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The impact of genetic counseling on Direct-to-consumer and Patient-initiated testing

by

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Submitted in Partial Fulfillment of the Requirements
For the Degree of Master of Science in Human Genetics and Genetic Counseling
School of Pharmacy and Health Sciences
Keck Graduate Institute

2020

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Acknowledgements

I would first like to thank my thesis committee, Emily Quinn, MS, LCGC and Jamie Zdrodowski, MS, CGC for their support and advice during the research and writing process. They provided valuable feedback while developing and implementing this survey and were flexible as changes came up. Emily has been a close mentor who was enthusiastic throughout the writing process. She helped me stay motivated and passionate about completing this work. I would also like to express my appreciation to Nicholas Gorman, MPH, EdD for his thoughtful work, collaboration, and statistical analyses of survey responses. His help developing an analysis plan and recommendations on data presentation were valuable to this process. I would also like to thank our program director, Ashley Mills, MS, LCGC for her teaching, enthusiasm, and support throughout our genetic counseling program.

Lastly, I would like to thank my friends and family for their love and support, especially my parents, Jason, and Moe. The decision to go back to school would have been much more difficult without knowing that I would have your support and encouragement along the way.

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Introduction

Direct-to-consumer (DTC) testing provides consumers access to genetic testing without involvement or advice from a genetics specialist. By sending a saliva sample to a DTC company, consumers can have their DNA analyzed for information on ancestry, paternity, disease-related risks, and many other heritable traits. Approximately 246 companies offer DTC testing with 71 companies offering health-related testing, including 23andMe, AncestryDNA, and Helix (Phillips, 2016). DTC testing has recently expanded to include reports on diet, exercise, carrier testing, and pharmacogenetics (Niemiec, Kalokairinou, & Howard, 2017). While this expansion has increased consumer access to genetic interpretations, these reports can often provide a false sense of security and rarely cover enough genetic information to make significant interpretations about disease risk.

To provide an example of difficulties in navigating DTC test results, imagine ordering testing from a company like 23andMe. You open an email link and get a test report that indicates that you do not have any of the 3 *BRCA* mutations that 23andMe identifies. You feel relieved and might be less concerned about following recommended cancer screenings, typically mammograms for women and prostate cancer screenings for men (Bellcross, Page, & Meaney-Delman, 2012). You might also have a lower perceived risk than your actual cancer risk. Without reading and understanding the full report, you may not realize that most cancers caused by *BRCA* mutations are not caused by the 3 mutations that 23andMe looks for, and that these mutations are mostly relevant to people with Ashkenazi Jewish ancestry. If you do not have Ashkenazi Jewish ancestry, this information is likely irrelevant to your *BRCA*-related cancer risk. This scenario highlights a common disconnect between DTC test results and impressions of disease risk. If you

had a strong family history of breast cancer, further genetic testing might be medically advisable. If you met with a genetic counselor, after looking at your family history, they might recommend full sequencing of the *BRCA1* and *BRCA2* genes, *PALB2*, and other genes associated with increased breast cancer risk (Miki et al., 1994; Rahman et al., 2007; Wooster et al., 1994). This level of detail can help provide a more comprehensive view of risk and is built upon personal ethnicity and family history. With the guidance and involvement of a genetic counselor, the meaning and possible impact of direct-to-consumer testing results can be clearly explained, and possible follow-up testing can be provided.

Currently, direct-to-consumer testing does not typically include involvement or guidance from genetic counselors. Consumers order a genetic test online and then view their results. If consumers hope to make health-related decisions based on testing, involving a genetics professional can be a valuable way to help interpret and explain limitations of DTC results. While studies have investigated the uptake of genetic counseling after DTC testing and some outcomes related to understanding test results after counseling, changes in attitudes and understanding of specific results provided in DTC reports have not been examined before and after genetic counseling sessions. The purpose of this study is to examine genetic counselors' opinions about how genetic counseling sessions may change an individual's understanding, perceived value, and anxiety related to direct-to-consumer (DTC) and/or patient-initiated genetic testing. With the guidance of a genetic counselor, health advice can be tailored to the patient and health outcomes may improve.

Background

Motivations for ordering DTC testing

Consumers often order DTC testing with the hope of gaining information relevant to their health, ancestry, and to find blood relatives. A ranking of motivations for pursuing DTC-PGT (personal genomic testing) from 23andMe and Pathway Genomics found that top motivations were personal disease risk, desire to improve health, and finding out about personal medication response (Koeller, Uhlmann, Carere, Green, & Roberts, 2017). Since DTC test results can influence health management, it is important for consumers to understand the limitations and values of DTC genetic testing reports as they relate to interpreting disease risk.

Variant Detection

For individuals hoping to gain health-related information, DTC testing can have limited, incorrect, or misleading information. DTC tests typically scan for variants associated with diseases or specific traits that have been identified via genome-wide association studies (GWAS) (Bellcross et al., 2012). These studies look at single nucleotide changes, or SNPs (single nucleotide polymorphisms), that are associated with a given disease by comparing individuals from a specific population with and without a disease to people with and without a given nucleotide change. Variants identified via GWAS are not typically generalizable and DTC results do not incorporate familial risk (Bellcross et al., 2012). The problem with applying these studies to the general public is that typically SNPs associated with a specific disease are relevant to a small portion of the total population, like how a set of 3 common *BRCA1* and *BRCA2* variants are relevant to individuals with Ashkenazi Jewish ancestry and are considered founder mutations (Bellcross et al., 2012). SNPs that are identified in DTC testing typically have odds ratios of less than 1.5% for common diseases (Bellcross et al., 2012). If a DTC test looked for an SNP associated with colon cancer, which had an odds ratio of 1%, a test indicating that someone

did not have this SNP would provide almost no information on this person's colon cancer risk. In this case, 99% of colon cancers are not caused by the mutation identified in testing. This highlights the limited generalizability of most genetic testing for SNPs.

Ambry genetics recently ran a study on variants associated with diseases that were identified in reports from DTC companies (Tandy-Connor et al., 2018). Testing to confirm the DTC results came from medical providers, typically genetic counselors, medical geneticists, or oncologists, who had ordered testing to confirm the variants identified via DTC genetic testing. Of the 49 patients tested, 43 (87.8%) of them received reports indicating that they had variants associated with cancer risk caused by mutations in genes including *BRCA1* and *BRCA2*, *CHECK2*, *TP53*, and *ATM*. After full gene sequencing to look for variants in the genes reported, 40% of the DTC results were found to be false positives. This means that 4 out of 10 patients who were given reports claiming they had a disease-causing variant did not actually have the variant. These findings indicate a significant problem with quality and interpretation of genetic testing by DTC companies. In addition, 8 variants were classified as increasing disease risk by either DTC or third-party interpretation (TPI) analysis in the genes *ATM*, *BRCA1*, *BRCA2*, *COL3A1*, and *COL5A1*. Pathogenic variants in the genes *ATM* and *BRCA1/2* are associated with increased risk for breast, ovarian, and prostate cancer, while pathogenic variants in *COL5A1* and *COL3A1* are associated with the connective tissue disorder Ehlers-Danlos syndrome. These variants were identified as benign, or not causing disease, by databases including 1000 Genomes and Ambry (Auton et al., 2015; Tandy-Connor et al., 2018). DTC testing often provides inaccurate data that may be irrelevant to the consumer ordering testing.

While the previous study indicates high false positive rates in DTC testing, we do not know their false negative rates. A false negative report would falsely indicate that a patient does

not have a risk-predisposing mutation, when they do have a mutation associated with disease risk. When patients may act based on DTC test results, companies' incorrect negative or positive results can have serious consequences. In the case of a false negative, someone may think they have reduced risk based on a result indicating that they do not have any of the screened mutations associated with cancer. If this patient has a mutation that increases cancer risk, an early cancer could be missed. For healthcare providers unfamiliar with genetics, seeing a DTC test result that indicates increased or decreased risk could also guide how they treat patients, or even their recommendations for preventative surgeries. If a patient presents a test report indicating they have a *BRCA1* or *BRCA2* mutation, a doctor may follow guidelines of recommending prophylactic bilateral mastectomy and salpingo-oophorectomy (removal of the ovaries and fallopian tubes) without confirmatory testing (Ludwig, Neuner, Butler, Geurts, & Kong, 2016). Overall, direct-to-consumer testing has limited value and reliability.

Consumer Expectations from DTC results

In a study which surveyed 23andMe and Pathway Genomics customers, PGEN participants reported lower genetics self-efficacy scores after receiving DTC results (Carere, Kraft, Kaphingst, Roberts, & Green, 2016). Genetics self-efficacy was assessed using a 5-item Likert scale, with consumers rating how strongly they agreed with statements like "I am confident in my ability to understand information about genetics" and "I have a good idea about how genetics may influence risk for disease generally" on a scale from 5 (all strongly disagree) to 35 (all strongly agree). There was a significant decrease in genetics self-efficacy scores from baseline to 6-months after viewing DTC test results. This decrease may reflect decreased confidence in understanding and interpreting the relevance of genetic information to health after viewing complex, lengthy results. This also likely indicates that consumers overestimate their ability to interpret and understand genetic information and their confidence changes after

attempting to interpret results on their own. This decrease highlights the value of genetics specialists in helping patients interpret and understand genetic testing results, as well as explaining some of the limitations of DTC testing. In addition, this study included a series of nine true-false questions to gauge genetics knowledge. Consumer baseline scores in genetics knowledge were very high and increased slightly 6-months after viewing results. With this small increase in genetics knowledge, it is difficult to gauge whether the change in genetics knowledge resulted from subjects' increased understanding of genetics after viewing and understanding test results, or from other variables including pretest sensitization, ceiling effects, and maturation. In this case, maturation would describe a general increase in public genetics knowledge during the 6 months after respondents completed baseline surveys. Pretest sensitization refers to a change in test scores because respondents may have learned answers after seeing the baseline survey. The authors also acknowledge the likely influence of a ceiling effect, since the average baseline score of 8.15 was very close to the maximum score of 9.

Further analysis following the PGEN study revealed that perceived risk of breast, prostate, and colorectal cancer increased when DTC results reported elevated risk, while perceived risk decreased when DTC results reported that an individual had an average population risk (Carere et al., 2015). With lung cancer, perceived risk increased, regardless of whether results indicated an average or increased cancer risk. Having a decreased perceived risk of different types of cancer can have negative health consequences if patients decide to avoid preventative screenings and have a false sense of low risk.

Genetic Counseling Following DTC Testing

The PGEN study also surveyed individuals to help understand motivations for pursuing genetic counseling following DTC testing. Out of 1,026 individuals enrolled in the study, only 4% sought genetic counseling. Compared to participants who did not seek genetic counseling,

participants who pursued genetic counseling tended to report poorer health, were more likely to have had previous genetic counseling, were younger, and more likely to have children. They also tended to report higher uncertainty about what test results meant about personal risk (60% of GC seekers vs 27.2% of non-GC seekers) (Koeller et al., 2017).

Unlike the PGEN study, the Scripps Genomic Health Initiative enrolled patients in a DTC PGT program which included no-cost access to genetic counseling services (Darst, Madlensky, Schork, Topol, & Bloss, 2013). Consumers would receive test results from Navigenics, a DTC company which offered free genetic counseling services to help explain results to consumers; Navigenics was acquired by Life Technologies in 2012 and no longer offers DTC testing (Dorfman, 2013). Among the 1,325 participants, 14.1% spoke with a genetic counselor; when an active effort was made to contact patients about GC services via phone or email, utilization rates of GC went up to 27.3% (Darst et al., 2013). When asked about reasons for meeting with a genetic counselor, 43.9% reported wanting to take advantage of a free service and 42.2% wanted more information on risk calculations. After genetic counseling, 85% reported that genetic counseling helped them improve their understanding of test results and 75.5% reported that it improved their understanding of genetics overall. Of those who did not pursue genetic counseling, 55.6% believed that they already understood their results and approximately 20% reported wanting to use genetic counseling but were 'too busy'. This indicates that even when genetic counseling services are provided at no cost, utilization rates following DTC testing are low. However, amongst patients who see a genetic counselor, they generally report very high satisfaction and find value in the interpretation and explanations provided by genetic counselors.

Results with high psychological burden

With an increasing number of DTC companies providing information on disease risk relating to serious conditions such as breast cancer, Alzheimer's, and Parkinson's, genetic testing companies have a responsibility to offer support. Without pre-test counseling, consumers may not have considered how they might be affected by a result indicating that they are at increased risk of developing all the diseases analyzed via DTC testing. They may not have thought through how a positive result might affect their relatives and children, and their own mental health. Consumers may be left with burdensome results that can shake up their world, with nothing more than an email link to information on the disease and the mutation that they carry. When providing such burdensome results, DTC companies should make access to pre- and post-test genetic counseling available to consumers (Middleton, Mendes, Benjamin, & Howard, 2017). Considering that studies on positive genetic testing results have found increased anxiety and depression amongst patients after disclosure of the genetic test results, an individual who can provide resources and tools for support is valuable to navigating the emotional distress that positive results can bring (Benusiglio et al., 2017; Brédart et al., 2017; Scherr, Christie, & Vadaparampil, 2016).

Ancestry Testing

Ancestry testing has been used to estimate pan-ethnic heritage and users can often opt into receiving information on relatives identified via ancestry testing. These ethnicity estimates are based on current native and indigenous populations, and similarities to their DNA is used to estimate a person's geographic ancestry (Kirkpatrick & Rashkin, 2017). While these can often provide accurate continental information, estimates of specific regions within a country are likely inaccurate. These estimates are unable to account for large migrations and population bottlenecks, including epidemics or famine. For these reasons, the 'native population' DNA used

as a proxy for pan-ethnic origin today may have originated or moved from other locations and likely interbred with populations from other regions. For these reasons, test results with ancestry information are likely unreliable if they estimate ancestry within a small geographic area.

Individuals who order ancestry testing through companies like Family Tree DNA, 23andMe, Ancestry DNA, and National Geographic Geno 2.0 can download raw data VCF files, which can be uploaded for third-party interpretation. Third-party interpretation (TPI) companies, such as Promethease, Interpretome, and Livewello provide SNP analysis of the data file from DTC testing for additional information on ancestry, disease risk, metabolism, and other traits. Third party interpretation (TPI) of data from DTC test results rarely involves complete informed consent, medical oversight, or accurate interpretation (Badalato, Kalokairinou, & Borry, 2017). While the ability to know as much as possible about the data generated from a DTC test seems valuable, unfortunately these reports can often be misleading or incorrect.

Data Privacy

Many consumers are unaware of how their data will be used by DTC and TPI companies. In a review of a Canadian DTC company websites, researchers found that 67% of the companies reviewed did not provide adequate information for consumers to understand how their data might be used (Christofides & O'Doherty, 2016). Many DTC companies sell consumer information to pharmaceutical companies to use with the intention of developing drugs tailored to disease risk. In a study of DTC companies which offer health or ancestry testing, researchers reviewed companies for privacy policies or terms of service. Out of 30 reviewed companies, only 13% stated that they would not use consumer data for research or other purposes and 30% of the companies indicated that they planned to use consumer data for health-related research (Laestadius, Rich, & Auer, 2017).

Recently, 23andMe has partnered Genentech, which has paid \$10 million with the interest of gaining access to DNA sequencing information relevant to Parkinson's disease drug development (Mullard, 2015). The partnership includes plans to perform full genome sequencing on 3,000 patients with Parkinson's disease. With widespread sales of personal genomic data, the informed consent process should be thorough and clearly explain to consumers that their data is not private. Unfortunately, many consumers order direct-to-consumer with misperceptions about the privacy of their data due to an inadequate informed consent process.

Patient-Initiated Testing

Patient-initiated testing involves patients seeking out and ordering genetic testing, typically through an online platform. Genetic testing may be ordered to understand hereditary cancer risk or reproductive risks through carrier screening. Generally, patients pay on a cash basis for a genetic test of their choice. Their self-reported medical and family history is reviewed by a clinician before the test is ordered. Then the patient provides a saliva or buccal sample for genetic testing. Genetic testing results can typically be viewed through an online portal, often with the option to discuss results with a genetic counselor.

Some patient-initiated testing includes genes on the ACMG (American College of Medical Genetics) 59 list, which is a list of genes for which reporting findings on clinical exome or genome sequencing is recommended (Kalia et al., 2017). These genes are considered medically actionable, meaning that patients with pathogenic mutations in any of these genes have possible screening or treatment options available for the associated condition. As an example, genes associated with Lynch Syndrome, MLH1, MSH2, MSH6, and PMS2, have management recommendations including colonoscopies every 1-2 years, starting age 20-25 or 2-5 years before earliest diagnosis of colon cancer in a family (National Comprehensive Cancer Network, 2019).

Genetic Counselor Involvement

With the limited relevance to healthcare and poor reliability of DTC test results, it can be valuable for consumers to meet with genetic counselors to clearly explain the limitations of testing and to identify possible health-related risks based on family history. Genetic counselors can also provide possible follow-up testing with certified labs when disease risk based on family history or DTC testing suggests that additional testing may be useful. Since false positive rates in DTC testing are high, it is very important to confirm variants reported via DTC testing or TPI through certified labs with full gene sequencing. Since genetic counseling demand currently exceeds the availability of genetic counselors, access to counseling about DTC testing results will likely depend on alternative models of GC. These models include telegenic, telephone counseling, and group counseling (McCuaig et al., 2018). Telegenic counseling involves meeting with patients via video conference, similar to skype. Group counseling sessions involve presenting to a group of individuals about disease risk and genetic testing, often with the opportunity to meet one-on-one following a group presentation and discussion. Although one-on-one counseling sessions typically have a more personal and patient-centered feel, alternative models are currently necessary to meet patient demand. For individuals who live far from a clinic or hospital with genetic counselors available, telehealth may be the only option. To meet with patients regarding DTC test results, group counseling sessions could provide a high-impact way to explain DTC test results and some of the limitations in testing. Following a group counseling session, patients could meet one-on-one with counselors to discuss the findings within their personal DTC report. Genetic counselors can help navigate DTC results and suggest certified labs to run follow-up analyses

Previous studies have not done an in-depth comparison of consumers' understanding of test results, genetics understanding, and feelings of anxiety or depression resulting from DTC

test results. This study seeks to understand the change in consumer attitudes and understanding of test results by surveying genetic counselors. I expect that genetic counselors see increased understanding of limitations in DTC test results, increased genetics knowledge, and reduced anxiety and depression related to testing results after genetic counseling sessions regarding DTC results.

Materials and Methods

Survey Distribution and Content

This study involved distributing a survey via email listserv to the National Society of Genetic Counselors' (NSGC) members through their Student Research E-blast service. Participants were at least 18 years old and genetic counselors. Respondents were asked for basic demographic information including ethnic origin, age, gender, zip code of practice, and area of practice (clinical, industry, or research). The survey asked about impressions of patient understanding and feelings related to DTC and patient-initiated genetic testing results. Examples of questions included level of agreement with statements like 'Before counseling, patients have anxiety related to their DTC genetic testing results' and 'I am in favor of consumers being able to self-order clinical testing of larger panels, including genes outside of the ACMG 59 gene list.' A copy of the survey is available in Appendix A. The survey asked for impressions of how consumers feel about their DTC results before compared to after genetic counseling sessions. The survey also asked about consumer anxiety, understanding of the medical significance of results, limitations of results, how genetic variants influence health and disease risk, and how well consumers understand their results report.

Counselors were asked on a 5-item Likert scale from 'strongly disagree' to 'strongly agree' about impressions of patient-initiated and DTC testing. Questions included whether they support consumers being able to self-order genetic testing of the ACMG 59 gene list, self-order large panels including genes outside of the ACMG 59 list, whether pre-test counseling is necessary to have informed consent, whether speaking with a genetic counselor regarding results

should be optional, and level of concern regarding the expansion of DTC and patient-initiated testing.

Lastly, a series of open-ended questions asked counselors to describe what information regarding DTC testing and results consumers find most helpful, the misconceptions consumers have surrounding testing, challenges in dealing with DTC results, and experiences with patient-initiated testing.

Data Cleaning

Data cleaning was performed in order to detect invariant or careless respondent answers and ensure that all participants qualified to participate in the study. All 60 respondents consented to participate in the study. Of the 60 respondents, 6 (10.0%) failed to provide any information after providing consent and were thus removed from the dataset, resulting in a final sample of 54.

The amount of time respondents spent completing the survey was examined to rule out responses from bots. The survey took participants, on average, 31.0 minutes to complete ($SD = 86.4$), with examination of shortest completion time (0.8 minutes) and longest completion time (9.6 hours) showing several outliers. Visual inspection of subjects' responses revealed that those who completed the survey extremely quickly simply left many questions blank. These subjects were left in the dataset, as their responses appeared to reflect good faith efforts from human respondents (i.e. not near-instantaneous responses to all questions by bots), and missing data is simply excluded from pertinent analyses. Extremely long response times (> 1 hour) were observed for 4 respondents, but all 4 responses appeared to represent good faith efforts to respond to the survey, and could be explained by practices such as opening the survey in another tab and forgetting about it. As a result, no subjects were removed due to completion times.

Examination of subject's written responses, histograms of numeric variables, and frequency tables of categorical variables failed to detect any evidence of careless, malicious, or invariant responses, so no additional responses or subject were removed from the dataset.

Data Analyses

Study data were collected and managed using Qualtrics. Data analysis was performed using SPSS Software for independent samples t-tests, chi-square tests of independence, and paired samples t-tests. For statistical analysis, $p < 0.05$ was considered a significant difference.

Independent samples t-tests were run to compare differences in age, percentage of genetic counseling sessions including discussions of direct-to-consumer test results, and percentage of genetic counseling sessions including discussions of patient-initiated test results between clinical and industry/research/other specialties. Chi-square tests of independence were used to compare differences in ethnicity and gender between clinical and industry/research/other specialties. Paired samples t-tests were run to report perceived differences in patient outcomes before and after counseling sessions discussing direct-to-consumer test results. Independent samples t-tests were run to compare projected changes in patient outcomes surrounding direct-to-consumer test results between clinical and non-clinical genetic counselors. Lastly, independent samples t-tests were used to compare clinical and non-clinical genetic counselor differences in attitudes toward direct-to-consumer and patient-initiated genetic testing.

Results

Demographics

Table 1. Demography of Responding Genetic Counselors (n = 53 – 54)

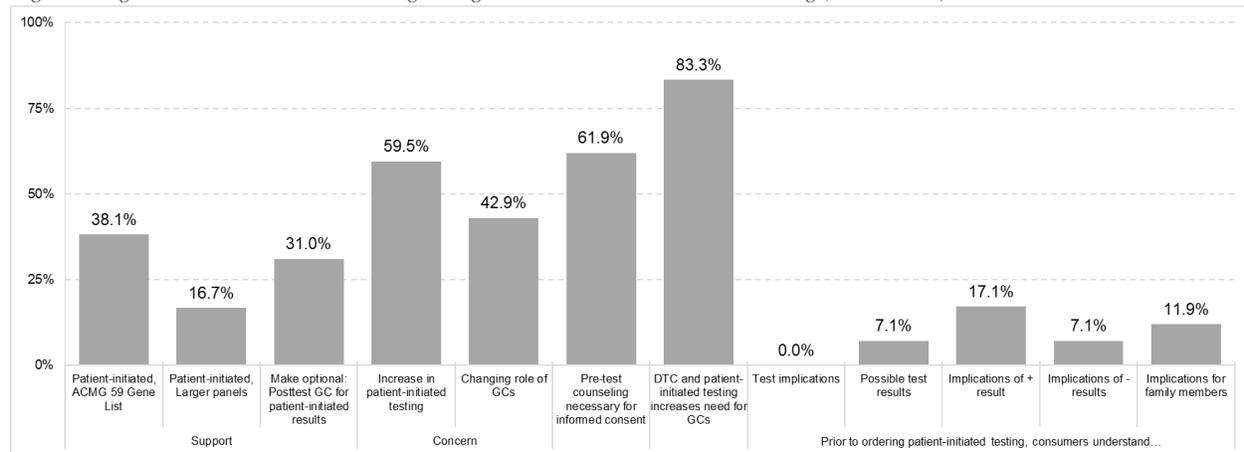
Demographic	Overall		Clinical		Industry/Research/Other		p*
	M (SD)	Freq (%)	M (SD)	Freq (%)	M (SD)	Freq (%)	
Age	30.3 (7.3)		29.6 (6.8)		35.3 (8.9)		.05
Discussion: DTC (%)	6.9 (14.0)		7.7 (14.5)		0 (0)		.25
Discussion: Patient-initiated (%)	2.7 (5.3)		3 (5.5)		0 (0)		.001
Ethnicity							.49
Asian / Asian Indian		3 (5.6)		3 (6.4)		0 (0)	
White		51 (94.4)		44 (93.6)		7 (100)	
Gender							n/a
Male		0 (0)		0 (0)		0 (0)	
Female		54 (100)		47 (100)		7 (100)	

* Comparisons between Clinical counselors and Industry/Research/Other counselors conducted via independent samples t-tests or chi-square tests of independence, where appropriate.

As shown in Table 1, limitations in the demography of respondents warrant consideration and should be kept in mind for the remainder of the analyses discussed below. Specifically, respondents to this survey were entirely female and overwhelmingly white, with the majority (87.0%) of respondents indicating working in a clinical setting. Amongst respondents from Industry, Research, or Other settings, none of their counseling sessions included discussions of DTC or Patient-initiated test results. Amongst clinical genetic counselors, about 8% of counseling sessions included discussion of DTC testing and 3% included discussions of patient-initiated testing. There was not a significant difference in percentage of counseling sessions involving discussion of DTC vs Patient-initiated testing between clinical and industry/research/other counselors. Percentage of counseling sessions including discussions of patient-initiated testing were significantly higher amongst clinical genetic counselors. In addition, respondents exclusively from Industry, Research, or Other settings were significantly older than respondents exclusively working in Clinical settings.

Genetic counselor attitudes towards direct-to-consumer and patient-initiated testing

Figure 1. Agreement* with Statements Regarding Patient-Initiated and DTC Testing (n = 41 - 42)



* Note: Agreement refers to a score of 5 “strongly agree” or 4 “agree” on a 5-pt Likert-type scale

Level of agreement with statements regarding direct-to-consumer and patient-initiated genetic testing was outlined in Figure 1 to show genetic counselor attitudes towards genetic testing.

Agreement was noted when counselors selected ‘agree’ or ‘strongly agree’ on a 5-point Likert-type scale. Most genetic counselors surveyed agreed that pre-test counseling with a genetic counselor is necessary for consumers to have informed consent before ordering patient-initiated testing. A majority also expressed concern about the expansion of patient-initiated testing and the evolving role of genetic counselors. Over 80% agreed that as DTC and patient-initiated testing options expand, the need for genetic counselors is going to increase. Fewer than half of genetic counselors supported consumers being able to self-order clinical testing of the ACMG 59 gene list, and even fewer supported consumers being able to self-order clinical testing of larger panels, including genes outside of the ACMG 59 gene list. About one third of genetic counselors agreed that speaking with a genetic counselor after receiving patient-initiated test results should be optional. Just under half of genetic counselors were concerned about how the role of clinical genetic counselors will change as DTC and patient-initiated testing options expand. Fewer than 10% of genetic counselors agreed that consumers understand the possible test results before

ordering patient-initiated testing and that consumers ordering patient-initiated testing understand the health-related implications of a negative result. Fewer than 20% of genetic counselors agreed that consumers ordering patient-initiated testing understand the health-related implications of a positive result and how patient-initiated test results apply to family members. None of the genetic counselors surveyed felt that consumers understand the implications for long-term care, life, and disability insurance before ordering patient-initiated testing.

Perceived changes in patient outcomes with genetic counseling

Table 2. Summary of Paired Samples *t*-tests Comparing Projected Changes in Patient Outcomes Surrounding Direct to Consumer Test Results ($n = 38 - 42$)

Patient Outcome	Counseling Session M (SD)		t	df	Cohen's D	p
	Pre	Post				
Anxiety	3.2 (1.1)	2.8 (1.0)	1.5	37	0.29	.14
Knowledge: Medical Sig.	1.7 (0.7)	3.9 (0.8)	-15.3	39	2.35	< .001
Knowledge: Limitations	1.5 (0.7)	3.7 (0.8)	-15.1	41	2.32	< .001
Knowledge: Disease Risk	2.2 (1.1)	3.9 (0.8)	-9.5	41	1.51	< .001
Knowledge: Report Results	2.3 (1.0)	4.1 (0.7)	-10.6	41	1.69	< .001

Perceived changes in patient outcomes with genetic counseling regarding DTC results, are outlined in Table 2. This comparison was made through a series of questions asking for the level of agreement with statements such as 'before genetic counseling consumers have anxiety related to their DTC genetic testing results'. A second version of this question was also asked regarding patient anxiety after a genetic counseling session. These pairs of questions regarding consumer attitudes and understanding were used to compare changes before and after counseling in consumer understanding of medical significance of DTC results, understanding of the limitations of DTC testing, understanding of how genetic variants can affect health and disease risk, and understanding of their DTC result reports. A series of paired samples *t*-test revealed that respondents estimate that, following genetic counseling regarding direct to consumer test results, there are statistically significant improvements in all patient outcomes except for anxiety (p 's < 0.05). Regarding the clinical significance of the reported statistically significant differences, all

statistical significant comparisons had Cohen's D scores of greater than 0.8, Cohen's suggested cut-point for large effects (Cohen, 1988). This suggests that the anticipated changes in clients scores is not only statistically significant, but also of a clinically meaningful magnitude.

Perceived changes in patient outcomes by area of practice

Table 3. Summary of Independent Samples *t*-tests Comparing Projected Changes in Patient Outcomes by Clinical versus Industry GCs ($n = 38 - 42$)

Patient Outcome*	Area of Practice		t	df	Cohen's D	p
	M (SD)					
	Clinical	Non-Clinical				
Δ Anxiety	-0.4 (1.4)	0.5 (1.3)	-1.3	36	0.64	.21
Δ Knowledge: Medical Sig.	2.3 (0.9)	1.8 (1.1)	1.1	38	0.54	.28
Δ Knowledge: Limitations	2.3 (1.0)	2.0 (1.0)	0.6	40	0.30	.56
Δ Knowledge: Disease Risk	1.6 (1.1)	1.8 (1.3)	-0.4	40	0.18	.70
Δ Knowledge: Report Results	1.8 (1.1)	1.4 (0.9)	0.8	40	0.37	.41

* Calculated as Posttest – Pretest score.

In order to compare estimated changes in patients' outcomes due to genetic counseling by clinical respondents compared to non-clinical ones, change scores (post-pre) were calculated for each of the variables in Table 2. As shown in Table 3, no statistically significant differences were observed between clinical and non-clinical respondents' estimations of how much patients' scores would change from before genetic counseling to after genetic counseling (p 's > 0.05). It is worth noting that among the non-clinical respondents, an already small group ($n = 7$), only 4-5 offered ratings on these variables, reducing their already small sample size further. Interestingly, while no results were statistically significant, two comparisons showed effect sizes greater than the threshold for Medium-sized effects (Cohen's $D = 0.5$; Cohen, 1988). In addition, while clinical genetic counselors perceived decreased anxiety after genetic counseling regarding DTC results, Non-clinical counselors perceived an increase in anxiety after genetic counseling.

Comparison in attitudes toward DTC and patient-initiated testing between clinical and non-clinical genetic counselors

Table 4. Summary of Independent Samples *t*-tests Comparing Attitudes Towards DTC and patient-initiated Genetic Testing of Clinical versus Industry GCs (*n* = 41-42)

Attitudinal Item	M (SD)		t	df	Cohen's D	p
	Clinical	Non-Clinical				
Support: self-order ACMG 59 gene list	3.7 (1.9)	2.8 (1.1)	1.0	40	0.49	.34
Support: self-order larger panels	2.5 (1.8)	2.8 (1.1)	-0.3	40	0.17	.76
Require: GC prior to patient-initiated testing	3.7 (1.3)	3.6 (1.1)	0.2	40	0.08	.83
Make Optional: Post-patient-initiated testing GC	3.0 (1.6)	3.0 (1.4)	0.0	40	0.00	1.00
Concern: Expansion of patient-initiated testing	4.0 (1.3)	3.4 (0.9)	0.9	40	0.48	.37
Concern: Changing role of GCs	3.4 (1.3)	3.2 (1.1)	0.2	40	0.16	.81
DTC and patient-initiated testing increase the need for GCs	4.6 (0.9)	4.0 (0.0)	1.5	36*	0.71	<.001
Consumers Knowledge: Patient-initiated Implication	1.5 (0.9)	1.8 (0.8)	-0.7	39	0.34	.47
Consumers Knowledge: Results of patient-initiated	2.1 (1.4)	2.4 (0.9)	-0.4	40	0.22	.68
Consumers Knowledge: Implications of + result	2.9 (1.5)	2.8 (0.4)	0.1	39	0.07	.93
Consumers Knowledge: Implications of - result	2.1 (1.4)	2.2 (1.1)	-0.1	39	0.07	.92
Consumers Knowledge: Implications for family	2.5 (1.3)	2.8 (0.4)	-0.4	40	0.24	.67

* Levene's Test for the Equality of Variances revealed a violation of the assumption of equality of variance; degrees of freedom were adjusted by SPSS to compensate

In order to compare attitudes toward DTC and patient-initiated testing between clinical respondents and non-clinical ones, a series of independent samples *t*-tests were conducted (see Table 4). Only a single comparison emerged as statistically significant; respondents from clinical settings more strongly agreed that as DTC and patient-initiated testing options expand, the need for genetic counselors is going to increase (all other comparisons $p > 0.05$). As previously discussed, small sample sizes in the non-clinical condition likely undermined the statistical power of these analyses, though Cohen's *D* values were suggestive, with a couple of comparisons approach the benchmark for "medium" sized effects (Cohen's $D = 0.5$; Cohen, 1988).

Discussion

This study highlights the value genetic counselors see in pre- and post-test counseling for patients, and a general concern about the expansion of patient-initiated testing. Fewer than half of genetic counselors surveyed supported consumers being able to self-order clinical testing of the ACMG 59 gene list, and even fewer supported consumers being able to self-order larger panels. In addition, most counselors felt that pre-test counseling prior to ordering genetic testing was necessary to have informed consent, and only 1/3 felt that genetic counseling after receiving patient-initiated test results should be optional. This highlights the general impression that genetic counseling is important to ensuring that consumers are well informed before and after ordering genetic testing. In addition, none of the genetic counselors surveyed felt that consumers understand the implications for long-term care, life, and disability insurance before ordering patient-initiated testing. Genetic testing can have implications to insurance coverage, which consumers should be aware of before ordering testing. Test results may show an increased risk for certain types of cancers or diseases, thus impacting coverage. Fewer than 10% of genetic counselors agreed that consumers understand the possible results before ordering patient-initiated testing, and that consumers understand the health-related implications of a negative result. Fewer than 20% of genetic counselors agreed that consumers ordering patient-initiated testing understand the health-related implications of a positive result and how patient-initiated test results apply to family members. Overall, counselors seem to place significant value on consumers being well-informed about the implications of, and possible results from, genetic testing. This concern translates to a strong value placed on having consumers meet with a

genetics specialist who can clearly review family history and provide informed genetic testing options, with a review of possible outcomes and results.

Genetic counselors also generally felt that patients have significant increases in understanding the medical significance of DTC results, understanding the limitations of DTC testing, understanding how genetic variants can affect health and disease risk, and understanding their DTC result reports after genetic counseling regarding DTC results. These increases indicate how valuable it can be for consumers to meet with a genetic counselor to discuss DTC results. They may learn about additional testing recommendations based on their family history and limitations to their current results. Genetic counselors did not express a significant decrease in patient anxiety following genetic counseling discussions regarding DTC results, likely because patients may learn about the limitations in DTC testing as well as develop a more comprehensive understanding of hereditary disease risk based on their family history, which could either increase or decrease anxiety. Generally, genetic counselors have observed increases in patient knowledge with possible short-term increases or decreases in anxiety surrounding awareness of hereditary disease risk.

While most differences in attitudes toward DTC and patient-initiated testing between clinical respondents and non-clinical genetic counselors were not significant, there were some interesting differences. Clinical genetic counselors expressed a slightly higher level of concern about the expansion of patient-initiated testing and the evolving role of genetic counselors. Clinical genetic counselors also expressed higher agreement that DTC and patient-initiated testing increased the need for genetic counselors, and this difference was significantly higher than that of non-clinical genetic counselors. Clinical genetic counselors may have experiences counseling patients who have received DTC or patient-initiated results, that increases their level

of concern and impression that expansion of genetic testing increases the need for genetic counseling.

It should also be noted that the results of this study are not representative of the genetic counseling community as a whole. This sample was heavily skewed towards white, women working in clinical settings. The genetic counseling community itself is very limited in its diversity as well, being roughly 90% white or Caucasian, 5% Asian, 3% Asian Indian, 1% Black or African American, and less than 1% American Indian, Alaskan Native, Native Hawaiian, or Other Pacific Islander (National Society of Genetic Counselors, 2019). It is difficult to know whether this skewed sample might have different views on patient access to self-testing, as compared with a diverse sample that more closely resembles the general population. Future research could include more of an effort to recruit non-white subjects, males, and subjects from non-clinical settings. For this study, our recruitment involved email distribution to a listserv for all members of the NSGC. In future studies, it might be helpful to reach out directly to organizations of genetic professionals of color, such as the Minority Genetic Professionals Network.

Although this study did not survey patients directly, it provides possible questions to ask as part of a survey of patients before and after counseling sessions. This survey is limited, as genetic counselor impressions of patient understanding and feelings surrounding genetic testing may not reflect actual changes experienced from a patient's perspective. We hope that these findings can be used to inform future studies that survey patients who have undergone genetic counseling before and after receiving DTC or patient-initiated test results. We also hope that these questions can be incorporated to compare patient understanding with in-person or

telegenetic genetic counseling to short information videos or reading materials presented to consumers before and after ordering testing.

Conclusion

Overall, this study highlights the value of genetic counseling on patient understanding before and after receiving genetic testing results. Interestingly, while genetic counselors expressed concerns about the expansion of DTC and patient-initiated testing, direct-to-consumer testing has recently declined. When comparing November 2018 to November 2019, direct-to-consumer genetic testing offered through Ancestry.com declined 38% and 23andMe testing declined 54% (Molla, 2020). This may indicate increased awareness amongst consumers of privacy concerns and limitations in health-related information offered by this type of testing. We also identified some of the values seen in genetic counseling. Respondents felt that patients have increases in understanding the medical significance of DTC results, understanding the limitations of DTC testing, understanding how genetic variants can affect health and disease risk, and understanding their DTC result reports after genetic counseling regarding DTC results. While genetic counselors are often involved in psychosocial aspects of counseling, there was not a strong perceived decrease in patient anxiety surrounding results following genetic counseling; this may be related to a thorough understanding of disease risk provoking a level of anxiety for some patients.

We have shown the strong value that genetic counselors see in informing patients about genetic testing. While this study highlights the importance of informing patients about limitations in testing, privacy concerns, and insurance coverage implications, it seems that consumers are beginning to pursue these tests with lower frequency. Genetic counseling continues to play a valuable role in informing patients about nuances of genetic testing while providing individual recommendations for testing. Genetic counselors can help inform patients

about genetic testing as part of a collaborative team between patients, genetic testing laboratory staff, physicians, and medical care providers.

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Appendices

Appendix A: Genetic Counselor Survey

Investigating Genetic Counselor Opinions and Experiences with Direct-To-Consumer (DTC) and Patient-Initiated Testing

You are invited to volunteer for a research study. While volunteering will probably not benefit you directly, your response will contribute to the growing body of literature addressing genetic counselor experiences and opinions on direct-to-consumer and patient-initiated testing. If you decide to volunteer, you will complete one questionnaire, which will take about 10 minutes. Volunteering for this study does not involve risk beyond what a typical person would experience on an ordinary day. Since your involvement is voluntary, you may withdraw at any time for any reason. Please continue reading for more information about the study.

STUDY PERSONNEL: This research project is led by Jennifer Hull, a graduate student in the Master of Science in Human Genetics and Genetic Counseling program at Keck Graduate Institute.

PURPOSE: The purpose of this study is to learn genetic counselors' opinions about how genetic counseling sessions may change an individual's understanding, perceived value, and anxiety related to direct-to-consumer (DTC) and/or patient-initiated genetic testing.

ELIGIBILITY: To be in this study, you must be at least 18 years old and be a genetic counselor.

CONFIDENTIALITY: Your individual privacy will be protected in all papers, books, talks, posts, or stories resulting from this study. In order to protect the confidentiality of your responses, all data will be stored in secure files. Your information and your responses will not be disclosed to anyone outside of the research team. Participant names or other identifying information will not be used in the final research document.

FURTHER INFORMATION: If you have any questions or would like additional information about this study, please contact Jennifer Hull at jhull18@students.kgi.edu. You may also contact Jennifer's faculty advisor, Emily Quinn, LCGC, at (909) 607-6474 or Emily_Quinn@kgi.edu. The Claremont Graduate University Institutional Review Board (the governing IRB for the Claremont college consortium) has approved this project. If you have any ethical concerns about this project or about your rights as a human subject in research, you may contact the CGU IRB at (909) 607-9406 or at irb@cgu.edu.

CONSENT: Selecting the box below means that you understand the information on this form and that you voluntarily agree to participate in it.

- I agree with the above statement
- I do not agree with the above statement

Thank you for agreeing to take this survey. The purpose of this survey is to help understand genetic counselor experiences with **direct-to-consumer** test results and thoughts on **patient-initiated testing** (described below).

Direct-To-Consumer (DTC) Testing

Direct-to-consumer (DTC) testing includes genotyping through SNP (single nucleotide polymorphism)-based arrays. Commonly used DTC companies include 23andme and Ancestry, which provide consumers information on ancestry, certain SNPs associated with diseases, and other non-medical pieces of information.

Patient-Initiated Testing

Patient-initiated testing includes clinical genetic testing for disease risk, without the need to see a provider before testing is ordered. Examples of patient-initiated testing include Invitae's Genetic Health Screen, which analyzes 147 genes, and Helix's GenePrism, which analyzes the 59 genes on the ACMG secondary findings list (aka "ACMG 59" list). Both services include access to post-test genetic counseling.

First, please answer some demographic information so we can learn a little more about you.

Ethnic origin (select all that apply):

- American Indian or Alaskan Native
- Asian
- Asian Indian
- Black or African American
- Hispanic or Latin American
- Native Hawaiian or other Pacific Islander
- White or Caucasian

Prefer not to respond

Other, please specify:

Number of years you have worked as a genetic counselor:

Age (in years)

Gender

Male

Female

Other, please specify:

Zip code where you practice:

What is your area of practice (select all that apply)?

Clinical

Industry

Research

Other, please specify:

Reflecting on the total number of genetic counseling sessions you have facilitated in the last year, please answer the following questions:

Approximately what **percentage** of your total number of sessions included discussion of DTC (direct-to-consumer) test results?

Approximately what **percentage** of your total number of sessions included discussion of patient-initiated test results?

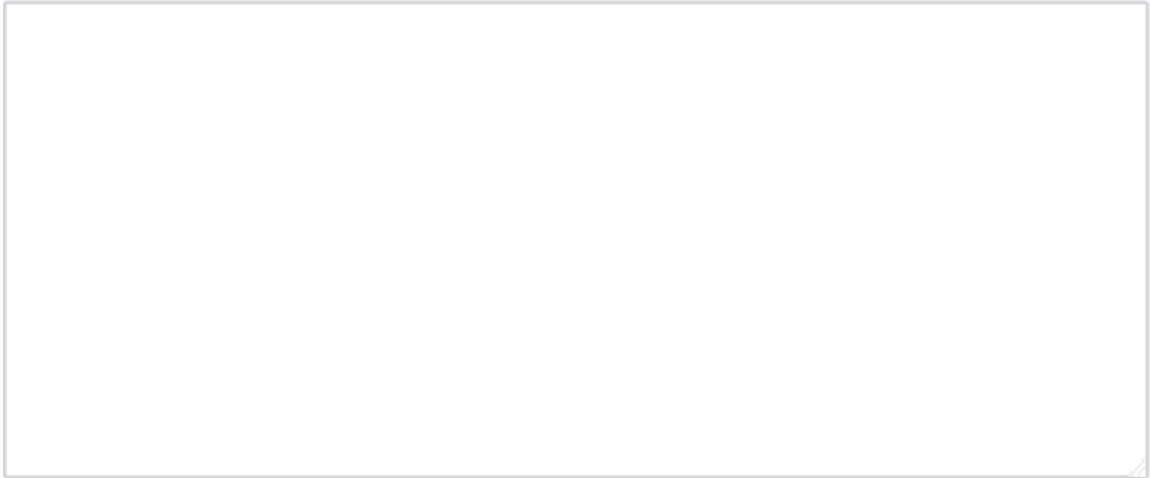
	Strongly disagree	Disagree	Neither agree nor disagree	Agree	Strongly agree	N/A	Unsure
Consumers understand the limitations of DTC testing after counseling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand how genetic variants can affect their health and disease risk before counseling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand how genetic variants can affect their health and disease risk after counseling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand their DTC result reports before genetic counseling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand their DTC result reports after genetic counseling	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

If you would like to provide additional comments regarding changes in patient understanding of results or anxiety levels with genetic counseling, please elaborate here:

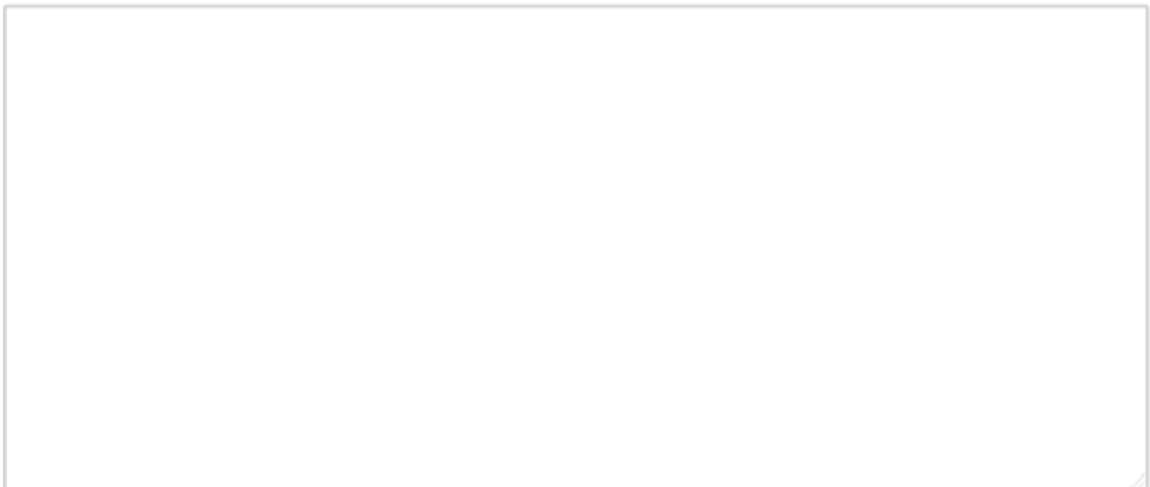
	Strongly disagree	Disagree	Neither agree nor disagree	Agree	Strongly agree	N/A	Unsure
I am concerned about how the role of clinical genetic counselors will change as DTC and patient-initiated testing options expand.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
As DTC and patient-initiated testing options expand, the need for genetic counselors is going to increase.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand the implications for long-term care, life, and disability insurance <u>before</u> ordering patient-initiated testing.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand the possible test results <u>before</u> ordering patient-initiated testing.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers ordering patient-initiated testing understand the health-related implications of a positive result.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers ordering patient-initiated testing understand the health-related implications of a negative result.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
Consumers understand how patient-initiated test results apply to family members.	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Finally, please answer some questions about your experience counseling about DTC results.

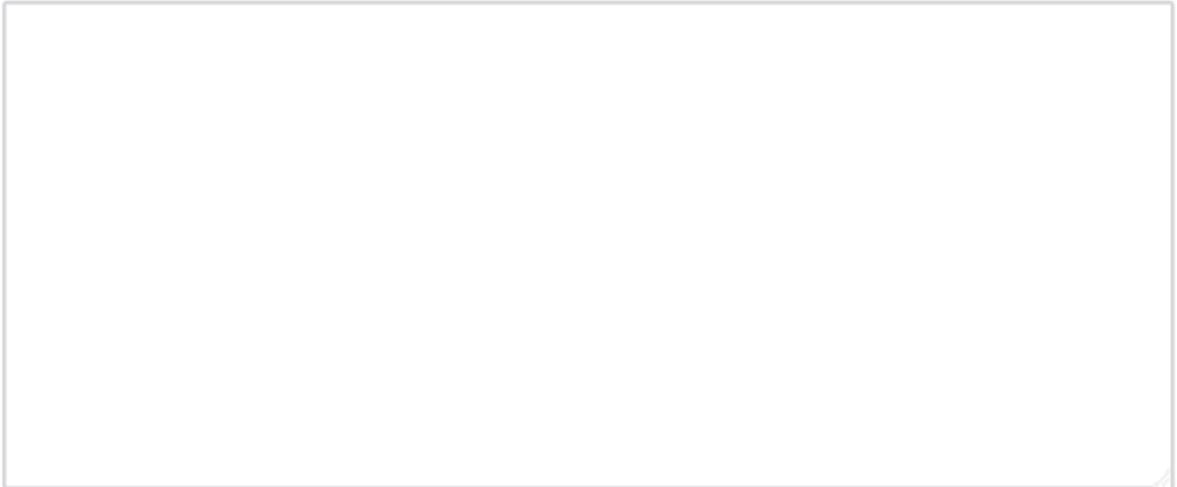
What information regarding DTC testing or DTC results do you think consumers find the most helpful?



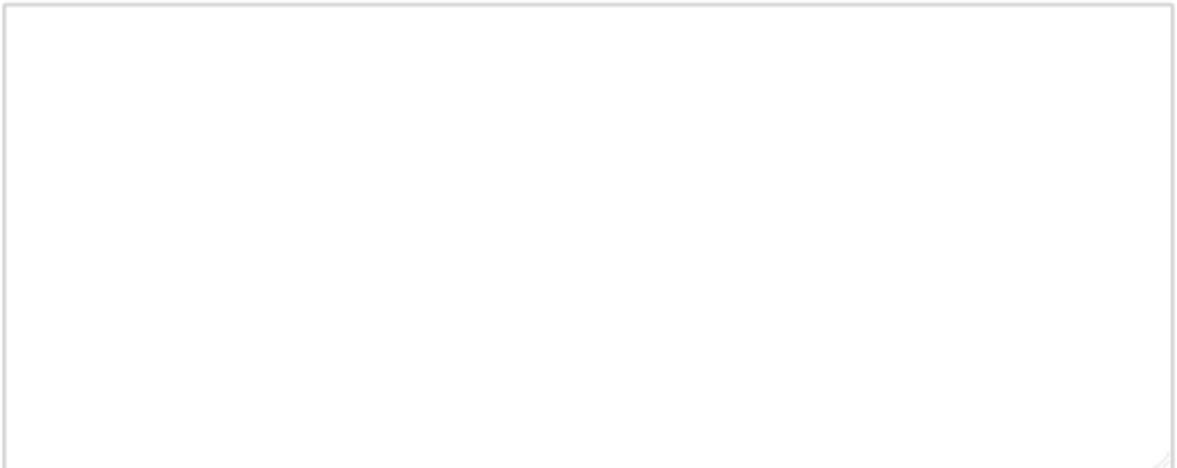
What misconceptions do you think consumers have regarding DTC results?



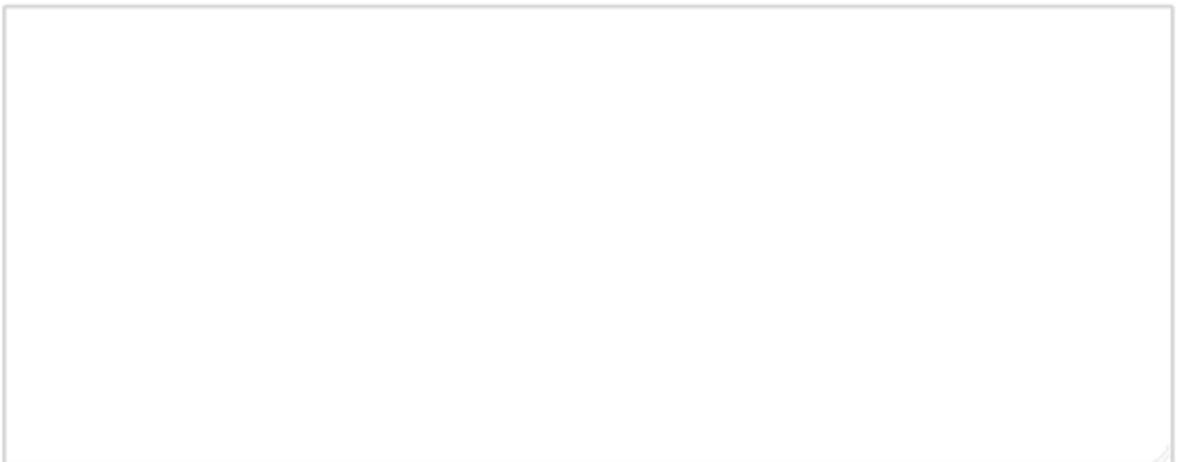
What are some challenges for consumers in dealing with DTC results?



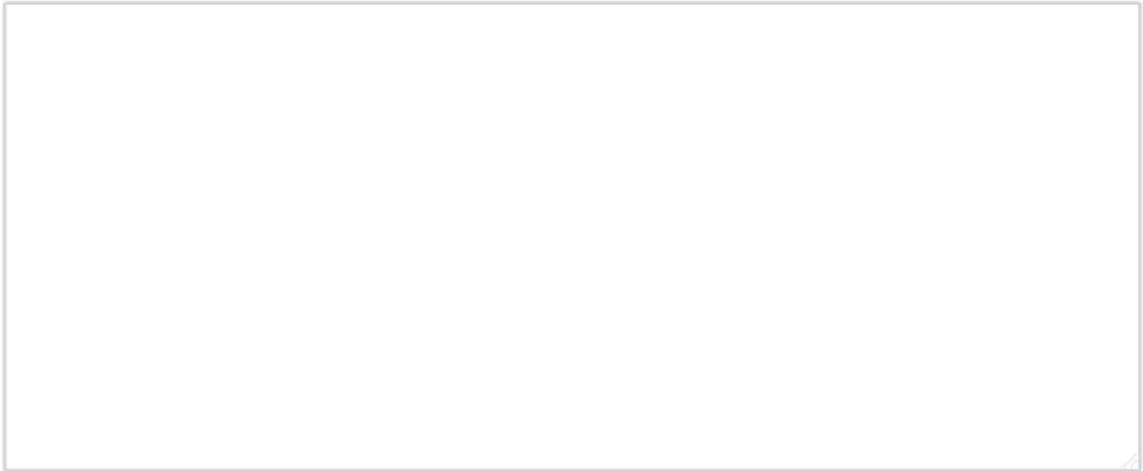
What are some challenges for genetic counselors in dealing with DTC results?



What are your experiences with or impressions of patient-initiated testing?



Do you have any additional comments you'd like to share about direct-to-consumer testing and/or patient-initiated testing?

A large, empty rectangular box with a thin grey border, intended for the user to provide additional comments. The box is currently blank.

Thank you again for your participation!

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