

THE IMPACT OF THE “AMERICAN SOCIETY OF BREAST SURGEON’S 2019
CONSENSUS GUIDELINES ON GENETIC TESTING FOR HEREDITARY BREAST
CANCER” ON CANCER GENETIC COUNSELORS

By

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ABSTRACT OF THE THESIS

The Impact of the “American Society of Breast Surgeon’s 2019 Consensus Guidelines on Genetic Testing for Hereditary Breast Cancer” on Cancer Genetic Counselors

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In February 2019, the American Society of Breast Surgeons (ASBrS) released a guideline recommending genetic testing be made available to all women with a breast cancer diagnosis. This guideline was based on findings in several papers outlining the number of individuals harboring mutations in a breast and ovarian cancer susceptibility gene who would not otherwise be found given the current National Comprehensive Cancer Network (NCCN) guidelines for cancer genetic testing (National Comprehensive Cancer Network, V1 2020). To determine the impact of this guideline on practicing cancer genetic counselors (GC), we created a survey asking GCs about their initial observations of the impact of the guideline on referral patterns from breast surgeons. Results showed 59% of respondents noted a change in referral patterns with breast surgeons referring more patients after the release of the ASBrS guideline. Thirty-seven percent of GCs reported that breast surgeons are referring all patients with breast cancer. GCs did not significantly change which breast cancer patients to whom they offered genetic testing. Our study found over 50% of cancer GCs agree that all women with a personal history of breast cancer should be offered genetic testing. This data supports increased coverage of genetic testing for these individuals as well exemplifies the increased demand for GCs in the cancer genetics setting.

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Introduction

In 2017 there were 268,600 new cases of female breast cancer in the United States ("American Cancer Society: Breast Cancer Facts and Figures 2017-2018," 2017-2018). Up to 10% of breast cancers are caused by heritable mutations ("American Cancer Society: Breast Cancer Facts and Figures 2017-2018," 2017-2018). Individuals with these mutations are diagnosed with hereditary breast and ovarian cancer (HBOC) syndrome. Identifying HBOC mutation carriers leads to increased surveillance, more effective treatment, and better preventative measures (Carbine, 2018; Narod, 2010; Trainer et al., 2010). Along with benefits to patient health, there are considerable cost savings for treatment of early versus later diagnosed breast cancer (Manchanda, Legood, et al., 2015).

Breast cancer patients have traditionally been referred to genetic counseling if they have a young age of onset, are triple negative, and/or have a family history of related cancers including breast, ovarian, and pancreatic cancer. Genetic counselors' (GC) roles are to assess the specifics of the patient's personal medical history as well as their family history to determine if the patient meets national guidelines for genetic testing as set by the principle guiding body, the National Comprehensive Cancer Network (NCCN). The GC then works with the patient to determine which specific genetic test should be ordered. Currently, there are genetic testing panels that offer testing of more than 80 genes. Informed consent for testing includes a discussion on the three possible results and the meaning of each for the patient as well as their family. It is also discussed that some results may not be actionable or have management guidelines and that there are limitations to the testing.

Guidelines for genetic testing were originally created by the NCCN to limit expensive testing to patients most likely to have a pathogenic variant. As the cost of testing has decreased, these guidelines have become less restrictive but more complicated and unclear as to which

specific genes should be tested. The most recent update to the criteria released in December 2019 begins with the heading, “Testing Criteria for High-Penetrance Breast and/or Ovarian Cancer Susceptibility Genes.” This often includes *BRCA1*, *BRCA2*, *CDH1*, *PALB2*, *PTEN*, and *TP53* among other. Yet, it is unclear which genes should be tested for when the patient meets testing guidelines. The guideline also states that testing is clinically indicated in 4 different scenarios with each scenario having a number of additional sub-bullets further restricting testing. For instance, scenario number 3 is headed, “Personal history of cancer” and then further clarifies which type of cancer, age of onset, and/or additional family history criteria the patient must meet. To add to the lack of clarity in the guidelines, there is an additional page listing scenarios in which testing “may be considered.” This guideline leaves much up to the interpretation of the ordering health care provider. Because of this, the specific genetic test/ panel offered to a patient could be different depending on the ordering health care provider and their testing practices (National Comprehensive Cancer Network, V1 2020).

Recent studies measured the number of individuals who harbor HBOC mutations that would have been missed if testing was restricted to those who meet NCCN guidelines. The first study, completed by Yang et. al in 2018 entitled “Underdiagnosis of Hereditary Breast and Ovarian Cancer in Medicare Patients: Genetic Testing Criteria Miss the Mark,” tested over 4000 Medicare patients with a 40-80 gene panel depending on the patient’s family history of cancer. The results indicated that 10.5% patients who met NCCN guidelines were found to have a likely pathogenic (LP) or pathogenic (P) variant while 9% of patients who did not meet NCCN guidelines were found to have a likely pathogenic or pathogenic variant. The authors concluded that “A substantial number of Medicare patients with clinically actionable genetic variants are being missed by current testing criteria and suggest the need for significant expansion and simplification of the testing criteria for HBOC” (Yang et al., 2018). This study was unique in its

approach to use Medicare patients as study subjects. Due to their advanced age, many would have already had a personal history of cancer but the age of onset would have been wide-spread. Additionally, Medicare is often the first insurance group to update its policies around genetic testing coverage with private insurance companies following suit soon after. One criticism of this study was its inclusion of genes with autosomal recessive increased risk for cancer, such as *MUTYH*, rather than only those with autosomal dominant inheritance. Even with this criticism, this study has been influential as it was one of the first to acknowledge that NCCN guidelines misses up to 50% of individuals harboring HBOC mutations.

A similar study by Beitsh et. al in 2019 summarized the results of testing by an 80-gene panel on 1000 patients with breast cancer, half who met NCCN guidelines and half who did not. The results showed 9.39% of those who met guidelines were found to have a P or LP variant and that 7.9% of those who did not meet NCCN guidelines were found to have a pathogenic or likely pathogenic variant. They concluded “Nearly half of patients with breast cancer with P/LP variant with clinically actionable and/or management guidelines in development are missed by current testing guidelines. We recommend that all patients with a diagnosis of breast cancer undergo expanded panel testing.” This study was also criticized for including autosomal recessive HBOC genes (Beitsch et al., 2019).

Based off of the findings in both of these studies, in February 2019, the American Society of Breast Surgeons (ASBrS) released an official statement which recommends, “Genetic testing should be made available to all patients with a personal history of breast cancer” (“Consensus Guideline on Genetic Testing for Hereditary Breast Cancer,” 2019). It is unknown how this recommendation will change the patient population for HBOC genetic testing and how this will impact cancer genetic counselors. Of note, this guideline was also unclear as to which genes should be included in testing of patients as it says “testing should include *BRCA1/BRCA2* and

PALB2, with other genes as appropriate.” Similar to NCCN guidelines, this guideline is also unclear as to which genes should be included in the testing of patients and leaves the specifics to the discretion of the ordering health care provider.

To analyze the impact of the new ASBrS guideline as perceived by cancer GCs, a survey was developed with two main goals. The first was to assess whether there was an increase in the number of patients referred by breast surgeons due to the ASBrS guideline. The second goal aimed to determine if GCs have changed their practice with respect to which patients are offered genetic testing. Additional goals of the survey were to gather GCs’ personal opinions on the utility and practicality of testing all patients with breast cancer, and to garner patient’s tendency to pursue testing when they do not meet NCCN guidelines for testing.

Material and Methods

Survey development

This survey was developed to explore the impact of the ASBrS guideline updates on the genetic counseling community. The survey began with demographic questions which were developed following those used by the 2019 NSGC Professional Stats Survey. Additional questions were developed to assess the GCs experience with cancer genetic testing as well as the patient population they served. The second part of the survey was centered on GCs perceived changes in breast surgeon's referral patterns. The next portion contained questions on the GCs' practices when ordering genetic testing for patients referred due to the ASBrS guideline. The last portion of the survey asked for the GC's personal opinions on the ASBrS guideline.

Survey distribution

A survey was created and distributed to all GCs who are members of the National Society of Genetic Counselors (NSGC) as part of a weekly digest of student research surveys under the subsection of cancer research (Appendix A). This is part of the NSGC's Student Research Protocol Program as described on NSGC.org (Appendix B). The survey was originally sent out on November 27, 2019 and a reminder email was sent one week later on December 4, 2019. Responses that only answered demographic information were removed from the data set. This proposed protocol was approved by the Rutgers University Health Sciences Institutional Review Board.

Data analysis

Qualtrics was used for distribution and collection of the survey. Survey answers were sent to a link at Qualtrics where data was stored in a password protected electronic format. No identifying information such as names, email addresses, or IP addresses were collected by Qualtrics. Therefore, the responses were anonymous.

Statistical analysis was completed using Qualtrics for all surveys received that answered questions beyond demographics. Descriptive statistics and chi squared analysis were used depending on the question type and the sample size. The percentage of the total number of responses was computed for questions that allowed for only one response. Results for questions that allowed for more than one response were computed as a percentage of those who checked the response over the total sample size for the question.

Visual charts were created to show the trends of answers collected. Additional comments submitted by counselors were used to develop discussion on plausible next steps for the genetic counseling profession and breast surgeons who are ordering additional testing for patients.

Results

Summary of Results

The survey was issued to all GCs who are members of NSGC. Through analysis of survey responses, we discovered there was an increase in the number of referrals, although the specific patterns of referrals as well as GCs' personal opinions on the guideline varied widely. Most patients who did not meet NCCN testing criteria still proceeded with testing. The most common deterrent to testing was high cost.

Demographics and Patient Population

A total of 63 responses were started and 52 surveys were fully completed through Qualtrics. The response rate for the survey was 6.7% based on the approximate number of clinical cancer GCs (773) who are members of NSGC according to the 2019 Professional Status Survey (NSGC, 2019).

The gender and years of experience of our survey respondents were consistent with that of the total population of practicing genetics counselors according to the 2019 NSGC professional practice survey with 92.5% of survey participants being female ($p > .72$). Our study population was younger than the NSGC population with 90.5% of survey respondents under the age of 40 compared to 70% of individuals in NSGC ($p < .004$). Additionally, 62.3% of survey respondents had five or less years of experience as a genetic counselor.

Respondents worked in a variety of settings and from regions throughout the United States. Specific respondent characteristics can be found in Table 1.

Survey respondents were asked about their patient population. Most of the respondents (77.4%) reported over 75% of their patients were seen for hereditary cancer genetic counseling. There were three individuals who reported only 0-25% of their patients being seen for a cancer

indication. The percentage of GCs who had less than half of their patients covered by Medicare was 92.11%.

<u>Table 1</u>	
<i>Genetic Counselor Demographic Information</i>	
<u>Characteristic</u>	<u>Number (%)</u>
Gender	
Female	49 (92.5)
Male	4 (7.5)
Age	
20-29	29 (54.7)
30-39	19 (35.8)
40-49	4 (7.5)
Prefer not to respond	1 (1.9)
Years practicing	
1-2	22 (41.5)
3-5	11 (20.8)
6-8	8 (15.1)
9-11	5 (9.4)
11+	7 (13.2)
Type of Institution	
Academic Medical Center	22 (41.5)
Large Hospital System	15 (28.3)
Community Hospital	14 (26.4)
Private Genetic Counseling Company	2 (3.8)
Geographic Region	
Region 1: CT,MA,ME,NH,RI,VT, Canada	6 (11.3)
Region 2: DC,DE,MD,NJ,NY,PA,VA,WV	15 (28.3)
Region 3: AL,FL,GA,KY,LA,MS,NC,SC,TN, (Puerto Rico, Virgin Islands)	11 (20.8)
Region 4: AR,IA,IL,IN,KS,MI,MN,MO, ND,NE,OH, OK,SD,WI	
Region 5: AZ,CO,MT,NM,TX,UT,WY	12 (22.6)
Region 6: AK,CA,HI,ID,NV,OR,WA	5 (9.4)
% of patients seen for hereditary cancer risk	4 (7.5)
0-25%	
26-50%	3 (5.7)
51-75%	3 (5.7)
76-100%	6 (11.3)
Patients seen per month	41 (77.4)
0-10	
11-20	1 (1.9)
21-30	3 (5.7)
31-40	10 (18.9)
41-50	18 (33.9)
50+	10 (18.9)
	11 (20.7)

<i>Clinical Setting Demographic Information</i>	
<u>Characteristic</u>	<u>Number</u>
Patients with Medicaid	
0-25%	39 (75.6)
26-50%	10 (18.9)
51-75%	2 (3.8)
76-100%	2 (3.8)
Patients with Medicare	
0-25%	25 (41.2)
26-50%	27 (50.9)
51-75%	0 (0.0)
76-100%	1 (1.9)
Patients uninsured or charity care	
0-25%	49 (92.5)
26-50%	3 (5.7)
51-75%	0 (0.0)
76-100%	1 (1.9)
Patients identifying as minority	
0-25%	26 (49.1)
26-50%	20 (37.7)
51-75%	3 (5.7)
76-100%	2 (3.8)
Unsure	2 (3.8)

Changes in breast surgeon's referral patterns

GCs were asked if they observed any changes in breast surgeons' referral patterns after the ASBrS guideline was released. We found over half of GCs (22.6+35.8=59%) surveyed reported a change in the referral patterns of breast surgeons, with 41.5% of GCs reporting breast surgeons referring more patients than they did prior to the release of the ASBrS guideline (Table 2).

A chi-square test of independence was performed to examine the relationship between GC's assessment of breast surgeons' referrals before and after the release of the guideline. The data suggests there was a significant relationship, with GCs reporting breast surgeons were more likely to refer more patients after the release of the guideline, $X^2(1, N=53) = 8.4, p < .038$ (Figure 1). An analysis of responses stratified by years of experience showed this did not lead to a significant difference in GCs reports of breast surgeon's referrals ($p > .08$).

The specific patterns of referral by breast surgeons as reported by GCs varied widely. Approximately 33% of breast surgeons were referring according to the ASBrS guideline while 25% continued to refer according to NCCN and the remaining 42% not following either guideline.

Chi square analysis revealed that although referral patterns have increased in some cases, GCs opinions on the guideline (whether they agree or do not agree with the guideline) do not appear to have any influence on changes in breast surgeon's referral practices ($X^2(1, N=53) = 8.4$, $p = 0.181$).

<u>Table 2</u> <i>GCs' reported observations in the changes of referral patterns by breast surgeons</i>	<u>Number of GCs responding (%)</u>
Have you seen a change in the referral pattern of breast surgeons since the release of the ASBrS guideline?	
Yes	12 (22.6)
No	22 (41.5)
Only for some breast surgeons	19 (35.8)
Which of the following patterns have you seen in the referring practice of breast surgeons since the release of the ASBrS guideline?	
Referring in the same pattern	
Referring more patients	27 (50.9)
Referring less patients	22 (41.5)
	4 (7.5)

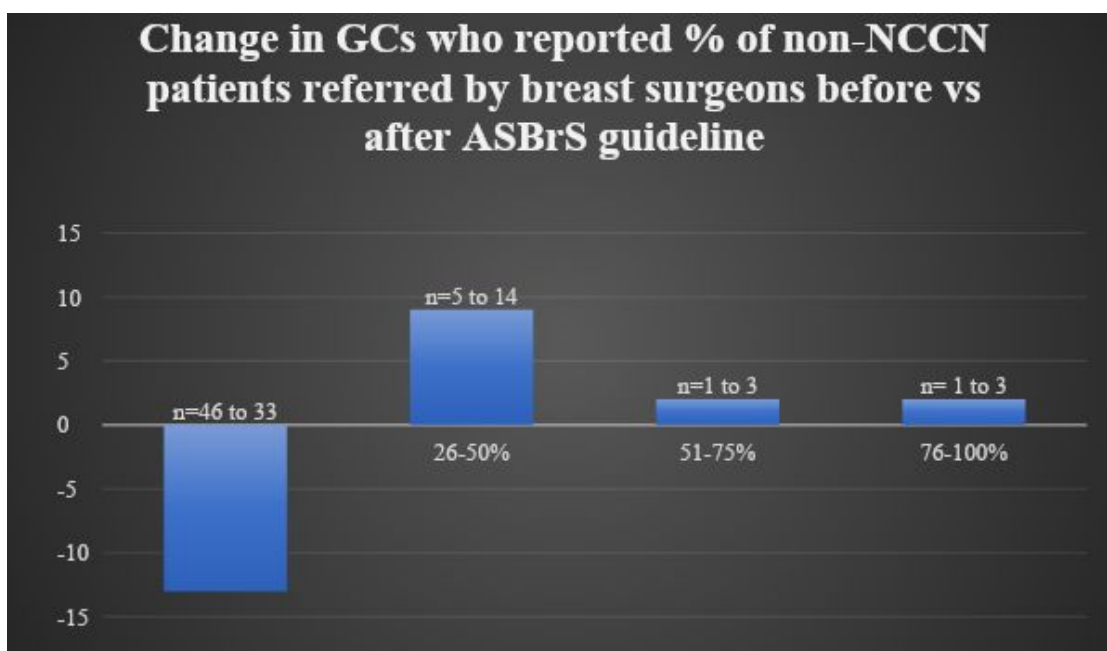


Figure 1- Change in GC reported percentage of non-NCCN patients referred by breast surgeons before vs after the ASBrS guideline. For example, after the release of the guideline, 13 less GCs reported that 0-25% of patients referred by breast surgeons did not meet NCCN guidelines.

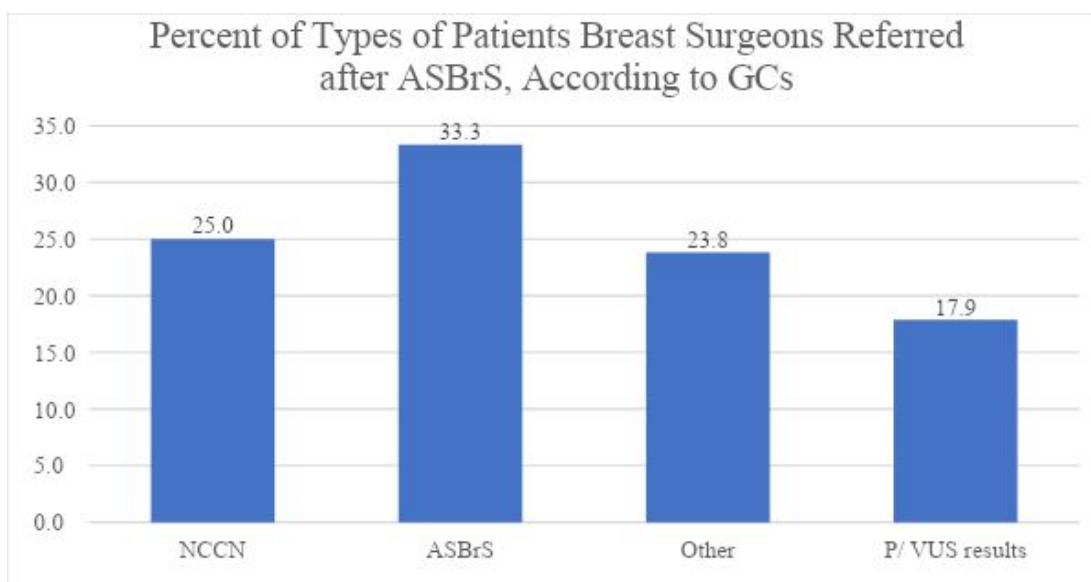


Figure 2- Percent of type of patient's breast surgeons referring for genetic counseling as reported by GCs.

Genetic counselor's approach to testing for those who do not meet NCCN guidelines

To establish how GCs handle patients who do not meet NCCN guidelines, we asked how often and what type of testing was being offered. Before the release of the guideline, 56% of GCs were offering testing to 76-100% of their patients. This increased to 66% after the guideline. Only 8/53 or 15% of GCs increased the number of patients they offered testing to after the release of the guideline. A chi-square test of independence was performed to determine if there was a significant change in the percent of patients GCs offered testing to before and after the release of the guideline. The percent of patients who were offered testing did not differ from before to after the release of the guideline, $\chi^2 (1, N=53) = 4.3, p < .23$. There was not a significant difference in the number of GCs who increased their referral rate when stratified by those who reported an increase in the number of patients referred as compared to those who did not ($p > .65$). There was a significant difference in how often GCs offered genetic testing to patients who do not meet NCCN guidelines when separated by years of experience in genetic counseling. Those with more experience (6+ years), were most likely to be offering testing 76-100% of the time (85% of this group).

When GCs do offer testing to patients who do not meet NCCN guidelines, they most often leave the choice in type of gene panel up to the patient (Figure 3). GCs were also asked why they were not offering testing to patients who have a personal history of breast cancer but do not meet NCCN guidelines. As seen in Figure 4, the most common reasons were the GC believing genetic testing was not warranted based on the patient's family history and the large out of pocket cost for the patient (41.8%). These results were not significantly different based on region or the demographics of the GCs patient population. GCs with few years of experience before the release of the guideline reported responses that were not statistically different from GCs with more years of experience.

Table 3 <i>Change in practice of offering testing to non-NCCN patients by GC by years of experience, % as total of row.</i>		
	1-5 years	6+ years
Decreased	3.2	0
Stayed the Same	40.6	5
Increased	15.6	10
Stayed 76-100%	40.6	85

Type of testing offered to patients who do not meet NCCN

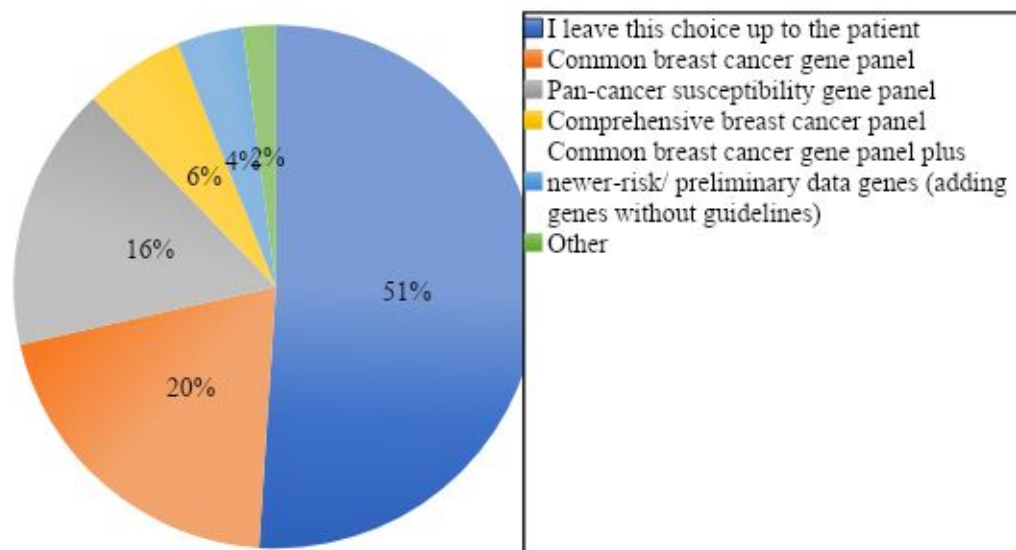


Figure 3- Type of genetic testing panel offered by GCs to patients who do not meet NCCN guidelines.

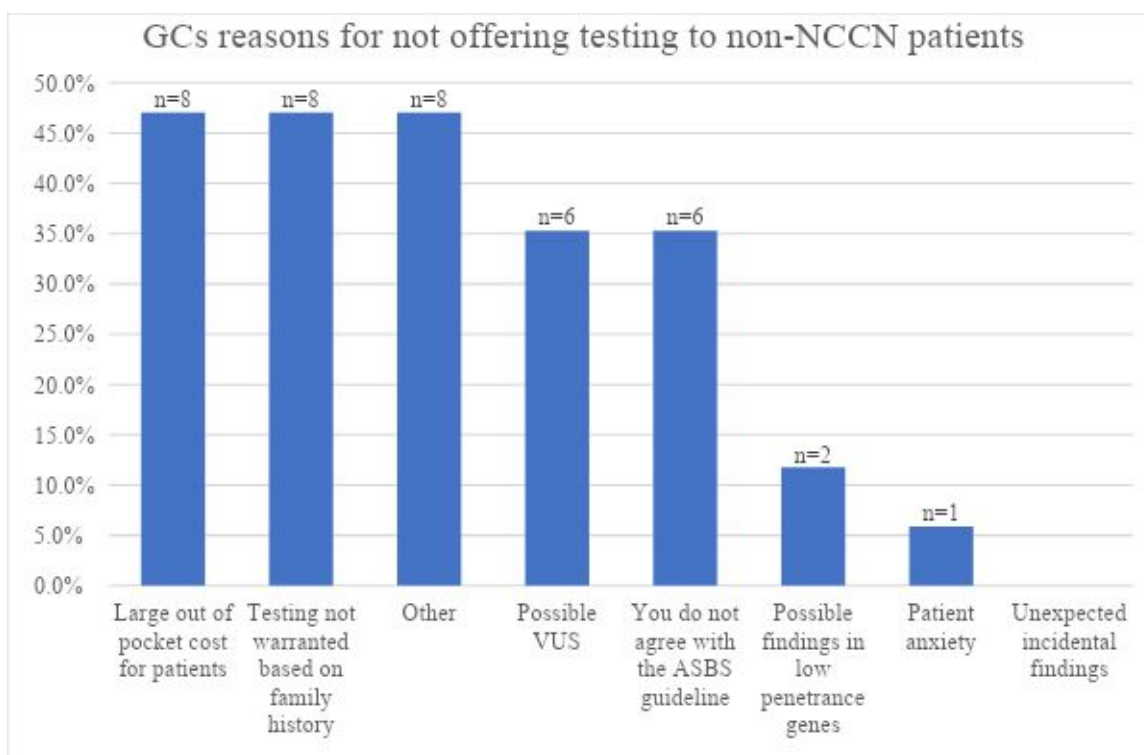


Figure 4- Reasons GCs do not offer testing to patients who do not meet NCCN guidelines but do meet ASBrS guidelines. Limited to 17 respondents who reported not offering testing to all patients.

Patients reactions to the offer of genetic testing

To determine how often patients who have a personal history of breast cancer but do not meet NCCN guidelines consent to genetic testing, we asked survey respondents if these patients were more or less likely to pursue testing as compared to patients who do meet NCCN guidelines.

Almost 57% of GCs said these patients are more likely or just as likely to pursue testing as patients who meet NCCN guidelines (Table 4). We also asked how often GCs had patients who did not meet NCCN guidelines consent to genetic testing. Most GCs (79.3%) said more than half of these patients consented to genetic testing (Table 5). The most common reason for these patients not consenting to genetic testing was the large out of pocket cost (64.8%), followed by

genetic testing not being a priority at this time (57.4%), and believing a mutation would not be found (50.0%, Figure 5).

<u>Table 4</u>		
<i>As compared to patients who do meet NCCN guidelines, are those who do not meet guidelines more or less likely to pursue genetic testing?</i>	Number	Percent
About the same	27	50.9
Less likely	23	43.4
More likely	3	5.7

<u>Table 5</u>		
<i>For all patients who do not meet NCCN guidelines, how often do they pursue genetic testing?</i>	Number	Percent
0-25%	8	15.1
26-50%	3	5.7
51-75%	18	34.0
76-100%	24	45.3

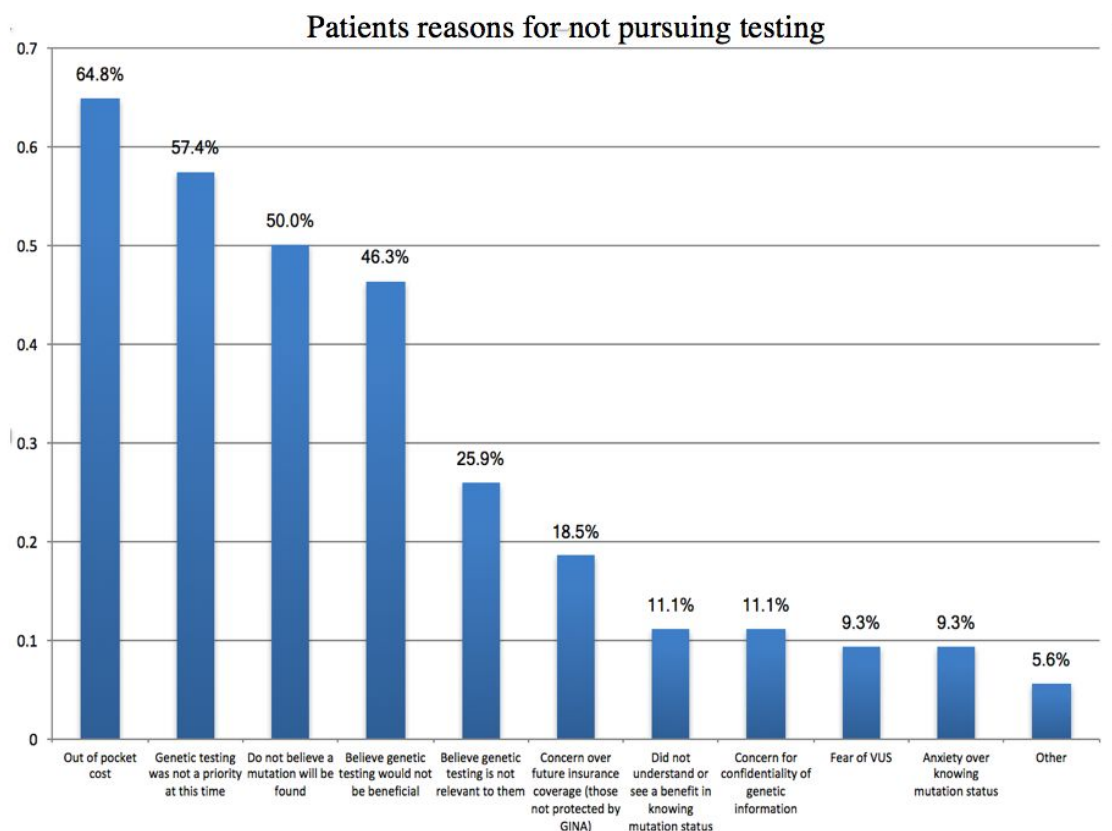


Figure 5- Reasons why patients who do not meet NCCN guidelines do not pursue genetic testing as reported by genetic counselors.

GCs opinion on ASBrS guideline

In order to determine GCs' opinions on the referral of patients who do not meet NCCN guidelines, respondents were asked if they agreed with the ASBrS guideline. GCs appear to have a near even split on their opinions towards the guideline with a large number of respondents (28.8%) reporting being unsure (Figure 6). Those who did not agree with the guideline most often stated their reasoning for not agreeing as insufficient data to support the guideline (Figure 7). These results were not significantly different based on the demographics of the GC including their age or years of experience.

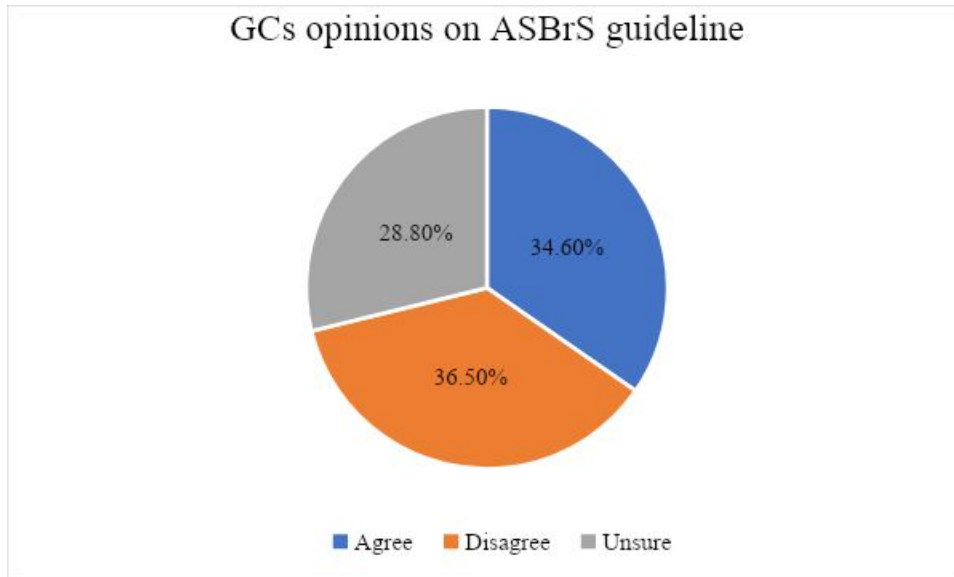


Figure 6- Percentages of respondents who agree, disagree, or unsure about the ASBrS guideline.

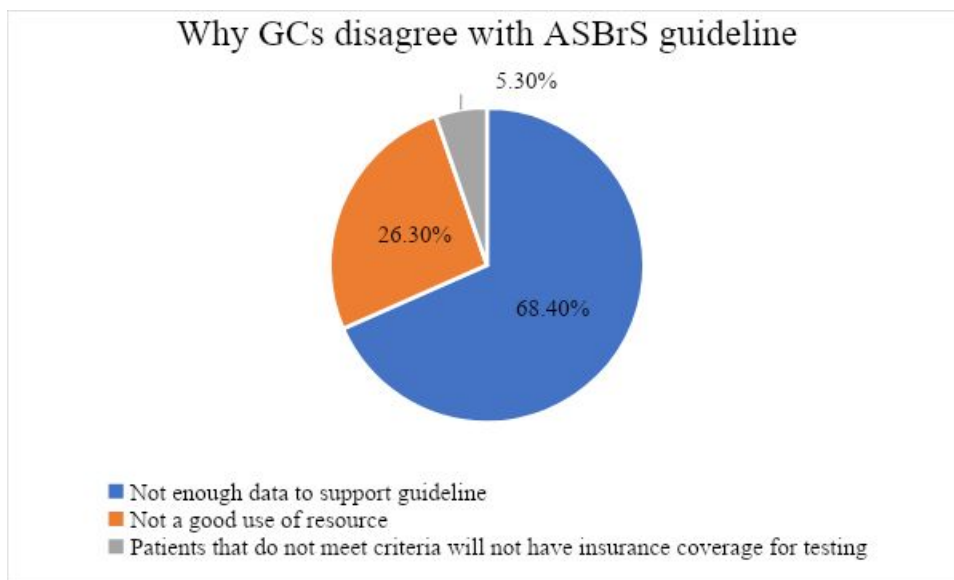


Figure 7- Why respondents disagree with the ASBrS guideline.

Genetic Counselors expanded thoughts on the guideline

Survey respondents were given an opportunity at the end of the survey to express any additional thoughts surrounding the new guideline. As far as the referrals from breast surgeons, three GCs noted that breast surgeons were warning patients of the potential out of pocket cost for testing and were therefore reducing the number of uninterested patients before they were sent to genetics.

One GC commented, “Our breast surgeons tend to offer [testing] to all patients and they do tell them that the testing will likely be out-of-pocket at \$250. So, patients seem prepared to pay this when they meet with me.” Another GC said, “Surgeons/oncologists usually mention it to patients and only refer those who are interested despite cost.”

One GC commented on breast surgeons strictly following the guideline. “I’ve also seen the breast surgeons become more adamant about ordering testing regardless of potential or known insurance issues, ‘because that’s what the guideline says.’”

Four GCs noted that the guideline has not had an impact on their offer of genetic testing to patients as they have always offered genetic testing to all patients. “I also offer genetic testing to any patient I see because it’s their right to pursue genetic testing and I would rather they do it through a GC then go out into the community and have someone else order it. So whether or not they meet criteria I give them the option to pay out of pocket for the testing.”

Finally, some used this free response section to discuss broader impacts of the ASBrS guideline on the GC community. One GC commented, “Though controversial, this guideline has sparked important conversations within our program on how to increase our patient load without sacrificing quality of care.” Another GC wrote, “I think it has pushed us to get ready for this change, when ultimately the data does support it. It has been interesting to deal with it when it was based on a study including genes it maybe shouldn’t have and has allowed for good teaching

moments.” Finally, another GC pointed out, “We need more support to develop alternative service delivery models that do not completely bypass GCs.”

<i>Table 6- GCs free Responses</i>
In Ontario we can only offer testing based on ministry of health criteria so this has had limited impact on the referral process and who I am allowed to offer testing to
We have additional internal data outside of the Beitsch paper which is why we offer all patients with breast cancer genetic testing.
We need more support to develop alternative service delivery models that do not completely bypass GCs
It's sometimes difficult to answer these questions because my institution is slower on the uptake of testing all breast patients. I also offer genetic testing to any patient I see because it's their right to pursue genetic testing and I would rather they do it through a GC then go out into the community and have someone else order it. So whether or not they meet criteria I give them the option to pay out of pocket for the testing.
Surgeons/oncologists usually mention it to patients and only refer those who are interested despite cost so my answers are skewed.
It should be noted that many of the patients that tested positive in the ASBS study were carriers of a <i>MUTYH</i> mutation. There was not a large enough number of women testing positive for a clinically actionable gene to warrant changes in practice. Thankfully, the breast surgeons at my institution value the opinion of the genetic counselors and understand that testing an 88 year old woman with breast cancer and no family history is not a good use of resources.
I find these blanket referral guidelines somewhat frustrating. Many of the patients that are only referred based on the ASBS guidelines are older, have many more health concerns, and sending them for another appointment can be challenging. Once I tell them that there is not much

<p>chance of finding a mutation, they often would have rather not come to see me, since time and travel are challenging for them.</p>
<p>Just FYI - I offer testing to all patients, but it may be out of pocket rather than insurance if they do not meet NCCN criteria</p>
<p>Our breast surgeons tend to offer to all patients and they do tell them that the testing will likely be out-of-pocket at \$250. So patients seem prepared to pay this when they meet with me.</p>
<p>Though controversial, this guideline has sparked important conversations within our program on how to increase our patient load without sacrificing quality of care.</p>
<p>I think there is benefit to offer testing to all patients with breast cancer with proper counseling prior to testing. I've seen an increase in the number of confused or upset patients with a positive or VUS results who were not expecting these results. I've also seen the breast surgeons become more adamant about ordering testing regardless of potential or known insurance issues "because that's what the guideline says". I have also noticed a significant increase in the presence of labs reps pushing this testing for the breast (and other) surgeons.</p>
<p>I think it has pushed us to get ready for this change, when ultimately the data does support it. It has been interesting to deal with it when it was based on a study including genes it maybe shouldn't have and has allowed for good teaching moments. I think it depends on how communicative the genetics department and other departments may be as to how this impacts communities. We are a very close community here. It hasn't changed our practice as we will offer self pay testing to anyone but it certainly has pushed us to keep thinking about the future and how we will handle it.</p>

Discussion

The goal of our study was to describe the initial impact of the ASBrS guideline on the practice of GCs. Our findings showed a significant number of GCs reported seeing an increase in the number of breast cancer patients referred for genetic testing after the release of the guidelines in February 2019. Our findings demonstrate that although more patients were referred to GCs, there was not a significant change in GC testing strategy. Many GCs are still performing risk assessment from medical and family history to deduce which patients are most likely to have a pathogenic finding and offering genetic testing accordingly. Although the guideline suggests testing should be expanded to more patients, each patients' unique medical situation as well as a psychosocial assessment of the patient must be used to determine the best course of action for offering genetic testing. This preliminary evidence shows GCs may be moving away from complete patient autonomy and non-directive practices that have been at the core of GCs guiding principles since the founding of the profession.

Interestingly, the results of this survey also revealed that GCs with over five years of experience were more likely to offer genetic testing to all of their patients, regardless of their family history or which guidelines they meet. This could perhaps be due to these GCs who have been practicing longer having more experience with pathogenic results found in patients they did not expect to test positive. Meanwhile, less experienced GCs may be more restrictive in who they offer testing to and stick closely to the NCCN guidelines they were taught to use in their graduate education program. We suggest additional studies be completed to better understand this shift in GCs use of non-directive counseling.

GCs reported breast surgeons referring patients for a variety of different reasons. Some surgeons seem to be referring all patients to genetic counseling before testing while others may be doing pretest counseling on their own and have thus decreased their referrals to GCs except in the

case of positive results. As far as communicating positive results, according to the GCs surveyed in our study, breast surgeons seem to be universally referring patients to GCs for counseling of positive and uncertain genetic testing results. GCs are advanced medical professionals with in-depth knowledge and understanding of the unique intricacies of genetic testing results and guide patients and their families. Here we propose that, if there are not enough GCs to see additional patients, alternative service delivery models will need to be developed. As one GC in the survey stated, “We need more support to develop alternative service delivery models that do not completely bypass GCs.” Previous studies have suggested alternative models and supported the efficacy of these models. These models include telephone counseling, telegenetics or counseling by video platforms, and pretest counseling in a group setting. These models were shown to increase access to GCs and lead to the same level of patient satisfaction as the traditional face-to-face model of genetic counseling (Buchanan, Rahm, & Williams, 2016).

An increase in genetic testing of patients also shows the need for proper education of breast surgeons and other healthcare providers on the importance of pretest counseling and informed consent for genetic testing. At this time, there is a lack of education about genetics in both medical school and in continuing education for physicians (Haspel & Saffitz, 2014). There is near universal support to increase education on these topics and physicians have commented on their desire and need to develop their understanding of genetics and genetic testing as personalized medicine continues to be viewed as the future of medical care (Haspel & Saffitz, 2014).

An additional critical finding in this study was the varied approach to which specific genetic test should be ordered for patients. The ASBrS has not made a statement on which genes should be included in the genetic testing of patients who do not meet NCCN guidelines or have a limited family history of cancer. As the studies the ASBrS guideline was based on included

testing for 80 genes, GCs would need to be offering a gene panel as extensive as this to achieve the same increase in patients found to have pathogenic mutations (Beitsch et al., 2019; Yang et al., 2018). Based off the results of this survey, this does not appear to be the case.

NCCN guidelines are also unclear as to which genes should be included in testing for patients who meet or do not meet their guidelines. The decision regarding breadth and depth of testing to be ordered remains at the discretion of the ordering health care provider after evaluating factors including family history, patient preferences, insurance coverage and turnaround time for results. This evaluation leaves a lot of room for variation between providers ordering genetic testing and this corresponds to the varied responses seen in our survey results. More guidance regarding what testing should be ordered for the patients may be useful at this time as there are options to order 80 gene panels at the same cost as single site testing.

Research studies have continually found that it is most cost-effective to do cancer genetic testing at a population level, meaning prior to a cancer diagnosis (Manchanda, Legood, et al., 2015; Manchanda, Loggenberg, et al., 2015). This would allow for patients screening and management to be individualized based on their genetic testing results in combination with their age and family history. This approach lessens the number of individuals found with late stage cancer as those with higher risk would be screened earlier in life and more frequently. Although this is the ideal approach to cancer screening, cancer population screening using genetic testing is not possible at this time because insurance companies continue to restrict the coverage of genetic testing. The hope is that the results of this study and others will persuade insurance companies to increase coverage for genetic testing and thereby decrease health-care costs in the long run.

Additional research should be done to show the increased number of patients being seen for HBOC who do not meet NCCN guidelines. Ideal studies would include a retrospective chart review of patients referred by breast surgeons and which guidelines they do or do not meet. A

survey to breast surgeons would also be useful as it would allow researchers to understand the reasoning behind their referral practices. Another suggestion would be to survey GCs again at a later time, after the guideline has had more time to settle in. The number of breast surgeons who follow the guideline may continue to increase with more time and communication amongst the community.

Limitations of this study included a small sample size and the number of GCs who reported less than two years of experience in the field. This could result in a false understanding of shifting referral patterns by breast surgeons. In addition, three respondents to the survey reported 0-25% of their patient population were referred for a cancer indication. They were left in the study as they responded to questions regarding breast surgeon referrals and therefore must have some experience with breast surgeons. More consideration should have been put into survey development in terms of which questions would ask for a “select all that apply” response verses those that were “choose one” response. This would have allowed for GCs to more accurately define their answers as well as helped to better define the meaning of responses. Additionally, the survey was sent to GCs and asked them to approximate the referral rates of breast surgeons, rather than gathering data directly from patient logs or breast surgeons. This study method could also be biased due to the GCs’ personal opinion on the guideline as well as recollection bias as the survey asked GCs to estimate referral rates from the previous 10 months.

In summary, our study results indicate an increase in the number of referrals from breast surgeons to GCs since the release of the guideline. Our findings suggest that with this increase in number of referrals, there is a greater demand for cancer GCs and indicate the need for alternative service delivery models as well as education of other healthcare providers on proper pretest genetic counseling. As more patients with a personal history of breast cancer who do not meet NCCN guidelines are referred for genetic testing, we hypothesize there will also be an impact on

which patients' insurance companies are willing to provide coverage for this testing. We propose that additional research is needed to show the increased number of patients being seen for genetic evaluation of HBOC as well as the rate of positive results in the population of patients who do not meet NCCN guidelines. These results may impact insurance coverage, genetic education, and the need for alternative service delivery models for genetic counseling.

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Appendix A

Survey Questions

Demographics of patients seen

1. Approximately what percentage of your patients are seen for evaluation of risk for hereditary cancer?
 - a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
2. How many patients do you see monthly?
 - a. 0-10
 - b. 11-20
 - c. 21-30
 - d. 31-40
 - e. 41-50
 - f. 50+
3. Approximately what percentage of your patients are on Medicaid?
 - a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
 - e.
4. Approximately what percentage of your patients are charity care or uninsured?
 - a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
5. Approximately what percentage of your patients are on Medicare?
 - a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
6. Approximately what percentage of your patients identify as a minority?
 - a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
 - e. Unsure

Demographics of GC's

7. How many years have you been practicing as a genetic counselor?
 - a. 1-2
 - b. 3-5
 - c. 6-8
 - d. 9-11
 - e. 11+
8. What type of institution do you work in?
 - a. Academic medical center
 - b. Community hospital
 - c. Large hospital system
 - d. Lab setting
 - e. Private genetic counseling company
9. What geographic region is your institute based in?
 - a. Region 1: CT,MA,ME,NH,RI,VT, Canada
 - b. Region 2: DC,DE,MD,NJ,NY,PA,VA,WV
 - c. Region 3: AL,FL,GA,KY,LA,MS,NC,SC,TN, (Puerto Rico, Virgin Islands)
 - d. Region 4: AR,IA,IL,IN,KS,MI,MN,MO, ND,NE,OH, OK,SD,WI,
 - e. Region 5: AZ,CO,MT,NM,TX,UT,WY
 - f. Region 6: AK,CA,HI,ID,NV,OR,WA
10. What is your age?
 - a. 18-29
 - b. 30-39
 - c. 40-49
 - d. 50-59
 - e. 60+
 - f. Prefer not to respond
11. What is your gender
 - a. Male
 - b. Female
 - c. Other
 - d. Prefer not to respond

Questions on the referral patterns of breast surgeons

12. Have you seen a change in the referral patterns of breast surgeons since the ASBS guidelines were released?
 - a. Yes
 - b. No
 - c. Only for some breast surgeons
13. Of all patients seen for hereditary breast and ovarian cancer in an average month before the ASBS guidelines were released, approximately how many patients were referred by breast surgeons for a personal history of breast and did not meet NCCN guidelines?

- a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
14. Of all patients seen for hereditary breast and ovarian cancer in the past month, approximately how many patients were referred by breast surgeons for a personal history of breast and did not meet NCCN guidelines?
- a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
15. For all patients referred due to the new guideline, how often do they pursue testing, regardless of whether they end up meeting NCCN guidelines or not?
- a. 0-25%
 - e. 26-50%
 - f. 51-75%
 - g. 76-100%
16. Which of the following patterns have you seen in the referring practice of breast surgeons since the release of the ASBS guideline?
- a. Referring more patients than before
 - b. Referring less patients than before
 - c. Referring in the same pattern as before
17. What types of patients are breast surgeons now referring? Choose all that apply
- a. Only referring patients with pathogenic or VUS results
 - b. Only referring patients with a family history of cancer
 - c. Only referring patients without a family history of cancer
 - d. Referring all patients with breast cancer
 - e. Other
18. Have you made any of the following personal efforts to keep breast surgeons and their patients up to date with genetic testing guidelines? Check all that apply
- a. Attend tumor board meetings
 - b. Host grand round lectures
 - c. Prepare patient pamphlet on genetic testing information
 - d. Other

Questions about the GC's practices in ordering genetic testing

19. Prior to the ASBS guideline, how often did you offer genetic testing to patients who did not meet NCCN guidelines but did have a personal history of breast cancer?
- a. 0-25%
 - b. 26-50%

- c. 51-75%
 - d. 76-100%
20. Since the ASBS guideline was released, how often do you offer genetic testing to patients who do not meet NCCN guidelines but do have a personal history of breast cancer?
- a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
21. For all HBOC patients to whom you offer testing and do not meet NCCN guidelines, how often do they pursue testing?
- a. 0-25%
 - b. 26-50%
 - c. 51-75%
 - d. 76-100%
 - e. I do not offer testing to patients who do not meet NCCN guidelines
22. If you do not regularly offer testing to individuals who do not meet NCCN guidelines, what are the reasons you do not? Check all that apply
- a. Does not apply as I offer testing to all patients
 - b. Possible VUS
 - c. Large out of pocket cost for patients
 - d. Incidental findings for mutations in genes you did not expect to be involved
 - e. Possible findings in low penetrance genes
 - f. Patient anxiety
 - g. Testing not warranted based on family history
 - h. You do not agree with the ASBS guideline
 - i. Other
23. For patients who do not meet NCCN guidelines, what type of testing are you most likely to offer?
- a. BRCA1/2 only
 - b. Common breast cancer gene panel
 - c. Common breast cancer gene panel plus newer-risk/ preliminary data genes (adding genes without guidelines)
 - d. Comprehensive breast cancer panel
 - e. Pan-cancer susceptibility gene panel
 - f. I leave this choice up to the patient
 - g. I do not order testing on these patients
 - h. Other
24. In your experience, which of the following have been top reasons for patients to decline genetic testing when they do not meet NCCN guidelines? Choose top three.
- a. Out of pocket cost
 - a. Do not believe a mutation will be found
 - b. Believe genetic testing is not relevant to them
 - c. Believe genetic testing would not be beneficial

- d. Fear of VUS
- e. Anxiety over knowing mutation status
- f. Did not understand or see a benefit in knowing mutation status
- g. Genetic testing was not a priority at this time
- h. Concern for confidentiality of genetic information
- i. Concern over future insurance coverage (those not protected by GINA)
- j. Other

25. As compared to patient's who do meet NCCN guidelines, are those who do not meet guidelines, more or less likely to pursue genetic testing?
- a. More likely
 - b. About the same
 - c. Less likely

GC's personal opinions on the ASBS guideline

26. Do you personally agree with the new guideline and think there are benefits to genetically testing all individuals with a breast cancer diagnosis?
- a. Yes
 - b. No
 - c. Unsure
27. If you answered "No" to the previous question, why?
- a. I did no answer "No" to the previous question
 - b. Not enough data to support the guideline
 - c. Not a good use of resources
 - d. Patients that do not meet criteria will not get insurance coverage for testing
 - e. Other
28. Are there any thoughts or observations you would like to share about changes in the field of cancer genetic counseling since the release of ASBS guideline?

Appendix B

Student Research Survey Program

The Student Research Survey Program is designed to provide students currently in Genetic Counseling Training Programs access to NSGC members for research purposes at a discounted rate. This opportunity is designed to increase student survey results and to provide exposure to a variety of genetic counselors.

In order to submit a Student Research Survey, students should complete the Student Research Survey application below. The application will ask for information such as contact info, topic of survey, desired initial send and reminder dates and a brief description about your survey. The purpose of the brief description is to provide the audience with a summary of the survey. If you would like to include other information deemed as important, please include it in your survey itself as a cover page or as a precursor to the first question. Please note: In order for NSGC to distribute your survey, you must incorporate a consent statement (or notice of exemption from ethical review from an IRB) and contact information for the researcher(s) in the survey.

Both initial and reminder surveys will be sent out in a weekly digest on Wednesdays with survey content due by Friday of the prior week (i.e. the deadline to submit your survey is the Friday prior to the next Wednesday the digest is scheduled to be sent out.) Each digest will be organized according to the topic areas students will select from when filling out their submission form. The subject line of each digest will be: "Student Research Surveys & Reminders - <<current date>>" and will come from NSGC ensuring prominence in members' inboxes.

Beginning on January 1, 2020, NSGC will also post all surveys distributed via the weekly digest to a members only standing page for up to two months.

Surveys will be distributed as submitted, and drafts will not be circulated prior to distribution.

All student research surveys must go through this process and cannot be posted in the SIG communities or discussion forums.

**Please note: desired eblast send dates are not guaranteed and can be delayed during high volume request periods.*