td>
</tr>
<tr>
<td>40-49</td>
<td>13</td>
<td>13.4%</td>
</tr>
<tr>
<td>50-59</td>
<td>25</td>
<td>25.8%</td>
</tr>
<tr>
<td>60-69</td>
<td>41</td>
<td>42.3%</td>
</tr>
<tr>
<td>70+</td>
<td>9</td>
<td>9.3%</td>
</tr>
<tr>
<td>Race</td>
<td></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>93</td>
<td>94.9%</td>
</tr>
<tr>
<td>African American</td>
<td>1</td>
<td>1.0%</td>
</tr>
<tr>
<td>Hispanic/Latino</td>
<td>1</td>
<td>1.0%</td>
</tr>
<tr>
<td>Mixed Ethnicities</td>
<td>3</td>
<td>3.1%</td>
</tr>
<tr>
<td>Highest education</td>
<td></td>
<td></td>
</tr>
<tr>
<td>completed</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Some high school</td>
<td>0</td>
<td>0.0%</td>
</tr>
<tr>
<td>High school diploma/GED</td>
<td>13</td>
<td>13.3%</td>
</tr>
<tr>
<td>Two-year degree</td>
<td>7</td>
<td>7.1%</td>
</tr>
<tr>
<td>Four-year degree</td>
<td>35</td>
<td>35.7%</td>
</tr>
<tr>
<td>Graduate/Professional degree</td>
<td>43</td>
<td>43.9%</td>
</tr>
<tr>
<td>Ann. Household Income</td>
<td></td>
<td></td>
</tr>
<tr>
<td>&lt;$25,000</td>
<td>1</td>
<td>1.2%</td>
</tr>
<tr>
<td>$25,000-49,999</td>
<td>4</td>
<td>4.8%</td>
</tr>
<tr>
<td>$50,000-99,999</td>
<td>17</td>
<td>20.5%</td>
</tr>
<tr>
<td>$100,000-149,999</td>
<td>9</td>
<td>10.8%</td>
</tr>
<tr>
<td>$150,000-199,999</td>
<td>18</td>
<td>21.7%</td>
</tr>
<tr>
<td>$200,000 or more</td>
<td>34</td>
<td>41.0%</td>
</tr>
<tr>
<td>Biological Children</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>64</td>
<td>65.3%</td>
</tr>
<tr>
<td>Adopted</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>3</td>
<td>3.1%</td>
</tr>
<tr>
<td>Received PGT results</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>88</td>
<td>89.8%</td>
</tr>
</tbody>
</table>

Beliefs regarding genetics and healthcare

In order to assess an individual’s understanding of genetic contribution to disease, participants were asked to rank a set of diseases on a scale of 1 to 5, with 1 being “genetics plays no role” and 5 being “genetics is the only factor”. (Figure 1) Of the 14 diseases, 11 are considered to be “common diseases” with a small genetic basis. Most of these common diseases are available for testing from the major personal genomics companies, including Navigenics and 23andme. In addition to the common diseases, three diseases have a Mendelian model of inheritance: Cystic fibrosis, Tay-Sachs disease, and sickle cell disease.
were included in the list. On average, individuals believed sickle cell disease to be the most “genetic” of the diseases (mean = 4.21, s.d. = 0.85) and obesity to be the least genetic (mean = 2.88, s.d. = 0.869). Across all 14 diseases, the mean overall score was 3.3. See Figure 1.

**Figure 1**: Beliefs regarding genetic contribution across all diseases as measured on 5-point scale. On this scale, “1” was defined as “genetics plays no role”, and “5” was defined as “genetics is the only factor”. Common diseases are in blue and Mendelian diseases are in yellow. The standard deviation for each answer is shown.

The researchers created a summary score for each respondent in order to assess individual differences in answering the “genetic contribution” question. The summary score was calculated by averaging the respondents’ answers for only the 11 common (non-Mendelian) diseases. This summary score is meant to represent how strongly an individual feels genetics contributes to common diseases. The mean summary score was 3.1 (st. dev. = 0.5, median = 3.2). Therefore, individuals tended to believe that common diseases were due to genetic factors rather than non-genetic factors. The distribution of summary scores is shown in Figure 2.
Figure 2: Genetic contribution summary score per respondent. This score was calculated by averaging each individual’s answers to the 11 common diseases shown in blue in Figure 1. This summary score represents how strongly individuals feel genetics contributes to common diseases, with a lower score indicating a belief of lower genetic contribution.

Respondents were given a variety of statements regarding genetics and their personal healthcare beliefs and asked to answer how strongly they agree with the statements on a 5-point Likert scale ranging from “strongly disagree” to “strongly agree”. (Table 3) The average answer for all statements was 4.13 (essentially agreed with the statements) indicating that individual felt they and their doctors were comfortable with genetic information and that genetic predisposition was an important part of medical care.

Table 3: Statements regarding genetics and healthcare beliefs. Answered on a 5-point Likert scale, with “1” defined as “strongly disagree” and “5” defined as “strongly agree”. The average answer (and standard deviation) to each statement is shown.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Mean</th>
</tr>
</thead>
<tbody>
<tr>
<td>I understand basic genetic principles.</td>
<td>3.95 (sd - 0.78)</td>
</tr>
<tr>
<td>I understand all of the risk information provided in personalized genome test results.</td>
<td>3.50 (sd - 0.80)</td>
</tr>
<tr>
<td>It is important to know my genetic predisposition to common diseases.</td>
<td>4.24 (sd - 0.62)</td>
</tr>
<tr>
<td>It is important to share my genomic test results with my doctor.</td>
<td>4.24 (sd - 0.87)</td>
</tr>
<tr>
<td>I feel my doctor understands my genomic test results.</td>
<td>4.08 (sd - 1.03)</td>
</tr>
<tr>
<td>I am an active participant in my healthcare management.</td>
<td>4.63 (sd - 0.51)</td>
</tr>
<tr>
<td>Understanding my genetic predispositions to common disease will change my medical care management.</td>
<td>4.15 (sd - 0.75)</td>
</tr>
</tbody>
</table>
**Personal experiences prior to testing**

Most participants in this study (53%, n=43) had heard of PGT through their physician. (Figure 3) The media was the second most common way they learned about PGT (27%, n=22). Only nine of the respondents had considered a PGT through a different company before choosing Navigenics (10.8%). Of those, seven individuals had considered testing with 23andme, one individual previously had testing through Genova Diagnostics, and one individual did not disclose which company they had considered.

**Figure 3:** Pie chart showing where individuals first heard of personal genome testing. Question was asked in open-ended response format and coded by the researchers into six categories. (n=81)
Seven participants (8.5%) did not discuss their intentions with anyone prior to purchasing the personal genome test. The remaining 75 participants discussed these intentions with a variety of individuals. The most common response (69.3%) was spouse or partner. About half (53.3%) of participants discussed their intention with their physician. Only 10.7% talked to their children about their decision. Some respondents discussed this with their siblings (4.0%), a parent (6.7%), other family members (10.7%), a friend (16.0%), or a physician besides their PCP (2.7%).

**Personal experiences after receiving results**

Participants were asked which members of the medical community they discussed their PGT results with. 28.8% (n=21) of the respondents consulted one of the genetic counselors at Navigenics. These conversations always involved interpretation of the test results and often (42.9%) involved discussion of family history. (Figure 4a) Two individuals responded that they had discussed their PGT results with geneticist or genetic counselor that was not employed by Navigenics. However, both individuals had training in genetics and/or genomics and interpreted the test result themselves. 75.3% (n=55) of the participants discussed their PGT results with their physician. Most of the time (94.5%), test result interpretation was discussed. In many cases (65.5%), physicians discussed medical management of diseases. In contrast, only 19% of conversations with Navigenics genetic counselors discussed medical management. All in all, 55.6% (n=40) of respondents talked to their doctor about their results, 11.1% (n=8) talked to a Navigenics genetic counselor, 18.1% (n=13) talked to both, and 15.3% (n=11) talked to neither. (Figure 4b)
Figure 4a: In multiple response format, individuals were asked what information they discussed with either the Navigenics genetic counselor (n=21) or with their physician (n=55) after receiving their PGT results.

![Bar chart showing discussion topics](chart1.png)

Figure 4b: Percent of respondents that discussed their PGT results with their physician (n=40), a Navigenics GC (n=8), both (n=11), or neither (n=13).

![Pie chart showing discussion partners](chart2.png)
Participants were asked if they had shared their PGT results with any family members. 13.7% had not shared this information (n=10). Of those who did, most shared their results with their spouse or partner. (Figure 5)

**Figure 5:** After receiving PGT results, individuals shared their results with friends and family as shown below. (n=63). Ten individuals (13.7%) did not share their results with any friends or family.

On a 5-point Likert scale, 84.9% of respondents (n=62) indicated that they were “satisfied” or “very satisfied” with their PGT results. Similarly, 85.7% (n=60) said that the PGT process met their expectations. The follow-up, open-ended responses revealed three main reasons for dissatisfaction. First, some individuals felt that the Navigenics disease panel was lacking diseases that were of high interest to them, such as *BRCA1/2* testing or pharmacogenomic studies. Secondly, some individuals were disappointed because they felt the results were not definitive enough or they felt uncertain about what the results meant. Lastly, a few individuals had administrative concerns such as sample mix-up, or not receiving their results in timely manner.
Participants were asked if they had changed any medical or lifestyle behaviors since receiving their PGT results. About half (54.8%, n=40) of respondents said they changed some behaviors. The researchers coded responses to the follow-up, open-ended question as either changes to medical management, lifestyle changes, or both. (Figure 6) Of the 40 individuals that reported making a change, 10% of individuals changed their medical management; 70% changed an aspect of their lifestyle; and 15% changed both. Two individuals (5%) did not report what type of change they made. The overall picture of changes that were or were not made is shown in Figure 7.

**Figure 6:** Bar graph showing types of changes individuals have made after receiving their PGT results. Question was asked in open-ended response format and coded by the researchers into multiple-response categories. N=40.

![Bar graph showing types of changes individuals have made after receiving their PGT results.](image)

Participants were then asked on a 5-point Likert scale, how useful they felt their PGT results were in making medical decisions. Fifty-seven respondents (78.1%) felt their results were “somewhat useful” or “very useful” in this respect. 14 individuals responded “neutral” (19.2%) and 2 responded “not that useful” (2.7%). There were no responses for “not useful at all”. Eight individuals gave reasons for why they answered “neutral” or “not that useful”.

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Five of the eight were because individuals were not at high risk for any of the diseases. Two respondents felt they still needed a better understanding of the results, and one person said their physician would not look at the results.

**Figure 7:** Overall picture of changes that have been made after receiving PGT results. N=73.

![Bar chart showing changes made after receiving PGT results.](image)

**Motivations for pursuing personal genome testing**

Participants’ motivations for purchasing a PGT are in Figure 8. In this multiple response question, the researchers asked respondents to choose their primary motivation(s). Of the 84 individuals that answered this question, 77.1% wanted the testing in order to define their chance of developing any common disease. 67.5% of individuals pursued testing to satisfy their curiosity.

An independent-samples t-test was used to examine differences in motivations for testing between individuals who reported having biological children, and those who did not. There was a significant, positive relationship between having children and choosing “define disease risk to my family” as a motivation (t(81)=3.222, p=.002). The same relationship was
seen with “concerned about something in my family history”. \( t(74.457)=2.270, p=.026 \)

Alternatively, there was a significant, negative relationship between having children and choosing the responses “to satisfy my curiosity” \( t(71.778)=-2.875, p=.005 \) and “motivate myself to make behavioral or lifestyle changes which will lower my risk for several common diseases” \( t(81)=-1.992, p=.05 \).

**Figure 8:** Individuals were asked in multiple response format, what their primary motivation(s) for pursuing PGT were. \( n=83 \)
Discussion

The purpose of this study was to explore the customers’ attitudes, beliefs and satisfaction with personal genome testing. Currently, this is a small population and a new field, thus little has been published in the medical literature. However, it is also a growing area and as our understanding of the genetic basis to common diseases grows, so will the use of PGT.

Who is having Navigenics PGT?

The population in this study is comparable to the data currently available from other sources regarding genetic or genomic testing. (Goddard, et al., 2007; Kalfoglou, et al., 2004; Stewart-Knox, et al., 2009) Individuals having PGT are generally white, well educated, and wealthy. Due to the costs of PGT, it is understandable that individuals of lower socioeconomic status would not be pursuing this type of testing.

In this study, most individuals had heard of PGT through their physician. However, this is likely a result of Navigenics’ relationship with various healthcare groups and the resulting recruitment bias. Therefore, it is not likely to be representative of the DTC population, but does show how influential these relationships are. The second most common response was learning of PGT through the media including Newsweek Magazine, New York Times, Wired magazine, and multiple television shows, all of which are aimed at educated and affluent individuals.

Another descriptive aspect of this population that is interesting involves the genetic contribution scale. Overall, the population in this group did not recognize the difference between common diseases and classic Mendelian genetic diseases, indicating that better education of the lay public is required to gain full understanding of genetic diseases. In
addition, the genetic contribution summary score was calculated to see how strongly individuals felt genetics contributed to common diseases. The average summary score indicated that respondents felt genetics played a major role in common disease development. On the contrary, it has been reported elsewhere that individuals tend to “believe that common health condition could be attributed relatively equally to genes and behavior.” (McBride, et al., 2009) One explanation for this difference could be in the manner the question was asked. In the current study, the respondents were asked to rate between ‘genetic’ and ‘non-genetic’ factors, whereas in McBride, et al., the rating scale was from ‘genetic’ to ‘behavior’ factors.

In further analysis, this summary score did not correlate with a variety of other sample characteristics, including satisfaction with test, decision to share test results with physician, decision to consult a Navigenics, annual household income, or changing behaviors after receiving results. When comparing the summary score with how useful individuals felt their PGT results were in making medical decisions, a positive correlation approached significance ($r=0.229, p=0.51$).

**Motivations**

The motivations for pursuing PGT are shown in Figure 8. The results of this study confirm the results from the CPMC data that has been released. (Gordon, et al., 2009) In both studies, defining disease risk, motivating ones’ self to make lifestyle changes, and learning about disease risk to family were significant reasons for having a PGT.

From previously published research, it has been seen that the risk to family members is a major motivating factor for pursuing genetic testing in highly penetrant diseases, such as rare familial cancer syndromes. (Espllen, et al., 2007; Lynch, et al., 1997) This is not the case when examining more common, complex diseases, such as bipolar disorder. (Meiser, et al.,
Results similar to those reported in the study of bipolar disorder are seen here. The top four reasons to pursue PGT are: to define my chance of developing any common disease, to satisfy my curiosity, to prepare for future medical concerns, and to define my chance of developing a specific common disease. These can be grouped as reasons of personal medical management or personal knowledge. Family is the fifth most common reason (to define the disease risks to my family). This trend seems logical. If there is a strong genetic component to disease development, such as in hereditary cancer or Huntington’s disease, then the risk to develop disease is high, and therefore the risk to family members is also high. However, if the genetic contribution is low, then despite the fact that the predisposing polymorphisms follow Mendelian segregation patterns, the risk to family members is also low. As further support to this trend, the researchers examined differences in motivations between individuals who have biological children and those who do not. The analysis showed that if an individual had biological children, they were more likely to view risks to family and family history of a disease as motivators for testing. If an individual did not have biological children, they were more likely to proceed with testing in order to lower personal risk for disease and out of curiosity.

Behavior Changes

In this study, about half of the respondents reported changing some health behaviors after receiving their PGT results. (Figures 6, 7) Previous research in this area is limited. In collaboration with Navigenics and others, the Scripps Translational Science Institute is currently conducting a 20-year clinical trial to assess the lifestyle changes that individuals make after personal genome testing. (clinicaltrials.gov, 2009) Medcan Clinic, a comprehensive, concierge health clinic in Toronto, carried out an internal study with their
patients who completed Navigenics PGT. They found that 69% of their population made changes to their diet, 77% changed physical activity levels, and 31% increased medical screening or testing. (personal communication, J. Davies) These percentages are higher than those we report here. (Figures 6, 7) One reason for this difference could be that the Medcan Clinic population is unique in both their financial and medical resources. One of the primary goals of this clinic is to disease prevention and it is reasonable to assume higher rates of follow-up in this comprehensive setting. Additionally, in the current study, not every individual had PGT with physician involvement. We are unable to determine how many individuals in this DTC category then sought the medical advice of their physician.

It is also interesting to examine the difference that disease penetrance has on behavior change. If we compare the rates of screening uptake in the PGT setting to those seen in hereditary cancer syndromes, it is possible that the overall change in lifetime risk makes a difference to individuals. For example, in hereditary breast/ovarian cancer syndrome, approximately 60% of all individuals increase screening after receiving a mutation-positive test result. (Peshkin, et al., 2002) This is most likely due to the high penetrance of the disease, along with the proven effectiveness of mammography. On the other hand, individuals may not feel the immediacy of preventive actions if the personal risk is not as high.

Implications for Genetic Counseling

Many genetic counselors have a negative view of DTC genetic testing and feel that genetic testing should be limited to the clinical setting. (Heuer, et al., 2008; Hock, et al., 2008) Similarly, the ACMG 2008 policy statement on DTC states that a qualified healthcare professional should be involved in all aspects of genetic testing, including counseling,
ordering, and interpretation. (ACMG, 2008) As the above discussion regarding behavior changes shows, the greatest medical advantage of PGT is seen when a physician is involved in ordering and utilizing the test results.

In regards to personal genome testing (with or without the involvement of a physician), many physicians and researchers feel that the clinical utility of PGT is low. (McCarthy, et al., 2008) In other words, the information gained from SNP analysis is not yet useful enough to support meaningful changes in clinical management. The variations examined have low penetrance, which decreases their overall predictive value. However, as evidenced by the genetic contribution scores reported above, the lay population does not understand low penetrance genetics, when compared to classic Mendelian diseases.

In contrast to these professional views, the results of the current survey show that most consumers are satisfied with the PGT process, results, and its usefulness in making medical decisions. Very few individuals mentioned uncertainty of test results as a reason for their dissatisfaction with the testing. In light of this, genetic counselors should be sure to have an open-mind when working with individuals who are pursuing PGT. In addition, they should encourage proper and accurate understanding of what the risk information means.

Limitations

Due to the retrospective nature of this study, recall of respondents’ actions and feelings before pursuing PGT may be different than they actually were at the time. In addition, there are limitations to this study as a result of the sample population. It was a convenience sample and may not be representative of the entire Navigenics customer population. Nor does this study take into account the perspectives of customers from other personal genomic testing companies that do not have physician partnerships. The sample
population may also be skewed as a result of the incentive that was offered. If an individual were unhappy with the information or communication that they have been receiving from Navigenics, they would be less likely to be enticed by the Navigenics subscription raffle or may have removed their names from the email list. Finally, there were some specific design flaws within the survey that limit the interpretation of the data. For example, the researchers are unable to determine which individuals ordered their PGT through their physician and which were strictly direct-to-consumer customers.
Conclusion

The aim of this study was to describe who has personal genome testing, why they are purchasing it, and what they are doing with the information. Most of the population reported here was interested in PGT for personal medical management or for curiosity about the results. Most of the survey respondents have a favorable view of PGT and about half report using their results to make changes to their medical management or lifestyle. Therefore, despite the hesitation in the medical and genetics community regarding the clinical utility of genomic testing, consumers are happy with what the testing provides. Future studies should look more carefully at how many individuals discuss their PGT results with their physicians and what the conversation involves. In addition, long-term studies, such as the one being done by Scripps, can provide a wealth of knowledge about outcome measures and primary prevention of common disease.
References


Twenty-Seventh Annual Education Conference of the National Society of Genetic Counselors (Los Angeles, California, October 2008). Glenside, PA: Arcadia University.


Appendix A: Recruitment notice

From our Genetic Counselors:
Our Genetic Counseling team would like to let you know about a research opportunity that might interest you.

A graduate student and researcher in genetic counseling at Brandeis University invites you to participate in an online survey because you have completed a personalized genome test from Navigenics. The study is exploring why individuals have genomic testing.

This research study is voluntary and completely anonymous. It only requires about 15 minutes of your time.

Upon completion of the survey, participants have the option of entering a compensation raffle. The two raffle winners will each receive a one-year Navigenics renewal subscription.

Please access the following website anytime between [date] and [date].

www.surveymonkey.com/samplelink

Please note:
- Participation is voluntary and your decision regarding participation will in no way affect your relationship with Navigenics.
- Participants can complete the research survey without entering the raffle.
- The data from this research may be shared with Navigenics for future use, but will always remain anonymous.
- The Brandeis University IRB, Waltham, MA, has approved this research study.

Questions regarding this study can be emailed to: ericabau@brandeis.edu, or irb@brandeis.edu

Thank you for taking the time to further research in personalized genomics!
Appendix B: Survey Questions

PERSONAL GENOME TESTS:
CONSUMER’S MOTIVATION AND PERSPECTIVE

The goal of this research project is to determine who has personalized genome testing and why. The following survey should take approximately 20 minutes. Please remember that your responses are anonymous and confidential. This survey is voluntary and you may stop at anytime. If you feel that the questions in this survey cause you distress, you may contact Shannon Kieran, MS, CGC (skieran@navigenics.com) or Barbara Lerner, MS, CGC (mebnl17@gmail.com).

If you would like to enter the raffle for a one-year Navigenics subscription, please complete the survey and follow the directions at the end. Thank you again for your participation!

1. **Demographic Questions:** The purpose of this section is to allow us to compare individuals that have completed a personalized genome test to the general population.

   1. What is your age?
      ___ 18-29 years
      ___ 30-39 years
      ___ 40-49 years
      ___ 50-59 years
      ___ 60-69 years
      ___ 70 years or older

   2. What is your gender?   Male       Female

   3. How many biological children do you have? ______

   4. Are you adopted?       Yes       No

   5. What is your race?
      ___ White
      ___ Black or African American
      ___ Asian
      ___ Native Hawaiian and other Pacific Islander
      ___ Hispanic/Latino
      ___ Other (please specify): ____________________________

   6. What is the highest level of education you have completed?
      ___ some high school
      ___ high school diploma/GED
      ___ two-year college degree
      ___ four-year college degree
      ___ graduate or professional degree
7. Please indicate the category that best describes your profession. *(Choose only one category.)*
   ___ Finance/banking
   ___ Government/law
   ___ Biological/physical sciences
   ___ Education
   ___ Industry/Manufacturing
   ___ Healthcare
   ___ Other *(please specify):* ___________________________
   ___ Not currently employed

8. What is your annual household income?
   ___ <$25,000
   ___ $25,000-49,999
   ___ $50,000-99,999
   ___ $100,000-149,999
   ___ $150,000-199,999
   ___ $200,000 or more

9. How did you learn about this survey?
   ___ My doctor told me about it.
   ___ I received information in an email from Navigenics.
   ___ Other *(please specify):* _______________________

II. Knowledge and attitudes towards genetics and healthcare: The following questions aim to categorize your knowledge and attitudes toward genetics and healthcare. There is no one right answer to these questions.

*Please rate the following on a scale of 1-5.*

10-23. To what extent do you believe the following diseases are due to genetics?
*(1 = genetics plays no role in disease development; 5 = genetics is the only factor in the disease development.)*

<table>
<thead>
<tr>
<th>Disease</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
</tr>
</thead>
<tbody>
<tr>
<td>10. breast cancer</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>11. Crohn’s disease</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>12. cystic fibrosis</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>13. diabetes (type II)</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>14. glaucoma</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>15. lactose intolerance</td>
<td>1</td>
<td>2</td>
<td>3</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>16. macular degeneration</td>
<td>1</td>
<td>2</td>
<td>3</td>
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<td>5</td>
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<tr>
<td>17. multiple sclerosis</td>
<td>1</td>
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<td>5</td>
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<tr>
<td>18. obesity</td>
<td>1</td>
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<td>4</td>
<td>5</td>
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<tr>
<td>19. prostate cancer</td>
<td>1</td>
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<td>3</td>
<td>4</td>
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<tr>
<td>20. psoriasis</td>
<td>1</td>
<td>2</td>
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<tr>
<td>21. rheumatoid arthritis</td>
<td>1</td>
<td>2</td>
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<td>4</td>
<td>5</td>
</tr>
<tr>
<td>22. Tay Sachs disease</td>
<td>1</td>
<td>2</td>
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<td>5</td>
</tr>
<tr>
<td>23. sickle cell disease</td>
<td>1</td>
<td>2</td>
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<td>4</td>
<td>5</td>
</tr>
</tbody>
</table>
Please respond to the following statements, indicating your level of agreement.

<table>
<thead>
<tr>
<th>Statement</th>
<th>Strongly Disagree</th>
<th>Disagree</th>
<th>Neutral</th>
<th>Agree</th>
<th>Strongly Agree</th>
</tr>
</thead>
<tbody>
<tr>
<td>24. I understand basic genetic principles.</td>
<td></td>
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</tr>
<tr>
<td>25. I understand all of the risk information provided in personalized genome test results.</td>
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<tr>
<td>26. It is important to know my genetic predisposition to common diseases.</td>
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<tr>
<td>27. It is important to share my genomic test results with my doctor.</td>
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<td>28. I feel my doctor understands my genomic test results.</td>
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<tr>
<td>29. I am an active participant in my healthcare management.</td>
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<td></td>
</tr>
<tr>
<td>30. Understanding my genetic predispositions to common disease will change my medical care management.</td>
<td></td>
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<td></td>
</tr>
</tbody>
</table>

**III. Your experience using a personalized genome testing service:** The following questions refer to your experience both before the testing and after receiving your test results. Again, your answers to these questions will be completely anonymous.

31. How/where did you first learn about personalized genome tests for predisposition to common diseases? *(Please be as specific as possible.)*

________________________________________________________________________

32. Did you consider a personalized genome test through another company prior to choosing Navigenics?
   ___ Yes *(please answer question 32a)*
   ___ No

   **32a.** Which company did you consider and why did you choose Navigenics?
   __________________________________________
   __________________________________________
   __________________________________________

33. Did you discuss your intention to purchase this test with anyone prior to testing? *(Select all that apply.)*
   ___ Spouse/partner
   ___ Child(ren)
   ___ Other family *(please specify): __________________________________________
   ___ Friend
   ___ Primary care physician
34. When your personalized genome test was ordered, did you order it through your doctor’s office or directly from Navigenics?
___ from my doctor
___ from Navigenics
___ through my employer

35. What were your primary motivations for purchasing a personalized genome test? (Select all that apply.)
___ To define my individual chance of developing any common disease.
___ To define my individual chance of developing a specific common disease.
___ To motivate myself to make behavioral and/or lifestyle changes which will lower my risk for several common diseases.
___ To motivate myself to make behavioral and/or lifestyle changes which will lower my risk for a specific disease.
___ To prepare for future medical concerns.
___ To define the disease risks to my family.
___ To satisfy my curiosity.
___ I am adopted.
___ My doctor recommended the test.
___ I am concerned about something in my family history.
___ My family history is incomplete.
___ My employee benefits program paid for it or subsidized some of the cost.
___ Other (please specify): ______________________________

36. Have you discussed your test results with one of the genetic counselors at Navigenics?
___ Yes (please answer question 36a)
___ No

36a. What type of information did you discuss with the counselor? (Select all that apply.)
___ test result interpretation
___ medical management
___ disease natural history
___ family history
___ other (please specify): ______________________________

37. Has your doctor discussed your results with one of the genetic counselors at Navigenics?
___ Yes
___ No

38. Have you consulted a genetic counselor or other genetic expert that is not associated with Navigenics regarding your personalized genome test results?
___ Yes (please answer question 38a and 38b)
38a. Why did you consult them?
__________________________________________________________________
__________________________________________________________________

38b. What type of information did you discuss with this specialist? (Select all that apply.)
___ test result interpretation
___ medical management
___ disease natural history
___ family history
___ other (please specify):

39. Have you discussed your personalized genome test results with your primary care physician or other medical specialist?
___ Yes (please answer question 39a)
___ No

39a. What type of information did you discuss with this physician? (Select all that apply.)
___ test result interpretation
___ medical management
___ disease natural history
___ family history
___ concern over developing a disease
___ other (please specify):

40. Please indicate your level of satisfaction regarding your personal genome test results?
___ Very satisfied
___ Satisfied
___ Neutral
___ Unsatisfied
___ Very unsatisfied
Describe why you feel this way.
__________________________________________________________________
__________________________________________________________________

41. Did the genome testing process meet your expectations?
___ Yes
___ No
Explain why you feel this way.
__________________________________________________________________
42. Have you changed any health behaviors since receiving your personalized genome test results?
   ___ Yes (please answer question 42a)
   ___ No

   42a. What types of changes have you made?
   ________________________________________________________________
   ________________________________________________________________
   ________________________________________________________________

43. How useful do you feel your personalized genome test results are at helping you make medical decisions?
   ___ Very useful
   ___ Somewhat useful
   ___ Neutral
   ___ Not that useful
   ___ Not useful at all
Please explain why you feel this way.
   ________________________________________________________________
   ________________________________________________________________
   ________________________________________________________________

44. Have you shared your personalized genome test results with family members?
   ___ Yes (please answer question 44a and 44b)
   ___ No (please answer question 44a)

   44a. Please explain why you did or did not do this.
   ________________________________________________________________
   ________________________________________________________________
   ________________________________________________________________

   44b. Which family members did you share this information with? (Select all that apply.)
   ___ Spouse/partner
   ___ Child(ren)
   ___ Sibling(s)
   ___ Parent(s)
   ___ Other (please specify): _______________________________________

45. Would you recommend personal genome testing to a friend?
   ___ Yes
   ___ No
Please explain why you would or would not do this.
   ________________________________________________________________
   ________________________________________________________________
   ________________________________________________________________
46. What does “personalized medicine” mean to you?
____________________________________________________________________
____________________________________________________________________

47. Is there anything else you would like to share about your experience with the process of ordering, receiving, and interpreting your personal genome test results?
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________