Improving Provider Use of NCCN HBOC Genetic Counseling Referral Guidelines Through Provider Education and Awareness

by

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Genetic counseling and testing is important for families with a strong history of breast and ovarian cancer. Primary care providers play an important role in identifying women with a family history of breast cancer and making appropriate referrals to genetic counselors. Previous research has shown only 50% of women who should be referred for genetic counseling ever receive a referral from a primary care provider (PCP), which may be due to a lack of provider education and awareness.[1; 2] Our study evaluated the effect of a 1.5 hour continuing medical education (CME) course presented to PCPs and NPs through Allegheny Health Network in Pittsburgh, PA. After completion of the educational session, we tracked referrals from each provider and determined appropriateness by the National Comprehensive Cancer Network (NCCN) HBOC referral guidelines. Seven providers completed the educational session. A post-educational survey showed all providers responded positively to the material, with 100% stating they learned something new and 85.71% (6/7) stating they would make changes in their practice. Providers correctly answered that most cases of breast cancer are not hereditary (82% correct responses), males have an increased risk for a pathogenic variant with a previous breast cancer diagnosis (82% correct responses), and there is increased screening available to women with a family history (100% correct responses). However, most providers struggled with the heredity of ovarian cancer (41% correct) and the equal importance of maternal and paternal family history (41% correct). Referral numbers did not increase significantly post-education compared to pre-education (12 referrals for each), although there was an improvement in appropriateness of referrals (10/12 pre-education

compared to 12/12 post-education). In summary, this study showed provider education by online video serves to improve provider knowledge in some areas and may improve the appropriateness of patient referrals. This has implications for public health by improving provider knowledge and appropriateness of referrals placed by providers to genetic counselors. More research is needed to determine the long-term effects of a single educational presentation and areas of improvement to increase genetic counseling referrals.

Table of Contents

List of Tables

List of Figures

Preface

I am so grateful to have an amazing support system standing with me that has driven me to succeed and create a project whose foundation can be continued in research beyond my years at the University of Pittsburgh. I would like to extend a thank you to all those who have supported me during this time.

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1.0 Introduction

Genetic counseling and testing are important for families with a strong history of breast and ovarian cancer. An estimated 1 in 400 individuals carry a pathogenic variant (PV) in *BRCA1* or *BRCA2*.[3] Pathogenic variants in *BRCA1* and *BRCA2* significantly increase a woman's risk for developing breast and ovarian cancer and can change long-term surveillance and management options. Primary care providers play an important role in identifying women with a strong family history of breast cancer and making an appropriate referral to a genetic counselor. Many primary care providers see themselves as a "gatekeeper" to genetic services for their patients.[4] This role in the clinical practice setting is important in ensuring appropriate patients are referred to genetic counseling services. With an increasing demand for genetic counseling services and increased attention on the BRCA genes, primary care providers are now a critical aspect in the patient screening process.[5]

Although playing a crucial sentinel role in the genetic testing process, previous research has shown only 50% of women who should be referred for genetic counseling ever receive a referral from a primary care provider.[1; 2] These numbers are concerning, as there are millions of women in the United States who are carrying an unidentified pathogenic variant in a high-risk breast cancer gene. Multiple studies have been performed assessing primary care provider's awareness of referral guidelines, knowledge of referral criteria, and confidence in making a genetic counseling referral.[1; 2; 6] These studies determined that many providers were not aware of referral guidelines, that providers were not confident in determining which patients were most appropriate to refer to genetics, and that some providers were unclear about the process of ordering a genetic counseling referral for their patients.

Our study seeks to increase provider knowledge about hereditary breast and ovarian cancer and its relationship to a woman's family history of cancer. With an increase in provider knowledge and confidence in using a patient's family history to determine breast and ovarian cancer risk, we hope to see a significant increase in the number of referrals made to genetic counselors.

To increase primary care provider's knowledge about hereditary breast and ovarian cancer, we presented a 1.5 hour CME course to providers in the Allegheny Health Network system. This course included information on hereditary breast and ovarian cancer, warning signs and symptoms in a patient's family history that would warrant a genetic counseling referral, and information about the screening recommended for women deemed to be at an increased risk for breast cancer. Along with information on the family history, this study paid special attention to "absolute criteria" for referrals, such as a family member diagnosed with ovarian cancer or a male family member diagnosed with breast cancer. This study also paid special attention to stressing the importance of the paternal family history, as previous studies have shown only 6% of providers properly obtain paternal cancer history from their patients.[2] This portion of the study aimed to increase the knowledge surrounding genetics and genetic counseling, as well as build confidence in placing a referral to genetics based on a patient's family history. This study gave a post-education survey to providers to assess the knowledge learned from the course and to assess perceived utilization of this information in their practice. Prior studies have shown a significant increase in provider knowledge and perceived confidence after an educational intervention.[7]

After the provider education, this study tracked data on referrals from the attending providers to the AHN Genetic Counseling Program. This will allow us to determine if the knowledge and confidence learned in the informational presentation translates to an increase in the number of referrals from these providers, but also an increase in the appropriateness of the referrals

that are placed. Following these provider's long term can help determine the efficacy of the informational intervention on genetic counseling referrals.

In summation, this project aims to improve provider knowledge surrounding genetic counseling and genetic counseling referrals, as well as to build provider confidence in accurately assessing a family history and placing a referral to genetic counseling for patients. This study also attempted to assess the volume and appropriateness of referrals placed to genetic counseling from the providers who attended the informational sessions to evaluate the effectiveness of the educational material presented.

1.1 Specific Aims

1. Measure and improve provider knowledge surrounding criteria for genetic testing referrals using a CME-approved educational lecture and standardized survey
2. Measure and improve provider confidence in assessing a family history for breast cancer risk and placing a referral to a genetic counselor using a CME-approved educational lecture and standardized survey
3. Increase the number of referrals made to the genetic counseling program by providers attending an informational presentation
4. Improve the quality of referrals made to the genetic counseling program following an informational presentation to providers

2.0 Literature Review

2.1 What is BRCA and why is it significant?

Genetic testing for hereditary breast and ovarian cancer syndrome has been available for approximately 25 years.[8; 9] The breast cancer 1 (*BRCA1*) and breast cancer 2 (*BRCA2*) genes are two of the most commonly recognized genes associated with hereditary cancer syndromes in the genetics world. This fame is due in part to the publicity these genes received in May of 2013.[5] Angelina Jolie, a famous actress and director, published an opinion article in the New York Times discussing her family's history of breast cancer and her decision to undergo genetic testing and preventative breast surgery (mastectomy), which helps to decrease her chances of developing breast cancer in her lifetime.[10]

Angelina Jolie, along with up to 750,000 women in the United States, carries a harmful change (also called a pathogenic variant) in one of the BRCA genes.[3] Pathogenic variants (PVs) in either *BRCA1* or *BRCA2* confer a lifetime risk of breast cancer of up to 72% and a lifetime risk of ovarian cancer up to 45%.[3] For women without a PV in one of the BRCA genes, the risk for developing breast or ovarian cancer is 13% and 1.2%, respectively.[3]

Due to the significantly increased risk of breast and ovarian cancer in women with an identified PV in *BRCA1* or *BRCA2*, women are eligible for increased screening for breast and ovarian cancer, preventative breast or ovarian surgery, and preventative medications to help reduce their risk for developing cancer in their lifetime.[11; 12] These preventative measures can help reduce a woman's risk for developing cancer by over 90%.[13; 14]

2.1.1 BRCA+ and recommended screenings

The cancer risks and screening methods for patients with a PV in one of the BRCA genes are well established. Studies have quantified the rate of adherence to recommended breast and ovarian cancer screening guidelines in women with an identified PV.[15; 16] Genetic counseling and receipt of genetic testing results are not beneficial if patients are non-compliant with recommended surveillance strategies.[17] Buchanan et al. [15]determined the percentage of unaffected BRCA+ women who followed through with the recommended cancer screening regimen after disclosure of their test results in the time period recommended by national guidelines.[18] Of the 97 women surveyed, 66% of women were adherent to the recommended breast and ovarian cancer screening guidelines. Overall, more than half of women were adherent to both the breast cancer and ovarian cancer screening guidelines (average 71%).

Schwartz et al. [17] found a significant behavioral impact on women receiving bilateral prophylactic oophorectomy after receipt of positive *BRCA1* or *BRCA2* results. 27% of individuals who carry a PV in *BRCA1* or *BRCA2* had a prophylactic bilateral oophorectomy within the first year of receiving their genetic testing results.[17]

Long-term follow-up studies also demonstrate an 80% uptake rate in women receiving bilateral mastectomy and/or bilateral prophylactic oophorectomy within 5 years after receiving a positive *BRCA1* or *BRCA2* genetic testing result.[16] Researchers also found there was a higher use of breast MRI in BRCA PV carriers compared to non-carriers (46% compared to 11%, respectively).[16] In summation, women with an identified PV in *BRCA1* or *BRCA2* are generally good about following through with the recommended screenings, as outlined by national guidelines.

2.1.2 Risk communication with family members

Another important factor in determining the utility of testing for BRCA PVs in individuals at an increased risk is the impact these results can have on patient's family members. Elrick et al. [19] studied the psychosocial and clinical factors associated with communicating about an increased risk of breast cancer among close family members. Of the 920 women surveyed, only 13% carried a PV in one of the BRCA genes, although based on the age at which these women were diagnosed, their close relatives would still be at an increased risk for developing breast cancer compared to the general population, regardless of the woman's genetic testing results. Results showed women shared their genetic testing results with an average of 70% of their family members.[19]

This finding is consistent with other studies, reporting that on average, women share their positive BRCA results with 54-88% of relatives.[20] McGivern et al. [20] found that participants who carried a PV in one of the BRCA genes were significantly more likely to share their results with other family members compared to women without a PV identified ($\chi^2 = 10.21, p < 0.001$). These results did not hold true in women with only a family history of breast cancer and no identified PV. Independent of BRCA variant status, women were more likely to communicate about cancer risk with female family members than with male family members (proportion of 0.82 and 0.64, respectively).[20] Women who are aware of their increased risk for cancer, either based on their personal histories, family history, or BRCA variant status feel compelled to share their risk with other family members.[20-23]

Communication about breast cancer risk benefits not only the patient, but can have an impact on close family members risk management and genetic testing decisions as well.[23; 24] One study reported that up to 64% of family members who are informed of a genetic risk for cancer seek genetic counseling and testing for themselves.[25]

2.2 Who is at risk for BRCA pathogenic variants and how are they being identified?

2.2.1 Patients at risk

The risk for developing breast and ovarian cancer in a woman's lifetime is significantly increased if she carries a pathogenic variant in one of the BRCA genes. What some patients are also aware of is that they are still at an increased risk for breast cancer based on their family history alone. Several studies have quantified breast cancer risk based on a woman's self-reported family history. Colditz et al. [26] used data from the Nurse's Health Study to determine the relative risk for unaffected women whose mother or sister was diagnosed with breast cancer. Risk categories were further stratified into breast cancer diagnoses before age 50 and breast cancer diagnoses after age 50. There was a significantly increased risk across all categories of family history, with the most significant increase in relative risk in women whose mother was diagnosed with breast cancer before age 50 (RR=1.69; 95% CI 1.39-2.05).[26]

Brewer et al. [27]also used family history to determine a woman's risk for breast cancer based on her reported family history. This study analyzed the family structure (number of first- and second-degree relatives in relation to the number of expected breast cancer diagnoses based on population incidence) in addition to the number of reported cancer diagnoses, to give a more accurate risk estimate. Women who reported two or more relatives with breast cancer had a 2.5-fold increased risk to develop breast cancer themselves.[27] The relative risk of breast cancer in a woman with one relative diagnosed with breast cancer was comparable to the study by Colditz et al. [26](RR=1.77 vs 1.69, respectively). When stratified by age, women who reported a relative diagnosed with breast cancer prior to age 45 had a relative risk of 2.47 ($p<0.0001$).[27]

Braithwaite et al. [28]analyzed family history of cancer and determined breast cancer risk in women who were over the age of 65. Women over the age of 65 with no personal history of breast cancer are still at an increased risk for breast cancer if they have one or more first-degree relatives diagnosed with breast cancer (HR= 1.48; 95% CI 1.35-1.61).[28] Even women who are at older ages and thought to have out lived much of their risk for breast cancer are still at an increased risk based solely on their family history.

These studies demonstrate the need for patients to report their family cancer history to their medical providers, in order to get an accurate breast cancer risk estimate. Not only do patients need to report the cancer history, but more specific risk estimates can be given with detailed information such as age of cancer diagnosis in relatives and relation of relatives to the patient themselves.

2.2.2 Patient identification

Several studies have previously attempted to quantify the proportion of women in the general primary care population who would be considered as a high-risk patient based on family history of cancer alone.[29-31] Quillin et al. [30] used a survey combined with pedigree information to attempt to quantify the proportion of women in a primary care clinic who would be candidates for BRCA1 and BRCA2 testing based on their self-reported family history. Of the 490 women surveyed, 4.5% met the criteria for a BRCA genetic counseling referral.[30] This equates to 1 in 22 women in a primary care clinic setting meeting criteria for BRCA testing based on family history of breast and ovarian cancer.

These findings have been replicated by other researchers, including through a trial intervention program through Allegheny Health Network's Cancer Genetic Counseling Program in partner with Glimmer of Hope.[32; 33] Over 12 months of an interventional program attempting to

quantify the number of women who would warrant a genetic counseling referral in primary care clinics within the Allegheny Health Network in Pittsburgh, PA, unpublished data showed 40.9% of individuals reported at least some family history of breast and/ or ovarian cancer.[33] These studies demonstrate the need for identification of patients and collection of family history in the primary care setting, as there is a respective proportion of individuals who warrant further genetic evaluation for a hereditary cancer predisposition syndrome.

In previous studies, the primary method used for patient collection of family history for many years was a self-administered survey where patients reported their family history and discussed the family history with their providers.[29; 30] One study designed an interactive tool to help educate patients and providers on hereditary breast and ovarian cancer and determine which patients would need an additional genetic evaluation based on the family history provided.[31] Of the patients who accessed the tool, 67% inputted their entire family history, with 77% of patients asking family members about cancer history directly. In addition, 96% of women opted to learn about their risk for a BRCA PV. A majority (65%) of patients who assessed their risk printed those results out and discussed them with their providers.[31] This study demonstrates the interest patients have in their own family history and the importance of a complete family history in discussion of risk estimates.

In addition, to patient interest, almost 90% of providers reviewed and updated a patient's family history, with 83% of providers discussing hereditary breast and ovarian cancer (HBOC) and BRCA mutations with their patients.[31] Most providers were invested in their patient's family history of cancer and were willing to have a conversation with their patients about the need for an additional genetics evaluation.[31]

Providers who review a patient's family history can use a current and routinely updated set of guidelines established by the National Comprehensive Cancer Network (NCCN) to identify patients who are considered at an increased risk for a hereditary cancer syndrome and who can consider genetic testing based on personal risk factors and family history.[18] Per these guidelines, women who are diagnosed with breast cancer at a young age (defined as age 45 or younger), women who have a family history of breast, ovarian, prostate or pancreatic cancer meeting specific requirements (such as three or more individuals on the same side of the family with breast cancer), or women with a previously identified pathogenic variant in their family should all consider genetic testing.[18] These guidelines are the current standards by which providers should identify patients who could consider genetic testing based on their reported family history.

2.3 Role of primary care providers in patient identification and referrals

2.3.1 Role of PCPs

Primary care providers (PCPs) are an important part of many patients' medical care. They are often the front line of care for patients and can see a wide variety of indications in their clinics. Because of this, PCPs are expected to be knowledgeable in a wide variety of conditions which has lately included hereditary cancer syndromes. PCPs are often the front-line healthcare worker tasked with addressing patient's concerns about a variety of topics including their preventative healthcare and future screening and disease management.[34]

While a provider's knowledge surrounding hereditary cancer syndromes does not need to be extensive, they are responsible for assessing a patient's family history, determining a patient's

cancer risk, and they need to be equipped to make an appropriate referral to a genetic counselor if needed.[35; 36] PCPs also need to be able to make recommendations around screening for patients and inform management based on personal and family history.[18] Providers are also encouraged to review genetic testing results for patients who present with a personal and/or family history of a PV in hereditary cancer genes such as BRCA1 and BRCA2.[34]

PCPs are an essential part of many patients care and can help identify patients at an increased risk for cancer based on their family history. This identification at the source of patient care can help increase the number of referrals to the genetic counseling department, and can inform patients of their future risks for cancer.[35; 37]

2.3.2 PCPs knowledge of BRCA and genetics

Previous studies have attempted to quantify provider's knowledge surrounding appropriate referral practices for genetic counseling and testing. Burke et al. [2] conducted a study using standardized patients in a primary care facility to measure the accuracy and completeness of provider's family history taking skills, provider's knowledge about hereditary breast and ovarian cancer syndromes, provider's breast cancer risk assessment for their patient, and the provider's willingness to place a referral to genetic counseling if necessary. Analysis of family history taking skills among providers found that about half of providers (61%) asked extensively about maternal relatives with cancer. However only 32% of providers asked about age of onset of cancer.[2] Less than half of providers asked about paternal history of breast cancer, and only 18% asked about paternal family history of ovarian cancer.[2] Furthermore, only 6% of all providers elicited a full paternal family history of breast and ovarian cancer. Other publications have studied the accuracy of providers in taking a complete family history of cancer and have found similar results.[38; 39]

When analyzing a patient's risk of breast cancer based on the family history, many providers are presenting risk estimates for patients based on incomplete information.[2; 40] In general, providers are good at determining that patients with a family history of breast cancer are at an increased risk compared to patients without a family history of cancer.[40] Providers have less success determining the importance of family history of breast cancer and ovarian cancer, and struggle to identify a patient's cancer risk when the family history of cancer is present only on the paternal side.[2; 40] Providers also tend to overestimate a woman's breast cancer risk in the setting of a family history of breast cancer and tend to underestimate a woman's risk in the setting of a family history of ovarian cancer.[41]

In addition to provider's incorrect assumptions and risk analysis based on family history alone, when surveyed, providers overestimated their ability and confidence in assessing a family history and estimating a patient's risk for breast and ovarian cancer.[41] These findings are similar across providers who have had formal education on cancer risk assessment and those who have had no previous formal training.[40]

2.3.3 PCPs referral practices

In addition to lack of appropriate analysis of family history and risk analysis, providers often struggle with placing appropriate referrals to genetic counselors. Burke et al. [2] analyzed the referral practices among providers after their patient intakes and cancer risk calculations for a set of standardized patients. Only 21% of providers recognized the need for genetic counseling with a significant maternal family history, and only 3% of providers referred patients who presented with a significant paternal family history of breast and ovarian cancer to a genetic counselor.[2]

Lack of patient referrals may be due in part to personal beliefs on the benefit of genetic testing held by PCPs. Hamilton et al. [34] conducted a meta-analysis on PCP's knowledge, attitudes and communication practices surrounding genetic counseling and testing for hereditary breast and ovarian cancer. Overall, PCP's attitudes regarding the utility of testing were favorable, with some providers noting concern for distressing legal, social and psychological effects that could come from completing genetic testing. Some providers also expressed their personal doubts about the utility of genetic testing to their patients. However, most providers were in agreement that genetic testing was helpful in determining patient's future risk, and could aid in determining future cancer screening for patients.[34]

PCPs are not perfect when it comes to the collection of family history in a clinical setting. Further, many providers are not well informed regarding the effect a paternal family history of breast and ovarian cancer can have on a woman's future cancer risk. Additionally, some providers are still skeptical of genetic counseling and testing, and are unsure of the benefits genetic testing can give patients and their families. All of these factors may contribute to the low genetic counseling referral rate in certain practices.

Although there is a significant proportion of patients who are identified as being high-risk, many of them will not be appropriately referred to a genetic counselor.[1; 4; 42] One referral-based study surveyed 3200 providers on their referral rates for BRCA1/2 testing.[43] Among the 1878 providers who responded, 41% reported adherence to referral recommendations in a high-risk patient setting (i.e. those patients who have a significant family history of breast and ovarian cancer and who would be appropriate to refer to genetic counseling). Conversely, 30% of providers reported they would refer to genetic counseling a patient who weas at an average risk of breast cancer based on their family history.[43]

Koil et al. [4] conducted a survey of providers in the Ohio region to determine referral practices by location. Of the 214 respondents, "51% reported having ever referred for an indication of hereditary breast cancer".

This study also classified disparities between clinic location, including urban/ suburban practice and rural practices. Providers practicing in an urban/ suburban location were more likely to refer patients for genetic counseling than providers practicing in a rural setting. Furthermore, many of the rural provider referrals to genetic counseling were placed due to patient interest, rather than a reported personal or family history of cancer.[4]

There needs to be an increase in education for providers not only on which patients should be referred based on personal and family history of cancer, but also on how to make a patient referral to genetic counselors in their area. Special attention needs to be paid to educating rural providers and providing genetic counselor contacts who work with patients in rural settings.

2.4 What does this all mean for the cancer genetics world?

Moving forward, more research needs to be done on changing the system starting from the PCP clinics. Much of the current research is on increasing patient awareness of their increased breast cancer risk and ensuring patients are compliant with their cancer screenings.[16; 29; 30] In order to increase referrals and ensure patients are being appropriately identified, some of the responsibility falls on the providers. PCPs are a gatekeeper for patients and are a crucial part of many patient's care. Targeting the knowledge and confidence levels of PCPs surrounding genetics and hereditary breast and ovarian cancer would help increase patient identification and referrals to genetic counselors.

Previous studies have demonstrated the efficacy of increasing PCPs knowledge through education on genetics and education on proper referral practices.Wilkes et al. [7] administered interactive web-based continuing education to PCPs in the California area. Providers who were in the intervention group (i.e. those who received the interactive education) had a greater increase in knowledge about genetics and genetic counseling compared to the providers in the control group.[7] Furthermore, providers in the intervention group reported an increase in confidence in using genetic counseling referral guidelines to assess risk for their patients.[7] There is limited research into intervention services targeting PCPs, although one meta-analysis showed half of studies reported an increase in provider knowledge following an educational intervention.[44] Further research needs to be conducted to determine the most effective method of increasing provider referrals to genetic counseling and ensuring the referrals made are for appropriate patients.

Our study aims to improve primary care provider education through a web-based informational video. This video highlights the NCCN HBOC referral guidelines and includes a tutorial on how to place an electronic referral to the Cancer Genetics Program. With many different approaches to increasing provider knowledge surrounding hereditary breast and ovarian cancer a short, interactive-based educational program is the one of the most effective.[7] These programs help to expand PCP's expertise in genetics and increase the volume of referrals while maintaining the appropriateness of the referrals placed. Our study will use this educational framework to demonstrate an improvement in PCP's confidence with using the NCCN HBOC referral guidelines and establish an increase in the number and appropriateness of patient referrals.

Although genetics can be confusing and overwhelming for PCPs, consistent, short and easily-digestible information sessions can help improve patient care and inspire confidence in providers to make sense of complex genetics topics. This study aims to improve upon previous

studies in the hopes of determining the most beneficial method of provider education in regards to hereditary breast and ovarian cancer. This advancement in education can serve to better patient care and increase awareness for genetics and genetic counseling services in a variety of clinical settings.

3.0 Manuscript

3.1 Background

Genetic testing for hereditary breast and ovarian cancer has been available for the last 25 years.[8; 9] The breast cancer 1 (*BRCA1*) and breast cancer 2 (*BRCA2*) genes are two of the most commonly recognized genes associated with a hereditary cancer syndrome. The incidence of *BRCA1* and *BRCA2* pathogenic variants (PVs) in the United Stated is estimated at 1 in 400 individuals.[3] PVs in *BRCA1* and *BRCA2* significantly increase a woman's risk for developing breast and ovarian cancer and can change long-term surveillance and management options.[11; 13; 45] Women at an increased risk are eligible for additional screening for breast and ovarian cancer, preventative breast and ovarian surgery, and preventative medications to help reduce their risk for developing cancer in their lifetime.[11; 12; 18] These preventative measures can help reduce a woman's risk for developing cancer by over 90%.[13; 14]

Genetic testing results are often not impacting only the patients themselves. Due to shared genetics among family members, an identified PV in a high-risk hereditary breast and ovarian cancer (HBOC) gene can impact other blood relatives. One study found that on average, women disclose their positive BRCA results with up to 88% of relatives.[20] This information about an increased risk for cancer can be important for an individual's relatives in determining eligibility for additional cancer screenings.

The risk for developing breast and ovarian cancer in a woman's lifetime is significantly increased if she carries a pathogenic variant in one of the BRCA genes. What some patients are unaware of is that they may be at an increased risk for cancer based on their family history alone.

Several studies have quantified breast cancer risk based on a woman's self-reported family history.[26; 28] Due to this increased cancer risk without an identified PV, there is a need for patients to report their family history of cancer to their providers, especially those providers at the front line of patient care.

Primary care providers (PCPs) are an important part of many patients' medical care. PCPs are often the front-line healthcare worker tasked with addressing patient's concerns about a variety of topics including preventative healthcare and future screening and disease management.[34] Many primary care providers see themselves as a "gatekeeper" to genetic services for their patients.[4] This role in the clinical practice setting is important in ensuring appropriate patients are referred to genetic counseling services. With an increasing demand for genetic counseling services and increased attention on the BRCA genes, primary care providers are now a critical component in the patient screening process.[5]

Although PCPs play a crucial sentinel role in the genetic testing process, previous research has shown only 50% of women who should be referred for genetic counseling ever receive a referral from a primary care provider.[1; 2] These numbers are concerning, as there are millions of women in the United States who are carrying an unidentified pathogenic variant in a high-risk breast cancer gene. Multiple studies have been performed assessing primary care provider's awareness of referral guidelines and confidence in making a genetic counseling referral.[1; 2; 6] Providers have limited success determining the importance of family history of breast and ovarian cancer, and struggle to identify a patient's cancer risk when the family history of cancer is present only on the paternal side.[2; 40] Providers also tend to overestimate a woman's breast cancer risk in the setting of a family history of breast cancer and tend to underestimate a woman's risk in the setting of a family history of ovarian cancer.[41] In addition to lack of appropriate analysis of family

history and risk communication, providers often struggle with placing appropriate referrals to genetic counselors. One study found that only 21% of providers recognized the need for genetic counseling with a significant maternal family history, and only 3% of providers referred patients to a genetic counselor who presented with a significant paternal family history of breast and ovarian cancer.[2]

Moving forward, more research needs to be done on changing the system starting from the front-line providers. Much of the current research is on increasing patient's awareness of their family history and ensuring patients are compliant with recommended cancer screenings.[16; 29; 30] The aim of this study was to improve provider education of genetics and genetic counseling services using an educational session focused on common misconceptions surrounding hereditary breast and ovarian cancer including the equal importance of maternal and paternal family history, hereditary cancer risks among men and women, and care and management following positive genetic testing results.[2; 38; 42] This study also aimed to assess the improvement of patient referrals after provider education to demonstrate retention and application of the material presented.

3.2 Methods

3.2.1 Setting and Participants

This study took place in Pittsburgh, Pennsylvania at Allegheny Health Network (AHN). Primary care providers (PCPs) were eligible for study participation if they were an MD, DO, NP, English-speaking, and a currently practicing provider through Allegheny Health Network (AHN). Participating providers were required to have access to the AHN internal Continuing Medical

Education (CME) site. PCPs were sent an informational email advertising the educational session at the beginning of the study (Supplemental Figure 1). The educational session was also advertised as part of multiple presentations and AHN monthly departmental meetings throughout the study duration. In total, 7 providers were recruited between September 2021 and December 2021. Participants were offered 1.5 CME credits after the completion of the educational session. Only providers who completed the educational session were included as part of this study.

3.2.2 Study Design and Procedure Overview

Study procedures were IRB approved by Allegheny Health Network and the University of Pittsburgh. Providers were offered an educational video and post-education survey as part of this study. The educational video consisted of a general genetics overview, information on hereditary breast and ovarian cancer (following the National Comprehensive Cancer Network Hereditary Breast and Ovarian Cancer guidelines), examples of patient pedigrees, and a tutorial on placing an electronic referral to the AHN Cancer Genetics Program (Supplemental Document 1). These topics were chosen based on previous research and special consideration was given to eliciting family history from both sides of the family, providing concrete examples of a family's cancer history and tools for identifying appropriate patients to refer for genetic counseling.[44] Learning objectives for the study were for providers to be able to identify individuals at risk for a hereditary cancer syndrome and for providers to be able to provide medical management recommendations for a BRCA gene mutation carrier. The post-education survey consisted of 6 true/false knowledge-based questions on the material presented, 6 yes/no questions on the stated learning objectives, 7 Likert scale questions on confidence and knowledge surrounding patient referrals, and 3 free response questions on proposed changes to the presentation. Providers were able to attempt the post-

education survey as many times as they would like. A score of 100% on the post-education survey was not necessary in receiving CME credit for this course. Providers who began the educational video did not have a designated time restriction and were enrolled in the study upon completion of the post-education survey.

After completing the post-education survey, the number of referrals and appropriateness of referrals was tracked for each provider. Patient referrals were tracked from October 2021 through January 2022. Patient referrals were tracked for varying lengths of time dependent on the month each provider completed the educational survey. For example, if a provider completed the educational survey in October 2021, their patient referrals were tracked from November 2021 through January 2022 (3 months total). Referral numbers were averaged over the number of months each provider's referrals were tracked for and compared pre-educational session to post-educational session.

Patient referral numbers were tracked using Allegheny Health Network EPIC electronic medical record system (EMR). Appropriateness of referrals was determined on a yes/no basis and followed the National Comprehensive Cancer Network Genetic/Familial High-Risk Assessment: Breast, Ovarian and Pancreatic Cancer guidelines v1.2022. Only information included in the referring provider's visit summary note was used to determine personal and/or family history of cancer. Referral data was de-identified and stored on an encrypted server housed at Allegheny General Hospital in Pittsburgh, Pennsylvania. Only de-identified data was used for statistical analysis.

3.2.3 Statistical Analysis

Data was analyzed using Stata SE (v16). Descriptive statistics were used to describe characteristics of the sample of providers and the post-education survey. A two-sample z-test was used to test for statistical significance. Statistical significance was set at $p<0.05$.

3.3 Results

There were seven providers who completed the educational video and post-educational survey. Providers were primarily female (85.71%), and registered nurse practitioners (71.43%). The average length of time to complete the educational video and post-education survey was 39.95 hours, with 42.8% (3 of 7) providers completing the educational session in less than one day. Length of time was determined from the time a provider accessed the educational session to the time a provider completed the post-education survey. The system used to determine length of time cannot determine how many of those hours were "active" hours when providers were interacting with the video or taking the educational survey. Providers were able to access the video as many times as they wished and were not required to complete the educational session in one sitting. Due to this, providers may have accessed the educational session and not been able to complete the video and survey in one sitting, which extended their length of time to complete the video and survey.

3.3.1 Provider recruitment

There were 7 providers who completed the educational session. This educational session was free for providers and those who completed the educational session and survey received CME credit. There are around 400 primary care providers at Allegheny Health Network. Less than 2% of eligible providers completed the educational session. There was difficulty in getting PCPs to complete the educational session, with multiple attempts made to increase participation including sending blast e-mails to offices, presenting at multi-disciplinary meetings and highlighting the presentation on the internal AHN CME site. Even with these attempts made, the rate of completion of this educational session by available providers was very low. This may also speak to the difficulty in getting providers to participate in optional educational sessions found in other studies [46]. Follow-up on why providers completed or why providers did not complete this educational session was not performed, but may be an area for further research.

3.3.2 Post-educational survey questions

Participants made a total of 17 attempts on the post-educational survey. It is unknown how many attempts each provider completed. Providers were allowed to attempt the survey as many times as they wished. Providers were not required to receive a perfect score (100%) on the post-education knowledge-based survey to receive CME credit, although this was not explicitly stated in the survey. Previous CME-approved lectures that contained a knowledge-based survey required providers to receive a perfect knowledge score to receive their CME credit. Due to this possible misconception of needing to receive a perfect score on the knowledge-based survey to receive CME credit, it is likely all providers took the post-education survey. The 6 true/false survey

questions included in the post-educational survey were used with permission from Cohn et al. [40], whose team conducted a similar study on provider's risk assessment knowledge regarding BRCA genetic testing.[40] While the same survey questions were used, the educational sessions presented to physicians was different between this study and the study by Cohn et al. [40].

A majority (82%, n=14) of provider attempts correctly identified that most instances of breast cancer were not hereditary, but only 41% (n=7) correctly identified that most instances of ovarian cancer are not hereditary. Most provider attempts (94%, n=16) correctly determined that there are different management recommendations for women who are at an increased risk for breast cancer due to an identified pathogenic variant. Additionally, 82% (n=14) of provider attempts in our study correctly identified that a male diagnosed with breast cancer is still at an increased risk for carrying a hereditary cancer pathogenic variant. This study found limited knowledge of the equal importance of maternal and paternal family history in cancer risk assessment (41%, n=7) (Table 1).

Table 1 True/ False Survey Statistics

Question	Correct Answer	% correct (total attempts)	Std. Dev
1.Most instances of ovarian cancer are hereditary	false	41% (7/17)	49.20%
2. Most instances of breast cancer are hereditary	False	82% (14/17)	38.10%
3. For cancer syndromes affecting mostly women, maternal history is more important than paternal history	False	41% (7/17)	49.20%
4. A male with a diagnosis of breast cancer is at risk for carrying a BRCA mutation even in the absence of family history	True	82% (14/17)	38.10%

5. If a person is found to have a hereditary cancer mutation, nothing can be done to improve screening for cancer	False	94% (16/17)	23.50%
6. A woman with a hereditary risk for breast cancer would have the same recommendations for breast screening as someone without a hereditary risk	False	100.00% (17/17)	0.00%

In addition to the true/false quiz questions, providers responded to five Likert scale questions on the effectiveness of the educational session and their perceived confidence with using the guidelines presented (Table 2). All of the participants were "likely to recommend" or "definitely would recommend" this educational session to their peers. A majority (85.71%, 6/7) of providers would make changes in their practice based on this session, with open responses including "[I will] be more aware of family history in my patients", "[I will] refer more patients with genetic risk", and "[I will] better educate patients on genetic counseling services" (Supplemental Figures 2-110Five out of seven providers (71.43%) stated they would use the NCCN HBOC referral guidelines frequently in their practice, and similarly, five out of seven providers (71.43%) also stated they feel confident in using the referral guidelines after the educational session.

Table 2 Likert Scale Survey Responses

Likert Scale Questions	Strongly disagree	Neither agree nor disagree	Agree/ Strongly Agree
1. I would use the NCCN referral guidelines frequently	Infrequently/ Very infrequently: 0/7 (0.00%)	Neither infrequently or frequently: 2/7 (28.56%)	Frequently/ Very frequently: 5/7 (71.44%)
2. I feel confident in using the NCCN referral guidelines	1/7 (14.28%)	1/7 (14.28%)	5/7 (71.44%)

3. The material was presented in a way that was easily understood	0/7 (0.00%)	0/7 (0.00%)	7/7 (100.00%)
4. I have learned new information from this presentation	0/7 (0.00%)	0/7 (0.00%)	7/7 (100.00%)
5. I have learned information I intend to apply to my practice	0/7 (0.00%)	1/7 (14.28%)	6/7 (85.72%)

All of the providers stated they learned new information from this session, and six of seven providers (85.71%) stated they learned something they intend to apply to their practice. When asked about positive aspects of the session, responses included "The material was presented in a way that made complicated information much easier to understand. I really liked the frequent use of graphs/tables", "[There is a] clear understanding and examples of when to refer [to genetic counselors], "knowing the percentage of cancers that may be hereditary was enlightening". There were no comments made about future improvements to the educational presentation.

3.3.3 Post-Education referral numbers

Following the educational session, provider referral numbers and referral appropriateness were collected. Referral appropriateness was determined only by the provider-entered personal and family history at the time of their visit and used the most updated version of the NCCN HBOC guidelines at the time of referral.[18] One provider completed the educational session in September 2021, one in October 2021, one in November 2021 and four completed the educational session in December 2021. Post-educational referral numbers were tracked for a total of thirteen months across the seven providers (Table 3). There was a total of twelve new referrals that came in over the total of 13 months of referral tracking for providers.

Table 3 Pre- and post-educational referral numbers

Provider	Average # referrals/month (pre-education)	Average # referrals/month (post-education)	# months tracked
1	2	2	4
2	0.66	1.33	3
3	0.00	0.00	2
4	0.00	0.00	1
5	0.00	0.00	1
6	2	0.00	1
7	0.00	0.00	1
Total	**4.66**	**3.33**	**13**

Referrals that met guidelines was determined by the information on personal and family history collected by the provider at the time of the visit using the most updated version of the NCCN HBOC criteria.[18] Of the twelve patients referred, 100% (12/12) met NCCN HBOC criteria post-education compared to ten of twelve patients (83.34%) who met criteria pre-education.[18] The data was trending towards significance ($p=0.0585$) for improved proportion of referrals that met guidelines post-education when compared to pre-education. There were two patients who did not meet referral criteria who were referred prior to the educational session. One patient had a previous history of endometrial hyperplasia with atypia diagnosed at 35 and no reported family history of cancer, and the other patient reported a mother with breast cancer diagnosed at 50 and a father with colon cancer (unsure of age of diagnosis) and no other reported personal or family history of cancer. This patient ultimately spoke with a genetic counselor who did not offer genetic testing based on her family history of cancer.

3.4 Discussion

This study attempted to improve the referral rate and appropriateness of patient referrals to the cancer genetic counseling program at Allegheny Health Network using an educational video and post-education survey. There has been a desperate need for improved genetics education among primary care providers (PCPs).[2; 34; 42] Several methods of improving provider education have been studied over the years including written material, educational videos, and simulated patients.[44; 47] Our study used a brief educational video to provide a background of genetics, examples of family histories and clear guidelines for making a referral to the genetic counseling department to help increase provider's confidence and knowledge surrounding hereditary breast and ovarian cancer. Several studies have been published on provider acceptance of tailored educational sessions.[48; 49] Post-educational survey and free-response questions indicated a high provider satisfaction with the online educational sessions, which has been previously shown in other studies.[49] In addition to the high satisfaction of the educational sessions, providers reported an increase in confidence in speaking with their patients and using the provided referral guidelines. Similar to this study, other studies have demonstrated a lack of prior genetics knowledge, with an increase in knowledge and confidence in discussing hereditary breast and ovarian cancer with patients following an educational intervention.[48; 50]

Educational videos have been thought to be the least time-consuming for providers, although most have only moderate effectiveness in post-education investigation.[44; 47; 51] PCPs are often busy providers who see a wide variety of indications in their offices and have limited time to participate in optional educational opportunities.[44] While this educational intervention was rated high in satisfaction and perceived use of information, similar to previous studies there was only

moderate effects seen post-education in terms of increased knowledge and increased referral numbers.[37]

In this study, most providers correctly answered the post-education question relating to the percentage of breast cancer due to hereditary causes. Providers also correctly identified the need for additional breast cancer screenings in women who are found to have a high-risk breast cancer pathogenic variant. This study showed good provider identification of males with a diagnosis of breast cancer having an increased chance of carrying a pathogenic variant. Using previous studies, it was determined that males with or without a cancer history were harder for primary care providers to identify as meeting NCCN HBOC testing criteria.[39; 41; 52] In addition, many studies often don't focus on males in the primary care space due to the lack of relevant breast cancer history in male patients themselves.[34; 42; 43] Due to this difficulty in the identification of males who should be referred for genetic counseling, the educational session presented in this study highlighted cancers in males and the importance of personal history.

Despite the lack of a significant increase in knowledge, there was high satisfaction and high retention of information among providers who completed the educational sessions. One participating provider has been in close contact with several of our genetic counselors to inquire on referrals for patients of hers that do not meet classic NCCN HBOC guidelines based on their reported family history of cancer. There have also been several messages from nursing staff on the appropriateness of additional provider's referrals. Providers are eager to participate in increased genetics education, but may need continued help in narrowing down which patients are truly considered high risk based on family history alone.

This study did not show an increase in the number of referrals post-educational session compared to the number of referrals pre-educational session. The average number of referrals per

month increased post-education compared to pre-education for one provider. This provider doubled the number of referrals placed post-education. All other providers either maintained their average referral numbers or decreased their average referrals pre-education compared to post-education. This may be due in part to lack of long-term follow up of referral numbers per month.

In addition to the lack of long-term follow-up, the number of referrals may not have changed significantly due to the time it takes providers to implement new procedures into their clinics. Many primary care provider offices are busy places, with a number of support staff functioning together. One provider who attempts to change a procedure in the clinic must inform other providers as well as the supporting staff of the changes before any significant procedural changes are seen. Referrals from participating providers were only tracked for a month following the educational session for four providers. This may not have been enough time to elicit a significant change in referral numbers in those provider's practices. Long-term follow-up of these providers may show an increase in referral numbers due to changes in a clinic's referral procedures.

Our study showed mild improvement in the appropriateness of genetic counseling referrals placed post-education compared to pre-education. Although there was not a significant improvement in the appropriateness of referrals as measured by this study, long-term follow-up may improve the referral appropriateness across providers who completed the educational sessions.[53] Few studies have followed providers for extended periods of time post-education, although the common trend of any provider education is the lack of sustained change in many practices.[47] Increasing provider education and monitoring changes over an extended period of time can help identify specific challenges facing individual clinic and providers from making sweeping changes in their practice.

3.4.1 Study Limitations

One of the important limitations to this study was the duration of post-education referral collection. Due to the limited length of time each provider's post-education referrals were tracked, there was not a significant change in referral practices noted across the seven providers. Although there was limited follow-up time reported in this study, additional studies have reported moderate to minimal changes following educational videos.[44] The most significant referral changes are seen in studies that consisted of extensive follow-up and additional tailored educational sessions spanning multiple months.[54] Additional follow-up with providers can be considered to determine how length of time from an educational session impacts referral rates and appropriateness of referrals. This follow-up can include supplementary educational sessions emphasizing the areas where providers previously struggled, such as the importance of maternal and paternal family history. Follow-up can also include information on changes made to the referral criteria to ensure all providers are aware of any guideline updates.

Another limitation of this study was the limited number of participating providers. Seven providers completed the educational session. There were limited responses to the post-education survey questions and comparison statistics were minimal due to lack of data. Previous studies have commented on the ineffectiveness of provider education, with an average survey response rate of 40-50%.[46] An additional study of similar design reported only half of providers enrolled completed the educational session offered.[49] Providers are often busy people and even with no associated financial cost, it can be difficult to get a large number of providers to complete an educational session. Aside from the length of time that may have been an issue, future educational sessions can be broken down into smaller, more manageable sections. Previous research has shown the optimal length of a recorded lecture video is between 6 and 15 minutes.[55; 56] Creating short,

informative videos on the same information presented in this study would help increase provider engagement and provide smaller sections of information.

Yet another limitation was the poor evaluation system withing the CME site the educational video was posted to. When determining the number of hours a provider spent on the educational session, the system only evaluated the time when the educational session was first started and when the educational session and following survey were completed. There was not a way to determine how many active hours providers spent on the educational session, which may be considerably less time than the total number of hours that was reported on. With a better system to determine active provider hours, there can be improvements made to the length of the videos and more insight can be gathered on provider's schedules. This can be useful information in determining the most appropriate length of time for educational videos to be presented to providers which may help increase provider engagement and participation.

Analysis was completed on a small number of patient referrals. Limited statistics can be performed with 12 patient referrals, making any generalizable conclusions about improvements in referrals impossible. If patient referrals were to be tracked for longer, or more provider's referral data was tracked, there could be more significant statistical findings. Allegheny Health Network is a small referral institution, with less than 500 referrals per month across all providers and all cancer types. When tracking referrals from specific providers for specific cancer types, there was limited statistical power that could be used in this analysis. Long-term follow-up or follow-up with more provider's data can be useful in determining any trends in referral improvement stemming from this educational session.

This was a small-scale study completed at one institution in Pittsburgh, Pennsylvania. Larger, more diverse studies would need to be completed to make any statements about result

generalization. This data is promising in the potential for increased referral numbers and increased referral appropriateness after a provider education session, although these results would need to be replicated at other institutions. Several studies have been published with similar study designs, although results across these studies have been comparable to this study.[49-51]

3.5 Conclusions

This study was conducted to highlight the importance of integrating genetics into the primary care space, which can improve access and awareness of genetic counseling services across a health network. The foundation to successfully completed genetic integration comes from proper and sustained education of primary care providers (PCPs). Discussions and education surrounding genetics is a complicated and often nuanced subject to undertake with patients, but with proper education and appropriate resources available to providers, there can be improvements in genetics knowledge and awareness. This study has shown that a single, brief educational session to a small group of primary care providers will not create widespread change, although this study was promising in that it provided insight into improvements that can be made to create a successful integration of genetics into the primary care space.

Sustained, routine and informative education with available genetic counseling support and additional provider resources are the key to creating genetics savvy providers. This study was focused on hereditary breast and ovarian cancer, although there is a myriad of additional hereditary cancer syndromes that are less well-known but still crucial to discuss with patients and their families.

This study aimed at increasing provider's knowledge and confidence using an educational CME course to improve the number and appropriateness of genetic counseling referrals for hereditary breast and ovarian cancer. Primary care providers are at the forefront of the healthcare field and are often the first line of care for many patients. These providers play an ever-increasing important role in monitoring their patient's health and ensuring they are connected with the appropriate specialty services. This study serves to demonstrate the improved referrals that stem from a brief educational session, but also highlights the expanding field of genetics and the need for further and consistent education for providers in the primary care space.

3.5.1 Further Research

Further follow-up in this field should be focused on creating additional educational sessions to be presented at routine intervals to providers. Increased access and reminders of genetic services can benefit in recruiting more providers to participate in additional educational sessions. The video created for this study is still being offered to providers at Allegheny Health Network, and can serve as a valuable resource for providers moving forward. With additional educational opportunities offered, more in-depth information and updates to current NCCN guidelines can be relayed to providers. The landscape of genetics is constantly changing and it can be difficult for providers to maintain a current knowledge of referral guidelines.[37; 40] Routine education can provide a space to pass on these updates in a scheduled manner. This can also ensure providers are presented with information on genetics at routine intervals to serve as a reminder for providers to ask patients about their family history.

Additional investigation can include studying the long-term impact on referrals by providers who completed an educational session. Long-term referral data would help identify areas

of educational need and help classify long-term retention of information by providers. If there are specific areas that providers are struggling in, additional education or support services can be offered to ensure appropriate patient referrals are being made. Houwink et al. [49]conducted a long-term study on the effects of continuing medical education and demonstrated that 6-months post-education, knowledge on genetics topics were not significantly increased over pre-test knowledge.[49]

4.0 Research Significance to Genetic Counseling and Public Health

The field of cancer genetics is a complex and detailed area with many nuances that are not always apparent to providers outside the genetics space. As front-line providers, PCPs need to be able to "investigate, diagnose and address health problems and hazards affecting the population".[57] In order to effectively identify health hazards, providers need to be appropriately trained to identify increased cancer risks in an individual based on their family history. Previous research has demonstrated a lack of genetics knowledge by PCPs.[34; 40] Improvements in PCPs knowledge of genetics can help providers feel more confident in investigating a patient's family history cancer, which can help them address any concerns for a hereditary cancer predisposition. This study focused on the lack of genetics knowledge by providing PCPs with an informational video highlighting some of the common misconceptions about hereditary breast and ovarian cancer and included examples of family histories to help providers apply the information to real-life situations.

In addition to being able to accurately identify patients with a potential hereditary cancer predisposition, providers need to have "effective communication and education of patient's health and the factors that influence it".[57] If providers are unable to relay important health information to their patients, there may be a lack of identification of patients who would benefit from genetic testing or additional cancer screenings. Many primary care providers (PCPs) have noted they have a limited knowledge about cancer genetics and they are unsure of how to talk to their patients about their family history.[41] Supplying providers with tools they can use to determine relevant cancer history in their patients is important in ensuring effective communication about a patient's cancer risk.[41] This study used real-life examples of family histories and walked providers through an assessment of the relevant information. Specific, targeted examples such as this can help providers

communicate with their patients and evaluate their family history for signs of a hereditary cancer predisposition.

Aside from investigating, diagnosing and communicating effectively with their patients, PCPs need to be able to effectively address health problems for their patients.[57] After PCPs have gained the knowledge and skills to communicate with their patients regarding their cancer risk, a provider needs to be able to appropriately address the concern for a hereditary cancer predisposition. Oftentimes this involves making a referral to the genetic counseling team.[43] This study included information on how to make an appropriate referral to a genetic counselor and determined the proportion of appropriate referrals after the educational video. Thorough patient education by providers will not significantly improve a patient's health if there is a lack of referrals. Appropriate referral action by providers will help patients address their health problems, which may improve health outcomes long-term.[15] Investigation, communication and action by providers all work together to ensure patients are receiving the best possible care to manage their health accordingly.

While PCPs play a crucial role in the identification of patients at an increased risk for cancer, there needs to be "an effective system that gives equitable access to specialty services that an individual needs to better their health".[57] This system involves a working relationship between PCPs and genetic counselors to ensure patients are receiving the best care. Genetic counselors need to partner with PCPs in their area to ensure proper and continuing education is provided, along with communicating to ensure appropriate referrals are made.[48] There cannot be an effective or equitable system in place without the cooperation of both entities. Genetic counselors are often the providers who are up to date with the rapidly-changing genetics landscape and can provide the most updated information to their PCPs. Working together, genetic counselors can help build a

successful education plan for the providers working with them to establish a foundation of genetics knowledge among all providers.[58] Even without physical genetic counselors on-site with providers, some studies have been conducted using virtual technology to act in place of a genetic counselor.[53; 59] These studies have been met with more success than traditional education methods.[44; 60] Genetics providers need to be open to consistently working with PCPs to ensure effective and complete education to provide equitable access and intervention to all patients.

In summation, primary care providers have a duty to inform patients of their risks for disease and to offer solutions for their patients who are at an increased risk. Educating PCPs so they can properly discuss hereditary cancer risks with their patients is a growing area of research to determine the most effective and sustainable education plan. With improved provider education, fair and equitable access to these specialty genetics services can improve healthcare and create a healthier patient population.

5.0 Appendix Supplemental Figures

Please join Dr. Janette Gomez and Kyla Morphy, genetic counselor as they present a talk on the referral guidelines for individuals with a family history of breast and ovarian cancer.

Janette Gomez, MD
Breast Surgeon

Kyla Morphy, MS, LCGC
Genetic Counselor

Title: HEREDITARY CANCERS : FAMILY HISTORY ASSESSMENT, GUIDELINES, MANAGEMENT.
Lecture available August 01, 2021 -January 31, 2022

Objectives:

- Identify individuals at risk for hereditary cancer syndrome
- Be able to provide medical management recommendations for BRCA gene mutation carrier.

Format: Online lecture

AMA PRA Category 1: MD, DO, CRNP, PA Attendance: All other healthcare providers

Angela R. Olesko | **Physician Relations Manager Senior** | **AHN**
Cell: 412.523.9858 | { HYPERLINK "mailto:angela.olesko@ahn.org" }

Allegheny Health Network

Figure 1 CME Promotional Flyer

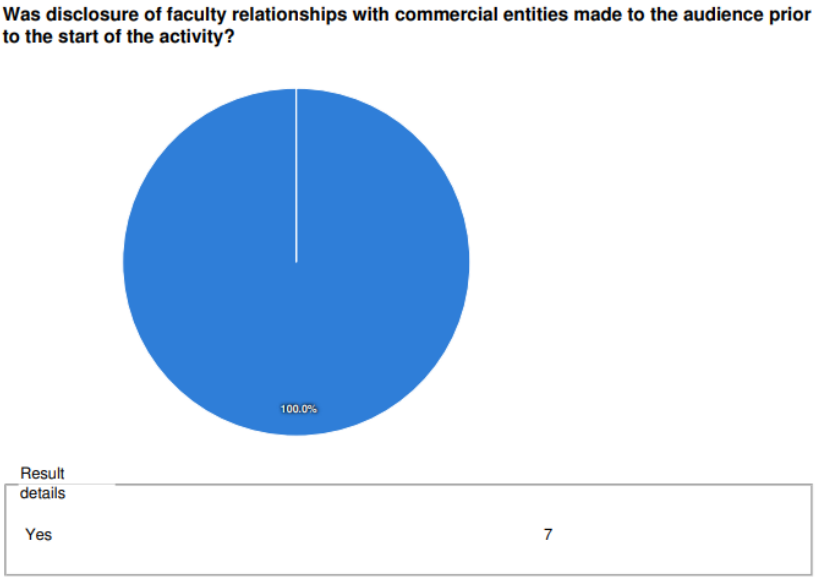


Figure 2 Faculty relationships and statement on financial disclosures

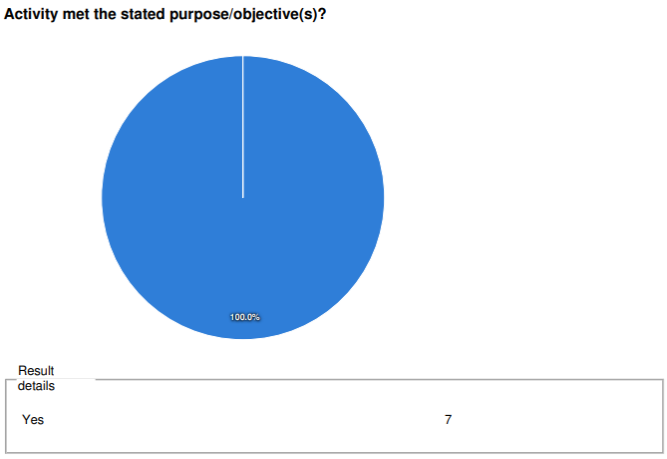


Figure 3 Presentation statement on the listed learning objectives

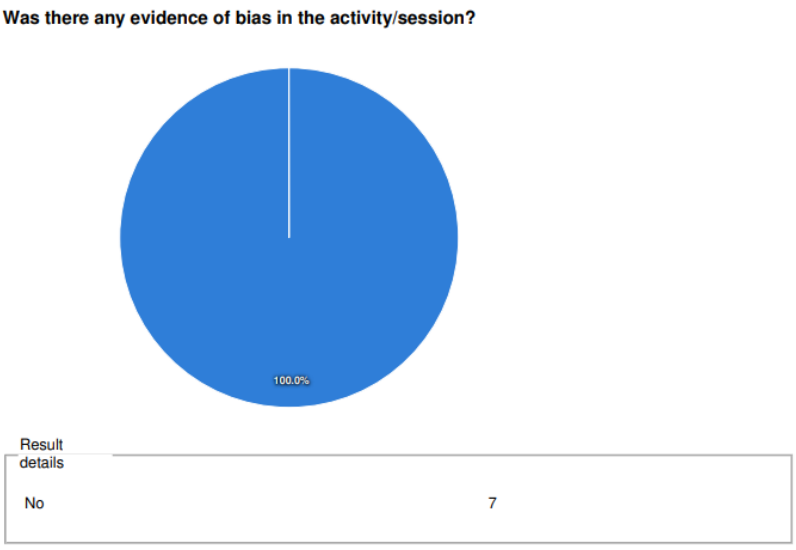


Figure 4 Percieved bias in the presentation materials

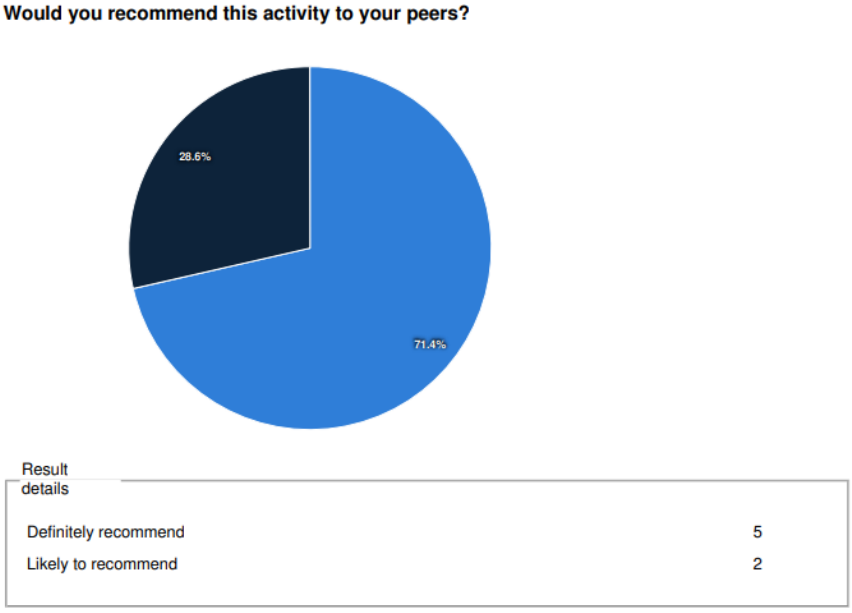
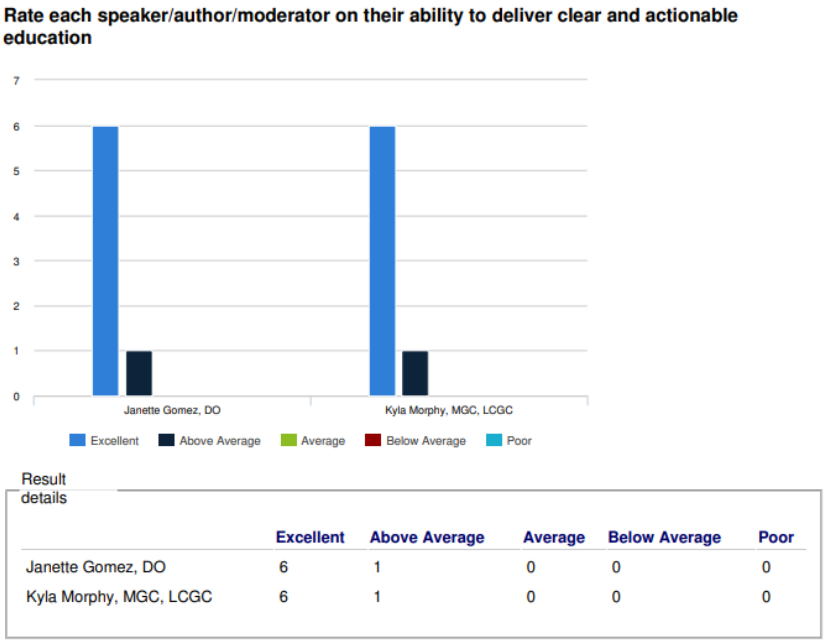
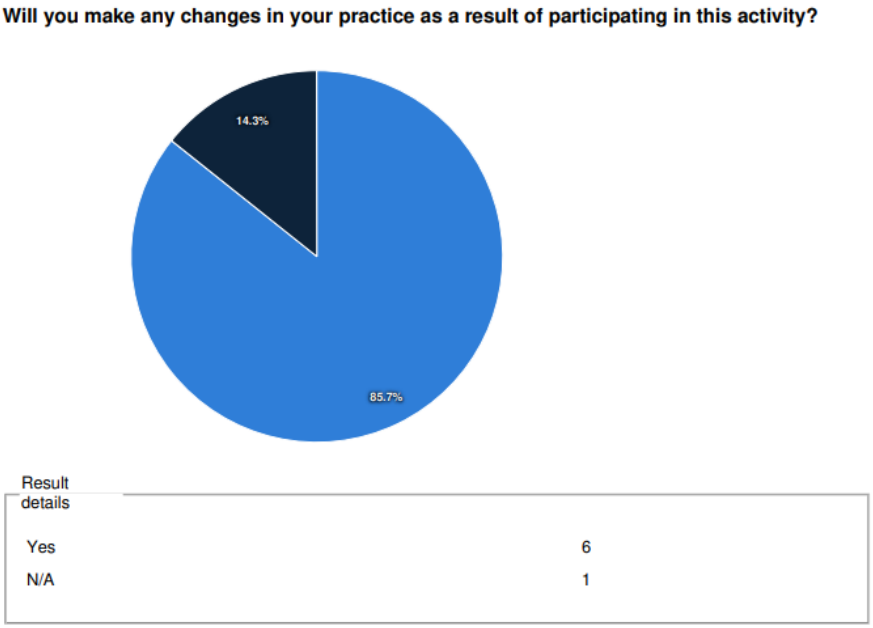


Figure 5 Peer recommendation of the presentation



	Excellent	Above Average	Average	Below Average	Poor
Janette Gomez, DO	6	1	0	0	0
Kyla Morphy, MGC, LCGC	6	1	0	0	0

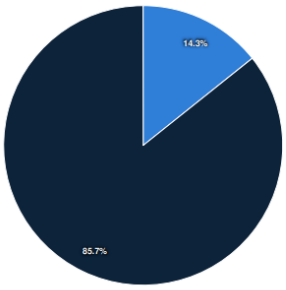
Figure 6 Ratings of each presenter and their ability to deliver clear and actionable education over the course of the CME lecture



Yes	6
N/A	1

Figure 7 Clinical changes to be made as a result of the presentation

If yes, what changes will you make?

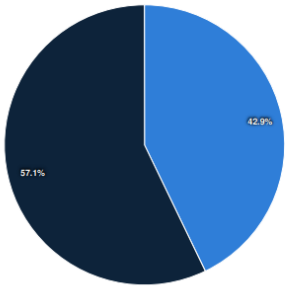


Result details

Left Blank	1
User entered value	6
Average submission length in words (ex blanks)	10.83
i will continue to be more aware of family history in my patients to screen whether or not they are a candidate for genetic testing.	
refer more patients for counseling	
Genetic referral to those with genetic risk	
Will be able to better educate oncology patients on genetic counseling services.	
knowing when a referral needs to be made	
make use of the flowsheet to make referrals	

Figure 8 Free responses entered to question on clinical changes to be made as a result of the presentation

What were some positive aspects of the educational presentation?

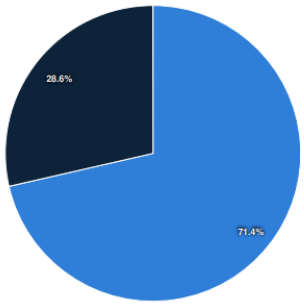


Result details

Left Blank	3
User entered value	4
Average submission length in words (ex blanks)	13.25
better understanding of patients that should be referred for counseling	
The material was presented in a way that made complicated information much easier to understand. I really liked the frequent use of graphs/tables.	
clear understanding and examples of when to refer	
just knowing the percentage of cancers that maybe hereditary was enlightening	

Figure 9 Free responses to positive presentation aspects

What could be improved for future educational presentations?



Result details

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User entered value	2
Average submission length in words (ex blanks)	1.50

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Figure 10 Free responses for presentation improvements

6.0 Bibliography

1. Meyer, L.A., Anderson, M.E., Lacour, R.A., Suri, A., Daniels, M.S., Urbauer, D.L., Nogueras-Gonzalez, G.M., Schmeler, K.M., Gershenson, D.M., and Lu, K.H. (2010). Evaluating women with ovarian cancer for BRCA1 and BRCA2 mutations: missed opportunities. Obstet Gynecol 115, 945-952.

2. Burke, W., Culver, J., Pinsky, L., Hall, S., Reynolds, S.E., Yasui, Y., and Press, N. (2009). Genetic assessment of breast cancer risk in primary care practice. Am J Med Genet A 149A, 349-356.

3. National Cancer Institute. (2020). BRCA Gene Mutations: Cancer Risk and Genetic Testing. In.

4. Koil, C.E., Everett, J.N., Hoechstetter, L., Ricer, R.E., and Huelsman, K.M. (2003). Differences in physician referral practices and attitudes regarding hereditary breast cancer by clinical practice location. Genet Med 5, 364-369.

5. Desai, S., and Jena, A.B. (2016). Do celebrity endorsements matter? Observational study of BRCA gene testing and mastectomy rates after Angelina Jolie's New York Times editorial. BMJ 355, i6357.

6. Mouchawar, J., Valentine Goins, K., Somkin, C., Puleo, E., Hensley Alford, S., Geiger, A.M., Taplin, S., Gilbert, J., Weinmann, S., and Zapka, J. (2003). Guidelines for breast and ovarian cancer genetic counseling referral: adoption and implementation in HMOs. Genet Med 5, 444-450.

7. Wilkes, M.S., Day, F.C., Fancher, T.L., McDermott, H., Lehman, E., Bell, R.A., and Green, M.J. (2017). Increasing confidence and changing behaviors in primary care providers engaged in genetic counselling. BMC Med Educ 17, 163.

8. Miki, Y., Swensen, J., Shattuck-Eidens, D., Futreal, P.A., Harshman, K., Tavtigian, S., Liu, Q., Cochran, C., Bennett, L.M., Ding, W., et al. (1994). A strong candidate for the breast and ovarian cancer susceptibility gene BRCA1. Science 266, 66.

9. Wooster, R., Bignell, G., Lancaster, J., Swift, S., Seal, S., Mangion, J., Collins, N., Gregory, S., Gumbs, C., and Micklem, G. (1995). Identification of the breast cancer susceptibility gene BRCA2.

10. Jolie, A. (2013). My Medical Choice. In The New York Times. (New York), p 25.

11. Robson, M., and Offit, K. (2007). Clinical practice. Management of an inherited predisposition to breast cancer. N Engl J Med 357, 154-162.

12. Finch, A.P., Lubinski, J., Møller, P., Singer, C.F., Karlan, B., Senter, L., Rosen, B., Maehle, L., Ghadirian, P., Cybulski, C., et al. (2014). Impact of oophorectomy on cancer incidence and mortality in women with a BRCA1 or BRCA2 mutation.

13. Rebbeck, T.R., Friebel, T., Lynch, H.T., Neuhausen, S.I., van 't Veer, L., Garber, J.E., Evans, G.R., Narod, S.A., Isaacs, C.F., Matloff, E., et al. (2004). Bilateral prophylactic mastectomy reduces breast cancer risk in BRCA1 and BRCA2 mutation carriers: the PROSE Study Group.

14. Rebbeck, T.R., Kauff, N.D., and Domchek, S.M. (2009). Meta-analysis of risk reduction estimates associated with risk-reducing salpingo-oophorectomy in BRCA1 or BRCA2 mutation carriers. J Natl Cancer Inst 101, 80-87.

15. Buchanan, A.H., Voils, C.I., Schildkraut, J.M., Fine, C., Horick, N.K., Marcom, P.K., Wiggins, K., and Skinner, C.S. (2017). Adherence to Recommended Risk Management among Unaffected Women with a BRCA Mutation. J Genet Couns 26.

16. Schwartz, M.D., Isaacs, C., Graves, K.D., Poggi, E., Peshkin, B.N., Gell, C., Finch, C., Kelly, S., Taylor, K.I., and Perley, L. (2012). Long-term outcomes of BRCA1/BRCA2 testing: risk reduction and surveillance.

17. Schwartz, M.D., Kaufman, E., Peshkin, B.N., Isaacs, C., Hughes, C., DeMarco, T., Finch, C., and Lerman, C. (2003). Bilateral prophylactic oophorectomy and ovarian cancer screening following BRCA1/BRCA2 mutation testing.

18. National Comprehensive Cancer Network. (2022). Genetic/ Familial High Risk Assessment: Breast, Ovarian and Pancreatic. In. (

19. Elrick, A., Ashida, S., Ivanovich, J., Lyons, S., Biesecker, B.B., Goodman, M.S., and Kaphingst, K.A. (2017). Psychosocial and Clinical Factors Associated with Family Communication of Cancer Genetic Test Results among Women Diagnosed with Breast Cancer at a Young Age. J Genet Couns 26.

20. McGivern, B., Everett, J., Yager, G.G., Baumiller, R.C., Hafertepen, A., and Saal, H.M. (2004). Family communication about positive BRCA1 and BRCA2 genetic test results.

21. Hughes, C., Lerman, C., Schwartz, M., Peshkin, B.N., Wenzel, L., Narod, S., Corio, C., Tercyak, K.P., Hanna, D., Isaacs, C., et al. (2002). All in the family: evaluation of the process and content of sisters' communication about BRCA1 and BRCA2 genetic test results.

22. d'Agincourt-Canning, L. (2001). Experiences of genetic risk: disclosure and the gendering of responsibility.

23. Wiseman, M., Dancyger, C., and Michie, S. (2010). Communicating genetic risk information within families: a review.

24. Finlay, E., Stopfer, J.E., Burlingame, E., Evans, K.G., Nathanson, K.I., Weber, B.I., Armstrong, K., Rebbeck, T.R., and Domchek, S.M. (2008). Factors determining dissemination of results and uptake of genetic testing in families with known BRCA1/2 mutations.

25. Gaff, C.L., Clarke, A.J., Atkinson, P., Sivell, S., Elwyn, G., Iredale, R., Thornton, H., Dundon, J., Shaw, C., and Edwards, A. (2007). Process and outcome in communication of genetic information within families: a systematic review.

26. Colditz, G.A., Kaphingst, K.A., Hankinson, S.E., and Rosner, B. (2012). Family history and risk of breast cancer: nurses' health study. Breast Cancer Res Treat 133, 1097-1104.

27. Brewer, H.R., Jones, M.E., Schoemaker, M.J., Ashworth, A., and Swerdlow, A.J. (2017). Family history and risk of breast cancer: an analysis accounting for family structure. Breast Cancer Res Treat 165.

28. Braithwaite, D., Miglioretti, D.L., Zhu, W., Demb, J., Trentham-Dietz, A., Sprague, B., Tice, J.A., Onega, T., Henderson, L.M., Buist, D.S.M., et al. (2018). Family History and Breast Cancer Risk Among Older Women in the Breast Cancer Surveillance Consortium Cohort. JAMA Intern Med 178.

29. Hughes, K.S., Roche, C., Campbell, C.T., Siegel, N., Salisbury, L., Chekos, A., Katz, M.S., and Edell, E. (2003). Prevalence of family history of breast and ovarian cancer in a single primary care practice using a self-administered questionnaire. Breast J 9.

30. Quillin, J.M., Krist, A.H., Gyure, M., Corona, R., Rodriguez, V., Borzelleca, J., and Bodurtha, J.N. (2014). Patient-reported hereditary breast and ovarian cancer in a primary care practice. J Community Genet 5.

31. Rupert, D.J., Squiers, L.B., Renaud, J.M., Whitehead, N.S., Osborn, R.J., Furberg, R.D., Squire, C.M., and Tzeng, J.P. (2013). Communicating risk of hereditary breast and ovarian cancer with an interactive decision support tool. Pateint Education and Counseling 92.

32. Bellcross, C.A., Leadbetter, S., Alford, S.H., and Peipins, L.A. (2013). Prevalence and healthcare actions of women in a large health system with a family history meeting the 2005 USPSTF recommendation for BRCA genetic counseling referral. Cancer Epidemiol Biomarkers Prev 22, 728-735.

33. Tri, N., Morphy, K., Bianchin, G., Schwaderer, K., Sobolewski, B., Voas, C. (2019). Glimmer of Hope Young Women's Breast Program. In. (Allegheny Health Network.

34. Hamilton, J.G., Abdiwahab, E., Edwards, H.M., Fang, M.L., Jdayani, A., and Breslau, E.S. (2017). Primary care providers' cancer genetic testing-related knowledge, attitudes, and communication behaviors: A systematic review and research agenda. J Gen Intern Med 32, 315-324.

35. Harris, R., and Harris, H.J. (1995). Primary care for patients at genetic risk. BMJ (Clinical research ed) 311, 579-580.

36. Watson, E.K., Shickle, D., Qureshi, N., Emery, J., and Austoker, J. (1999). The 'new genetics' and primary care: GPs' views on their role and their educational needs. Family Practice 16, 420-425.

37. Bell, R.A., McDermott, H., Fancher, T.L., Green, M.J., Day, F.C., and Wilkes, M.S. (2015). Impact of a randomized controlled educational trial to improve physician practice behaviors around screening for inherited breast cancer. J Gen Intern Med 30, 334-341.

38. Ready, K.J., Daniels, M.S., Sun, C.C., Peterson, S.K., Northrup, H., and Lu, K.H. (2010). Obstetrics/gynecology residents' knowledge of hereditary breast and ovarian cancer and Lynch syndrome. J Canc Educ 25, 401-404.

39. Vadaparampil, S.T., Scherr, C.I., Cragun, D., Malo, T.I., and Pal, T. (2015). Pre-test genetic counseling services for hereditary breast and ovarian cancer delivered by non-genetics professionals in the state of Florida. Clin Genet 87, 473-477.

40. Cohn, J., Blazey, W., Tegay, D., Harper, B., Koehler, S., Laurent, B., Chan, V., Jung, M.K., and Krishnamachari, B. (2015). Physician Risk Assessment Knowledge Regarding BRCA Genetics Testing. J Cancer Educ 30, 573-579.

41. Culver, J.O., Bowen, D.J., Reynolds, S.E., Pinsky, L.E., Press, N., and Burke, W. (2009). Breast cancer risk communication: assessment of primary care physicians by standardized patients. Genet Med 11.

42. Pujol, P., Lyonnet, D.S., Frebourg, T., Blin, J., Picot, M.C., Lasset, C., Dugast, C., Berthet, P., de Paillerets, B.B., Sobol, H., et al. (2013). Lack of referral for genetic counseling and testing in BRCA1/2 and Lynch syndromes: a nationwide study based on 240,134 consultations and 134,652 genetic tests. Breast Cancer Res Treat 141, 135-144.

43. Trivers, K.F., Baldwin, L.M., Miller, J.W., Matthews, B., Andrilla, C.H., Lishner, D.M., and Goff, B.A. (2011). Reported referral for genetic counseling or BRCA 1/2 testing among United States physicians: a vignette-based study. Cancer 117, 5334-5343.

44. Paneque, M., Turchetti, D., Jackson, L., Lunt, P., Houwink, E., and Skirton, H. (2016). A systematic review of interventions to provide genetics education for primary care. BMC Family Practice 17.

45. Narod, S.A. (2010). BRCA mutations in the management of breast cancer: the state of the art.

46. Puleo, E., Zapka, J., White, M.J., Mouchawar, J., Somkin, C., and Taplin, S. (2002). Caffeine, cajoling, and other strategies to maximize clinician survey response rates. Evaluation & the Health Professions 25, 169-184.

47. Davis, D.A., Thomson, M.A., Oxman, A.D., and Haynes, R.B. (1995). Changing physician performance. A systematic review of the effect of continuing medical education strategies. Journal of the American Medical Association 274, 700-706.

48. Drury, N., Bethea, J., Guilbert, P., and Qureshi, N. (2007). Genetics support to primary care practitioners - a demonstration project. J Genet Counsel 16, 583-591.

49. Houwink, E.J., van Teeffelen, S.R., Muijtjens, A.M., Henneman, L., Jacobi, F., van Luijk, S.J., Dinant, G.J., van der Vleuten, C., and Cornel, M.C. (2014). Sustained effects of online genetics education: a randomized controlled trial on oncogenetics. European Journal of Human Genetics 22, 310-316.

50. Bethea, J., Qureshi, N.F., Drury, N., and Guilbert, P. (2008). The impact of genetic outreach education and support to primary care on practitioner's confidence and competence in dealing with familial cancers. Community Genet 11, 289-294.

51. Davis, D., O'Brien, M.A., Freemantle, N., Wolf, F.M., Mazmanian, P., and Taylor-Vaisey, A. (1999). Impact of formal continuing medical education: do conferences, workshops, rounds, and other traditional continuing education activities change physician behavior or health care outcomes? JAMA 282.

52. Pal, T., Cragun, D., Lewis, C., Doty, A., Rodriguez, M., Radford, C., Thompson, Z., Kim, J., and Vadaparampil, S.T. (2013). A statewide survey of practitioners to assess knowledge and clinical practices regarding hereditary breast and ovarian cancer. Genetic Testing and Molecular Biomarkers 17.

53. Carroll, J.C., Wilson, B., Allanson, J., Grimshaw, J., Blaine, S.M., Meschino, W.S., Permaul, J.A., and Graham, I.D. (2011). GenetiKit: a randomized controlled trial to enhance delivery of genetics services by family physicians. Family Practice 28, 615-623.

54. Tu, K., and Davis, D. (2002). Can we alter physician behavior by educational methods? Lessons learned from studies of the management and follow-up of hypertension. The Journal of Continuing Education in the Health Professions 11, 11-22.

55. Berg, R., Brand, A., Grant, J., Kirk, J., and Zimmerman, T. (2013). Leveraging Recorded Mini-Lectures to Increase Student Learning. In. (Online Classroom), pp 5,8.

56. Guo, P., Kim, J., and Rubin, R. (2014). How Video Production Affects Student Engagement: An Empirical Study of MOOC Videos. In ACM Conference on Learning. pp 41-50.

57. Center for Disease Control and Prevention. (2020). 10 Essential Public Health Services. In. (

58. Harding, B., Webber, C., Rühland, L., Dalgarno, N., Armour, C., Birtwhistle, R., Brown, G., Carroll, J.C., Flavin, M., Phillips, S.P., et al. (2019). Bridging the gap in genetics: a progressive model for primary to specialist care. BMC Medical Education 19, 195.

59. Emery, J., Morris, H., Goodchild, R., Fanshawe, T., Prevost, A.T., Bobrow, M., and Kinmonth, A.L. (2007). The GRAIDS Trial: a cluster randomised controlled trial of computer decision support for the management of familial cancer risk in primary care. British Journal of Cancer 97, 486-493.

60. Talwar, D., Tseng, T.S., Foster, M., Xu, L., and Chen, L.S. (2017). Genetics/genomics education for nongenetic health professionals: a systematic literature review. Genetics in Medicine 19, P725-732.