Hereditary cancer screening attitudes and practices among primary care providers:
A mixed-methods study

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Abstract

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Purpose: Across the United States nationally recommended strategies for identifying asymptomatic individuals with predispositions to hereditary cancer syndromes are severely underutilized. The long term doctor-patient relationship between primary care providers (PCPs) and their patients makes them uniquely positioned to carry out such testing, but historically PCPs report being unknowledgeable about these topics. This study aims to 1) describe how much confidence PCPs have conducting clinical cancer consultations, 2) identify PCPs' current practices for obtaining their patients' personal and familial cancer histories, and to 3) understand how PCPs use their patients' personal and familial histories of cancer to inform their ordering of genetic testing.

Methods: An online survey was distributed and follow up semi-structured interviews were conducted among a group of PCPs located throughout Washington, Wyoming, and Montana. Binary logistic regression analyses were used to explore the relationship between background variables and the PCPs' confidence scores. Interview transcripts were analyzed via a directed content analysis approach.

Results: The regression analyses showed that PCPs who practiced in more urban rural urban continuum codes (RUCC) and scored higher on the *Consolidated Framework for Implementation Research* (*CFIR) self-efficacy subscale* were more likely to have higher confidence levels with respect to initiating the conversation about cancer risk and responding to their patients' questions about cancer risk based on their family history, as well as their confidence in explaining lifetime and age-related cancer risk to their patients.

The interview results revealed that most PCPs gather personal and familial cancer history from their patients via a general direct questioning process with the patient in tandem with another method. Most PCPs believe that collecting this information is important primarily because it informs their future cancer screening recommendation. Most PCPs are not ordering cancer risk genetic testing and those that are report they are really only comfortable ordering rather limited range of cancer genetic tests.

Conclusions: These results suggest that PCPs recognize the benefits these early identification strategies can provide, but they don't feel comfortable carrying out the ordering and interpretation of cancer risk genetic testing themselves. On this basis, better educational strategies, alongside implementation of updated procedures and automated prompts in electronic medical records are needed to further aid PCPs in successfully implementing universal screening and testing for predispositions to hereditary cancers in primary care settings.

1. INTRODUCTION

Inherited genetic mutations play a role in 5-10% of all cancer cases (NCI, 2017). The American College of Medical Genetics (ACMG) recommends that early identification of individuals with predispositions to hereditary cancer syndromes should be carried out at a population-level (Hampel, Bennett, Buchanan, Pearlman, & Wiesner, 2014). It has been recommended that identification of individuals with predispositions to hereditary cancer syndromes should be done via assessment of personal and familial cancer history followed by cancer risk genetic testing of high-risk individuals (Moyer & USPSTF, 2014 & EGAPP Working Group, 2009). Identification of asymptomatic individuals with predispositions to hereditary cancer syndromes can play an integral role in cancer prevention and treatment, which can ultimately save the lives of these patients (Järvinen, 2000, & Järvinen et al., 2009 & Kotsopoulos, 2018). Despite these recommendations, these early identification strategies are underutilized across the nation (Singh, Schiesser, Anand, Richardson, & El–Serag, 2010, Levy et al., 2011 & Vogel, 2012). For instance, among women with early onset breast cancers, only 30% had been offered BRCA1/2 testing (Levy et al., 2011). Given that BRCA1/2 testing is among the most well established cancer risk genetic tests (Lee, 2002) it would stand to reason that other cancer risk genetic tests are even more severely underutilized, particularly prior to cancer onset.

To improve the population-level performance of these strategies, the literature suggests that primary care providers (PCPs) should carry out these screenings due to their unique, long-term relationships with their patients (Selby, Bartlett-Esquilant, & Cornuz, 2018). However, PCPs face a wide variety of barriers to implementing these screening and testing schemes, including issues recognizing hereditary risk (Mahon, 2016 & Freedman 2003), a shortage of genetic counselors to provide counseling (Hoskovec et al., 2017), lack of time and resources

(Mahon, 2016), incomplete knowledge of potential benefits and preventative opportunities (Freedman, 2003), and a lack of confidence in that genetic testing related knowledge (Hamilton et al., 2016). Luckily, most of the barriers cited by PCPs are addressable. If we want to address these barriers in the future it is imperative that we seek to further understand PCPs' attitudes and practices surrounding hereditary cancer screening and genetic testing so that we can help implement these screening schemes into their everyday practices so these schemes can reach their true population-level potential.

Research indicates that most PCPs recognize the utility of having cancer risk genetic test results to inform cancer risk management strategies (Hamilton et al., 2016 & Lowstuter et al., 2008 & Guerra, Sherman & Armstrong, 2009, Evenson et al., 2016). However, most PCPs are not prepared to provide counseling about genetic testing to their patients (Freedman et al., 2003, Lowstuter et al., 2008, Mainous, Johnson, Chirina, & Baker, 2013 & Hamilton et al., 2016, Evenson et al., 2016, Hann et al., 2017, Dekanek et al., 2019). Further, despite this incomplete knowledge about genetic testing, the literature suggests that PCPs are still largely in favor of clinical genetic testing (Mainous, Johnson, Chirina, & Baker, 2013 & Guerra, Sherman, & Armstrong, 2009).

Exactly how PCPs put this incomplete knowledge of genetic testing to use with respect to cancer risk genetic testing remains unknown. The literature suggests that on average, PCPs do not collect family history information in sufficient detail to accurately detect cancer risk (Sweet, Bradley, & Westman, 2002 & Flynn et al., 2010 & Mahon, 2016). Further, exactly how PCPs collect hereditary cancer risk information, including what information they collect from their patients, and how that informs their ordering of genetic testing remains understudied.

This study sought to explore PCPs current practices for obtaining their patients' personal and familial cancer histories by investigating what PCPs bring up in the conversation about family history with their patients, and if the PCPs believe collecting personal and familial cancer history information is important. This study also sought to further understand how PCPs use their patients' personal and familial cancer history information to inform their ordering of genetic testing by asking PCPs if patients with personal or familial histories of cancer receive cancer risk genetic testing from them and if a personal or familial cancer history causes a PCP to change how they care for that patient. Finally, this study sought to describe how much confidence PCPs have performing 7 tasks that that are important to conducting a successful clinical cancer consultation. This was done by investigating, on average, how confident PCPs are conducting that clinical cancer risk consultation, and what factors influence high confidence scores.

2. METHODS

2.1 Parent Study

The parent study of this project is the Early Detection of GEnetic risk (EDGE) study. The larger goal of the EDGE study is to evaluate different methods for implementing population based screening and hereditary cancer risk genetic testing of high-risk individuals to improve hereditary cancer treatment and prevention in primary care clinics. 2 healthcare systems enrolled in the EDGE study. Healthcare system M is made up of 6 primary care clinics located in Washington state. Healthcare system M employed a total of 47 PCPs at the time of the study. Healthcare system B is also made up of 6 primary care clinics. These clinics are located in Montana and northern Wyoming. Healthcare system B employed a total of 106 PCPs at the time of the study. The EDGE study sent surveys to all 153 PCPs employed by Healthcare systems M and B at the baseline of the project. This project used data collected from that baseline PCP survey to identify and understand how the PCPs at the participating clinics obtain and use cancer risk information, including personal and familial cancer history to inform genetic testing and how confident these PCPs are conducting clinical cancer risk consultations with their patients.

2.2 Study Design

This study utilized a sequential mixed methods (Creswell, 2018), cross-sectional approach. In the first step participants completed an online survey. One of the questions in the survey asked if they would be willing to complete a follow-up interview. In the second step the PCPs who had agreed to be interviewed completed one-on-one semi-structured interviews with a member of the EDGE study team. IRB approval was obtained through The University of Washington prior to the beginning of the study.

2.3 Participants

The 12 participating clinics employed a total of 153 PCPs. For the purposes of this study PCPs were defined as clinic employees that were considered a primary care provider, who worked at least half time at that particular clinic, with at least one patient on their panel or regular patient load. We excluded residents, urgent care, flex, and float providers from our sample.

2.4 Quantitative Approach: PCP Surveys

2.4.1 Data Collection of PCP Surveys

We began by emailing the clinic managers at the 12 participating clinics telling them that we planned to send the survey link out to the PCPs soon. This way they were able to alert their PCPs to keep an eye out for the forthcoming survey invitation. Soon after, we emailed all 153 PCPs a hyperlink to the EDGE PCP survey and offered a $50 gift card for completing the survey. We then sent reminder emails at varying times of the day (6:00 am, 10:45 am, 3:00 pm) to maximize the chances of catching a PCP at a time that they would be willing to complete the survey. PCPs at healthcare system M were sent up to 12 reminders. PCPs at healthcare system B were sent up to 9 reminders.

2.4.1.1 *PCP Survey Measures*

PCP Confidence

To measure how much confidence PCPs have when conducting a clinical cancer consultation, the baseline EDGE PCP survey included a “Cancer and Genetics: Confidence” scale. This scale consisted of 7 items regarding a PCPs self-efficacy to conduct a cancer risk consultation. This scale was adapted from a scale used by Hann and colleagues that they created, piloted, and refined for use in the context of assessing health care providers’ self-efficacy in performing a clinical cancer risk consultations for ovarian cancer (2017). All 7 questions were answered on a four-point Likert-style scale with the following options: “not confident at all”, “not very confident”, “somewhat confident”, and “very confident”. The internal reliability for this scale was high for Hann and colleagues ($\alpha = .936$). The reliability for our modified version, the *Cancer & Genetics : Confidence* scale was good ($\alpha = 0.811$).

I performed a principle component analysis with varimax rotation to investigate if the 7 items in the survey carried equal weight (Abdi & Williams, 2010). The 7 items formed themselves into 2 domains with eigen values greater than one, resulting in two two-item subscales (Abdi & Williams, 2010). The first domain that emerged, *Addressing questions*, loaded 2 items onto it from the scale that addressed a PCP’s ability to initiate a conversation about cancer with their patients and in their ability to respond to patients’ questions about genetic testing for cancer risk. The *Addressing questions* domain has a Cronbach’s alpha of 0.426.

The second domain to emerge, *Explaining cancer risk,* originally loaded 3 items onto it from the *Cancer & Genetics : Confidence* scale that addressed items about explaining lifetime and age-related cancer risk and providing support to patients going through the assessment. The item regarding providing support to patients going through the assessment was removed to increase the Cronbach’s alpha from 0.690 to 0.857. These domains served as the main outcome variables for subsequent analyses.

Sociodemographic variables

The demographic section of the survey asked the PCPs to self-report their sex (male, female), race (White, Black or African American, Asian, American Indian/Alaskan Native, Other), and ethnicity (Hispanic or Latino, Not Hispanic or Latino). Due the small number of people of color in our study, the race variable was dichotomized into white (White) and nonwhite categories (Black or African American, Asian, American Indian/Alaskan Native, Other). Then, race and ethnicity were indexed into a single variable (Bhattacherjee, 2012).

Number of years the PCP has been practicing medicine

The number of years a PCP had been practicing medicine at the time of the survey was assessed by a survey item that asked them to indicate how many years they have been in practice. Answer choices ranged from “Less than 1 year” to “More than 30 years”. For descriptive statistics, the number of years in practice was dichotomized into providers with up to 10 years of experience and providers with more than 10 years of experience based on the bimodal trend in the histogram. This cut point is also where the median for the measurement fell. For all logistic regression analyses, the continuous version of this variable was used.

Number of years the PCP has been practicing medicine at their current clinic

The number of years a PCP had been practicing medicine at their current clinic at the time of the survey was assessed by a survey item that asked “How long have you worked at this clinic?” with answer choices ranging from “Less than 1 year” to “More than 10 years”. This variable was dichotomized into providers with up to 9 years at their current clinic, and those with 10 or more years at the clinic because there is a clear split in the data with most physicians working more than 10 years at the clinic. This dichotomized version of the variable was used for all descriptive and inferential analyses.

Degree(s) Held

What degrees a provider held at the time of the survey was assessed by a survey item that asked “What degree(s) do you currently have?” with answer choices of “MD”, “DO”, “PA-C”, “ARNP/APRN”, “PhD”, “MPH”, “MBA”, or “Other”. PCPs could choose multiple degrees and if they chose “Other” they were then prompted to enter in the type of degree they held that was missing from this list. This degree(s) held variable was trichotomized by the nature of the medical degree that the individual held, with the groupings being medical doctor degrees (MD/DO), physician assistant degrees (PA-C), and nursing degrees (ARNP/APRN/DNP). The trichotomized version of this variable was used for all descriptive and inferential analyses.

Provider Specialty

The PCP’s specialty at the time of the survey was assessed by a survey item that asked “What is your primary practice specialty? (If you have more than one, please choose the one where you spend the most time.)”. Answer choices included “Family Medicine”, “Internal Medicine” or “Other”. If they selected “Other” they were prompted to enter in the specialty they identified with that was missing from the previous options. This variable was dichotomized into doctors who specialized in Family Medicine and doctors who specialized in Internal Medicine. The dichotomous version of this variable was used for all descriptive and inferential analyses.

Rural Urban Continuum Code (RUCC)

RUCCs categorize addresses into different codes, ranging from 1 to 9, depending on population size, degree of urbanization, and adjacency to metropolitan areas (USDA, 2020). A RUCC was assigned to each provider by looking up the corresponding RUCC for the street address of the clinic where they were currently practicing. For the purposes of descriptive statistics, the RUCC variable was dichotomized into urban and rural categories, where urban areas had RUCC of 1-3 and rural areas had RUCC of 4 or more (Lister, Ellis, & Yoon, 2020; Segel & Lengerich, 2020). For all logistic regression analyses RUCC was used as a continuous variable.

Consolidated Framework for Implementation Research (CFIR) self-efficacy subscale

To determine the PCPs' beliefs in their own capabilities to use the results of genetic testing, family history of cancer, and population-based screening for hereditary cancer to better manage patient outcomes, PCPs were asked to rate their agreement on a 5 point Likert scale ranging from “strongly disagree” to “strongly agree” with the following statements: 1) “My training has prepared me to treat patients whose family history/genetics place them at high risk for medical conditions.” 2) “I am confident in my ability to use the results of population-based screening for hereditary cancer.” This scale was extrapolated from Orlando and colleagues' research where they identified PCPs' self-efficacy as a high priority CFIR construct for evaluating the implementation of genomic medicine interventions in clinical care (2017). The *CFIR self-efficacy subscale* score was calculated by averaging the PCPs' responses to the questions above and modeled continuously. The reliability of this subscale was good (α = 0.80).

CFIR knowledge and beliefs subscale

To determine the PCPs' attitudes and value placed on the intervention and familiarity with facts regarding population-based screening for hereditary cancers, PCPs were asked to rate their agreement on a 5 point Likert scale ranging from "strongly disagree" to "strongly agree" with the following statements: 1) "The information generated by population-based screening for hereditary cancer is important for patient care." 2) "I believe that population-based screening for hereditary cancer is relevant to my current clinical practice." Similarly to the *CFIR self-efficacy subscale*, this scale was extrapolated from Orlando and colleagues' research where they identified PCPs' knowledge and beliefs about genomic medicine interventions as a high priority CFIR construct in research pertaining to implementation of genomic medicine interventions in clinical care (2017). The *CFIR knowledge and beliefs subscale* scores were calculated by averaging the PCPs' responses to the questions listed above and modeled continuously. The reliability of this subscale was good (α = 0.84).

Active collection of familial cancer history information

To assess if a PCP actively collects their patients' familial cancer history information a survey item asked "Currently, do you actively collect familial cancer history from your patients?" Answer choices included "No", "Yes, but only for certain patients", and "Yes, I screen all patients for family history of cancer".

Method for collecting familial cancer history information

The method(s) a PCP uses to collect familial cancer history information was assessed by a survey item that asked “How is familial cancer history collected? (choose all that apply)”. Answer choices included “Paper form completed by patient (e.g. family history checklist)”, “Electronic form completed by patient (e.g. tablet entry by patient in waiting room or MyChart form patient completes at home)”, “Electronic form completed by clinician (e.g. standardized form, but not patient-facing)”, “Information is included in visit note (e.g. not a standardized form/location)”, or “Other”. If the PCP selected the “Other” box they were then asked to input what other method they use to collect familial cancer history information.

Use of familial cancer history information

To assess whether or not a PCP uses their patients’ familial cancer history information to inform their medical recommendations to their patients a survey item asked “Do you consider familial cancer history when making care recommendations?” Answer choices were “Yes” or “No”. PCPs who indicated that they do consider familial cancer history information when making care recommendations were then asked “How is that information used?”. For this question PCPs were then given the opportunity to freely enter in their response.

Use of screening tools

To assess which screening tools PCPs use with their patients PCPs were asked “Have you ever used any of the following tools with your patients?” and the list included “Family tree (pedigree)”, “Breast cancer referral screening tool (B-RST)”, “Breast cancer risk assessment tools (e.g. BRCAT/Gail model; Claus model; Tryer-Cuzick/IBIS model/ BOADICEA/ BRCAPRO)”, “Colorectal cancer risk assessment tools (e.g. PREMM; MMRpredict; MMRpro)”, and “Other”. For each screening tool listed PCPs had the choice of responding “No”, “Yes, but only for certain patients”, and “Yes, for all my patients.” If either of the yes categories was checked for the “Other” option the PCP was asked to specify what other screening tool they use. PCPs who indicated that they have used breast cancer risk assessment tools were asked to specify which breast cancer risk assessment tools they used and PCPs who indicated that they have used colorectal cancer risk assessment tools were asked to specify which colorectal cancer risk assessment tools they have used.

Experience talking to patients about cancer genetic testing

To assess how often PCPs talk to their patients about cancer risk genetic testing PCPs were asked “In general, how often do you talk to patients about cancer genetic testing in your practice?” Answer choices included: “Never”, “Rarely (less than once a year)”, “Occasionally (a couple times a year)”, “Often (a couple times a month)”, and “Almost always (several times a week)”.

Experience referring patients for cancer genetic counseling or testing

To determine if a PCP has experience referring patients for cancer genetic counseling or testing a survey question asked “Have you ever referred patients for cancer genetic counseling or testing?” Answer choices included “Yes” and “No”. PCPs who indicated that they have referred patients for cancer genetic counseling or testing were then asked “Where do you refer them to?” PCPs were allowed to freely type in their response to this question. Then these same PCPs who indicated that they do have experience referring patients for cancer genetic counseling or testing were asked “How often do you refer patients for cancer genetic counseling or testing?” Answer choices included “Never”, “Rarely (less than once a year)”, “Occasionally (a couple times a year)”, “Often (a couple times a month)”, and “Almost always (several times a week)”.

Experience ordering cancer genetic testing

To determine if a PCP has experience ordering cancer genetic testing a survey item asked “Do you have experience ordering cancer genetic testing?” Answer choices included “Yes” and “No”. PCPs who indicated that they do have experience ordering cancer genetic testing were then asked “How often do you order cancer genetic testing?” Answer choices for this question included “Never”, “Rarely (less than once a year)”, “Occasionally (a couple times a year)”, “Often (a couple times a month)”, and “Almost always (several times a week)”.

2.4.2 Data Analyses of PCP Surveys

2.4.2.1 ***Missing Data***

Following survey completion a total of 12 data points were missing from 10 surveyed PCPs. One PCP omitted their sex from the demographics section of the survey. This PCP’s sex was inferred from their name and a photograph of them from their clinic’s website. Two PCPs omitted information from the behaviors/current practices section of the survey. One of these PCPs did not respond to the survey question that asked them if they had ever used colorectal cancer risk assessment tools. The other PCP did not respond to the survey question that asked if they had any experience ordering cancer genetic testing.

The last 7 PCPs with missing data omitted information from the *Cancer and Genetics: Confidence* scale. These PCPs were re-contacted via the e-mail addresses they had provided in the survey and were asked to respond to the missing question(s). PCPs were contacted at one and two weeks following the first e-mail to encourage a higher response rate. One PCP was still unresponsive after these efforts, leaving only 1 missing data point from the *Cancer and Genetics: Confidence* scale. This was from the sixth question in the scale, which is one of the questions that loaded onto the *Addressing questions* domain. Data for this PCP was left out for all analyses that involved the *Addressing questions* domain.

2.4.2.2 ***PCP Survey Data Analyses***

The survey data was cleaned and analyzed using SPSS version 27.0.1.0 for descriptive statistics, univariate, and multivariate binary logistic regression analyses. This was an exploratory analysis, so both univariate and multivariate logistic regression models were run to evaluate the relationship between the predictor variables and high *Genetics and Cancer & Genetics : Confidence* scale scores. A total of 9 predictor variables were used, sex, race/ethnicity, number of years practicing medicine, number of years at current clinic, specialty, degree(s) held, the RUCC of the clinic, *CFIR self-efficacy subscale score*, and the *CFIR knowledge and beliefs subscale* score. The outcome variables, *Addressing questions* domain score and *Explaining cancer risk* domain score, were treated as dichotomous variables that were split at their medians, 2.0 and 1.5, respectively.

First, univariate binary logistic regression analyses were done with each of the nine predictor variables individually to examine the relationship between each predictor variables and the outcomes of interest. Then the multivariate binary logistic regression analyses were done in two steps. First, the seven demographic variables in their preferred forms were entered into the regression simultaneously: 2 category split for number of years at current clinic (≥10 years vs. <10 years), the number of years the PCP has been practicing medicine (continuous), the RUCC of the clinic they work in (continuous), sex (male vs. female), indexed race/ethnicity (White and Hispanic/Latino, White and not Hispanic/Latino, Nonwhite and Hispanic/Latino, Nonwhite and not Hispanic/Latino), specialty (internal medicine vs. family medicine), and degree held (doctor, physician assistant, nursing). Second, all nine predictor variables were entered into the regression equation simultaneously, including the *CIFR self-efficacy subscale* (continuous) and the *CFIR knowledge and beliefs subscale* (continuous). These subscale scores were included to examine what variability they contributed to the outcomes over and above the demographic variables. Significance levels were reported for each predictor variable in all of the univariate and the multivariate models. Statistical significance was defined as $p < 0.05$.

2.5 Qualitative Approach: PCP Interviews

2.5.1 Data Collection of PCP Interviews

At the end of the PCP survey all PCPs were asked if they would be willing to participate in a follow up interview. We planned to interview all PCPs who were willing to participate, hoping 50% would volunteer and offered an extra $50 gift card as an incentive. PCPs who agreed to the follow up interview were asked to input their preferred e-mail address for future contact. EDGE study team members used this information to follow up with all the PCPs that indicated they were willing to do the interview with an introductory message. EDGE team members then sent reminder emails at one and two weeks following the original introductory email to PCPs were who nonresponsive to encourage participation.

EDGE study team members conducted all interviews via Zoom using a semi-structured interview guide to assess PCPs current practices and beliefs about the collection and utilization of personal and familial cancer history information. Questions in the semi-structured interview that addressed current practices and beliefs about the collection of personal and familial cancer history information asked what information sources PCPs gather when assessing a patient for a hereditary cancer risk, how they gather personal and familial cancer history information, and why they think collecting personal and familial cancer history is important or unimportant. Questions that addressed utilization of personal and familial cancer history information included weather or not having a personal or familial history of cancer causes them to change how they care for that patient, and more specifically, if having a personal or familial history of cancer causes them to order a cancer risk genetic test for the patient.

Interviewers were trained in standard interviewing techniques and interviews lasted approximately thirty minutes each. We assigned each interviewee a unique alphanumeric code to ensure confidentiality for the interview participants. All interviews were audio recorded via Zoom, deidentified, then transcribed via an outside transcription company, Rev. Transcriptions were checked for accuracy by a coder before coding began.

2.5.2 Data Analysis of PCP Interviews

All deidentified transcripts were uploaded into Dedoose, a qualitative analysis program, for coding and directed content analysis (Hsieh & Shannon, 2005). A codebook was created that included a priori themes, but also allowed for emerging themes and subthemes to be captured. The codebook was originally based on the CFIR (Damschroder et al., 2009), but it was edited to include anticipated responses to the interview questions since not all the questions in the interview guide were explicitly linked to CFIR constructs. The codebook was further refined as successive transcripts were coded and new codes and themes emerged from the transcripts. New codes were made when the same concept arose in multiple transcripts.

Two research assistants coded the interview transcripts independently. When significant differences arose between the two coders, they met to reconcile those differences, making changes to the codebook when necessary. This coding strategy allowed for a single or multiple codes to be assigned to any portion of text depending on the intricacy of the statement or quotation. Kappa values were calculated at the beginning, middle, and end to ensure appropriate coding of important themes. At these time points initial kappa values were calculated for 2 interview transcripts, then the coders came together to reconcile their differences and final kappa values were calculated. End point kappa values were 0.85 and 0.95. Lastly, I analyzed the coded transcripts to identify sub-themes and themes that addressed the specified research questions.

3. RESULTS

3.1 Quantitative Results : PCP Surveys

3.1.1 Participant Characteristics

60 PCPs completed the survey for an overall response rate of 39.2% (60/153). 25 of the respondents were employed Healthcare system M and 35 were employed by Healthcare System B. Respondents were primarily female (63.3%, n=38), and White, not Hispanic or Latino (81.7%, n=49). Most respondents indicated that they have been practicing medicine for 10 years or less (60%, n=36) and have been at their current clinic for 5 years or less (53.3%, n=32). Most respondents hold an MD (51.7%, n=31) and are family medicine specialists (68.3%, n = 41) (Table 1).

3.1.2 PCPs' Current Behaviors

The majority (78.3%, n=47) of PCPs surveyed reported that they actively collect familial cancer history information from all of their patients, while a minority (20%, n=12) reported only collecting that information for certain patients of theirs, and only one PCP (1.7%) indicated that they do not actively collect familial cancer history information (Table 2). However, most PCPs indicated that they do not use any of the screening tools asked about in the survey. For instance, 80% (n=48) of PCPs indicated they do not use a family tree (pedigree), while 20% of PCPs (n=12) indicated that they only use it for certain patients (Table 2). Regarding the use of the breast cancer referral screening tool (B-RST), again, 80% (n=48) of PCPs indicated that they do not use it while a minority (18.3%, n=11) report using it with only certain patients of theirs, and only one provider (1.7%) indicated that they use it with all of their patients (Table 2). Only 17 PCPs (28.3%) indicated they use any breast cancer risk assessment tools (Table 2). Those PCPs most often reported using the BCRAT/Gail model (n=7), but personal and familial history (n=1) and the Tryer-Cuzick model (n=1) were also mentioned (Table 3). Only 4 PCPs

(6.7%) reported using any colorectal cancer risk assessment tools (Table 2) and one of those PCPs indicated that they use age and family history (Table 3).

There is a lot of variation in the methods that PCPs use to collect familial cancer history information. The most common response was that the information is included in the visit note (48.3%, n=29). Twenty-eight PCPs (46.7%) indicated they use a paper form that is completed by the patient, another 24 PCPs (40%) indicated they use an electronic form completed by the clinician and another 4 PCPs (6.7%) indicated they use an electronic form completed by the patient (Table 4). Other methods for the collection of familial cancer history information include an electronic form filled out by a nurse or medical assistant, that the information is entered directly into the family history in Epic, it is asked about verbally at preventive visits, that the nursing staff complete a form along with the patient, and that the patients are screened during their annual physicals (new patient on paper form then entered into Epic family history) (Table 4).

The vast majority of PCPs (95%, n=57) reported that they consider familial cancer history when making care recommendations (Table 5). The most common answers for how the PCPs use this information is that it causes them to change cancer screening recommendations (n=45) and consider referring the patient for genetic testing and/or genetic counseling (n=11) (Table 6). Other answers included that it causes a PCP to change other treatments or medications the patient is on (n=6), it prompts discussions of other cancer risk factors (n=2), it causes PCPs to refer the patient to another specialty, including oncology (n=2), and it causes PCPs to offer cancer risk genetic testing to the patient (n=1) (Table 6). Most PCPs (93.3%, n=56) indicated that they do talk to patients about cancer genetic testing and most of those PCPs (60.7%, n=34) indicated that they talk about cancer genetic testing occasionally (a couple times a year) (Table 7).

The majority (80%, n=48) of PCPs indicated that they have referred patients for cancer genetic counseling or testing (Table 5). Of the 48 PCPs who indicated that they have referred patients for cancer genetic counseling or testing, 1 PCP (2.1%) indicated that they never refer patients for cancer genetic counseling or testing, 17 (35.4%) indicated that the rarely do (less than once per year), 29 (60.4%) indicated that they occasionally do (a couple times a year), and 1 PCP (2.1%) indicated that they almost always do (several times a week) (Table 7). The PCPs who indicated that they do refer their patients for cancer genetic counseling or testing (n=48) most often indicated that they refer their patients to a genetic counselor (n=24), typically one within their own clinic or health system (n=14) (Table 8). Other places that they refer their patients to include genetics department(s) (n=5), a geneticist (n=5), the oncology department (n=3), another appropriate specialist (n=2), or a referral coordinator (n=1) (Table 8). Two PCPs mentioned that they do in office genetic testing and 1 PCP mentioned that they suggest the patient pursue direct-to-consumer genetic testing. Most PCPs (n=38) indicated that they refer their patients to other providers within their health system, while a minority (n=5) mention that they refer their patients to providers or institutions outside of their health system (Table 8).

Approximately one quarter of the PCPs (27.1%, n=16) surveyed indicated that they have experience ordering cancer risk genetic testing (Table 5). Of the 16 PCPs who indicated that they have experience ordering cancer genetic testing more than half (56.25%, n=9) indicated that they never order cancer genetic testing, a minority (37.5%, n=6) indicated that they rarely (less than once a year) order cancer genetic testing, and a single PCP (6.25%) indicated that they occasionally order cancer genetic testing (a couple times a year) (Table 7).

3.1.2.1 **PCPs' self-efficacy in conducting cancer risk consultation**

The distribution of answers to the first two questions on the *Genetics & Cancer : Confidence* scale follow a similar pattern, where more PCPs rate themselves on the higher end of the confidence scale, suggesting that PCPs have more confidence when it comes to simply initiating the conversation about cancer with patients and recording relevant information on a patient's family history of cancer (Table 9). For the rest of the questions on the scale, we see that more people fall in the middle two categories than anywhere else, suggesting that, on average, PCPs tend to have lower confidence when it comes to doing tasks that require them to respond to questions about cancer risk based on family history or about genetic testing for cancer risk, explaining lifetime cancer risk or age-related cancer risk, and providing support to patients going through the cancer risk assessment and based on family history and genetic testing (Table 9). Further, very few PCPs rated themselves in the "not confident at all" category for any of the questions on the *Cancer & Genetics : Confidence* scale (Table 9).

3.1.2.2 **Comparisons of PCPs' reported confidence among demographic factors**

For the *Addressing questions* domain males tended to score higher than females, white and Hispanic or Latino individuals tended to score higher than other race/ethnicity groupings, PCPs with more than 10 years in practice tended to score higher than those with fewer than 10 years in practice, PCPs with more than 10 years at their clinic tended to score higher than those with fewer than 10 years at their clinic, PAs tended to scored higher than the other degree categories, and PCPs working in urban settings tended to score higher than those working in rural settings (Table 10). However, none of these comparisons came to statistical significance (Table 10). There was no difference in a PCP's *Addressing questions* domain score between family medicine and internal medicine PCPs (Table 10).

For the *Explaining cancer risk* domain, we see that again, males tended to score higher than females, PCPs with more than 10 years at their clinic tended to score higher than those with fewer than 10 years at their clinic, and PCPs working in urban settings tended to score higher than those working in rural settings (Table 10). PCPs that identified as Hispanic or Latino, regardless of their race, tended to score higher than the other race/ethnicity groups, PCPs with less than 10 years in practice tended to score higher on the *Explaining cancer risk* domain than those with greater than or equal to 10 years of practicing medicine, MDs and DOs tended to score the highest among the degree categories, and PCPs who indicated that their primary specialty was internal medicine tended to score higher than family medicine PCPs (Table 11). Again, none of these comparisons were statistically significant (Table 11).

3.1.3 Univariate binary logistic regression results

In the univariate binary logistic regression analyses for the *Addressing questions* domain two predictor variables reached statistical significance. This was the number of years a PCP has been practicing medicine, modeled continuously (p-value = 0.042), and the CFIR self-efficacy subscale, modeled continuously (p-value = 0.009). Additionally, the RUCC of the clinic, modeled continuously, and the number of years the PCP has been working at their current clinic also came close to significance (p-value = 0.052 and p-value=0.064) (Table 12). For the *Explaining cancer risk* domain none of the predictor variables reached statistical significance. However, the PCPs' sex and RUCC of the clinic both came closest to statistical significance (p-value = 0.079 and p-value=0.095) (Table 13).

3.1.4 Multivariate binary logistic regression results

In the first step of the multivariate logistic regression model for the *Addressing questions* domain only the RUCC of the clinic came to statistical significance (p-value = 0.022). The type of degree held by the PCP also came close to significance (p-value = 0.086). Neither the number of years a PCP has been practicing medicine, nor the number of years a PCP has been practicing at their current clinic reached statistical significance in the multivariate model (p-value = 0.302 and p-value =0.182, respectively) (Table 14). When the *CFIR self-efficacy* and *CFIR knowledge and beliefs subscales* were added to the regression model RUCC of the clinic and the CFIR self-efficacy subscale both came to statistical significance (p-value=0.019 and p-value=0.027).

In the multivariate logistic regression model for the *Explaining cancer risk* domain that contained only the seven demographic variables no predictor variables reached statistical significance. The predictor variable that came closest to reaching significance was the PCP's sex (p-value = 0.101). The RUCC of the clinic where the PCP practices (p-value = 0.169) was the next closest to statistical significance (Table 15). The addition of the *CFIR self-efficacy* and

CFIR knowledge and beliefs subscale to the regression model did not change the results significantly. Again, none of the predictor variables came to statistical significance. RUCC was the closest variable to significance (p-value=0.136), followed by the PCPs' sex (p-value=0.151), and the PCPs' *CFIR self-efficacy subscale* score (p-value=0.167)

3.2 Results of PCP Interviews

3.2.1 PCP Interview Demographics

A total of sixteen PCPs completed an interview. Six of the interviewees were employed at Healthcare System M and 10 were employed by Healthcare System B. Interviewees were 50% male (n=8) and 50% female (n=8), primarily working in clinics in urban areas (75%, n=12), and had mostly been employed at their current clinic for 10 or fewer years (68.8%, n=11). A minority of providers (31.25%, n=5) fell in the higher scoring groups for both the *Addressing questions* domain (31.25%, n=5) and the *Explaining cancer risk* domain (31.25%, n=5) (Table 16).

3.2.2 Major Themes from the PCP Interviews

The qualitative data were collected to answer the following questions: 1) What do PCPs bring up in the conversation about family history? 2) Do PCPs believe collecting personal and familial cancer history information is important? 3) Does a personal or familial cancer history cause a PCP to change how they care for their patients? 4) Do patients with a personal or familial cancer history receive cancer risk genetic testing from their PCP?

3.2.2.1 ***What do PCPs bring up in the conversation about family history?***

Most PCPs gather both familial cancer history and personal medical history

The mostly commonly cited information sources PCPs claim to gather is the patient's familial cancer history (n=12), followed by the patient's own medical history (n=10) (Table 17). When asked how she identifies a patient who might need hereditary cancer genetic testing, participant MC-303 said:

> *"Yeah. So, if they are brand new to me as a patient, if they're transferring their care to me or if they're brand new to the clinic, I take the first few minutes to ask questions about their personal health history, surgical history, as well as family history"*
>
> Female, urban, high scoring on *Addressing questions* domain

A common theme among the interviewees was that they are specifically looking for an "unusual" familial and/or personal medical history. PCPs indicated that factors including age of onset, specific cancer diagnoses, degree of the affected relative, presence and patterns of multiple cancers, and the presence of hallmark disorders are particularly important in their clinical assessment for cancer risk. When asked how he currently identifies patients who might need hereditary cancer genetic testing, participant BC-105 explained:

> *BC-105: "Primarily based off of an unusual family history, or just in the course of our discussions something seems to suggest that maybe there could be an underlying genetic problem causing the patients issue."*
>
> *Interviewer: "Right. And what does unusual family history look like to you?"*
>
> *BC-105: "The classic things like breast cancer, ovarian cancer, uterine cancer, on the same family line. Just kind of several, that would be the big one. And let's see, in just other classic conditions, I had a patient with, family history of hypertrophic cardiomyopathy. So just kind of like buzzword disorders."*

Male, urban, low confidence scores both domains

PCPs' methods of collecting personal medical history and familial cancer history information are diverse

Most PCPs indicated that they personally collect familial cancer history information and personal medical history via direct questioning with the patient. Most of these PCPs also report using another method of collection in tandem with the direct questioning process. However, among PCPs who indicated that they collect familial cancer history information via direct questioning each PCP reported a slightly different method of performing this questioning and most reported using another method in tandem with the direct questioning process. For example, Participant MC-303 explained that their clinic has patients fill out family history intake forms, but she still prefers to do the questioning herself. She describes her rather thorough questioning process below:

> *"I generally start off by having the patient tell me if there's any pertinent family history, starting with their parents and then I break off to talking about maternal and paternal relatives. So, I make sure to ask questions about first degree relatives, which I think is probably the most important followed by second degree relatives. Third or fourth degree, I'm not as concerned. And then after I ask specific questions to first degree relatives, then I'll ask more sex-based cancer screening questions such as their family history of breast cancer in a mother or grandmother or male relatives. I ask if there's any history of ovarian or uterine cancer. And then I almost always ask about colon cancer for every individual in my clinic. And then for males, or people who identify as males, I ask about personal family history of prostate cancer, testicular cancer, and*

> *then again, colon cancer, almost always making sure that I hit. And then of course, if there's anything else that pops up as being uncommon, I add that in."*

Female, urban, high scoring on *Addressing questions* domain

Meanwhile, Participant BC-502 explained his less involved procedure:

> *"We usually just ask a general open-ended question. Any family history of illnesses that we need to know about? So, it's pretty general. We don't have a questionnaire or anything."*
>
> *Interviewer: "Yeah. So, you don't specifically ask for cancer but just general illness in the family?"*
>
> *BC-502: "Usually, yep."*

Male, rural, low confidence scores on both domains

PCPs expressed that direct questioning of the patient is their preferred method for collecting personal and/or familial cancer history information. They report that direct questioning process improves the quality of the data obtained compared to other methods and can be easily incorporated into the physical exam. Participant MC-404 explained:

> *"I personally do it through dialogue. My two cents are intake sheets people sometimes flip went through, they don't really answer truthful questions. I get a 40 minutes slot for new patients too. So, I always just think the more I can dive into that personal questions too, to figure out more about it, the better I get a response."*

Male, urban, high score *Explaining cancer risk* domain

Other methods for obtaining their patients' familial cancer history information include obtaining or looking at existing medical records, paper intake forms completed by the patient, putting the information directly into the electronic medical record (EMR), having another staff member collect it before they meet with the patient, or some combination of those methods. Participant BC-107 explains:

> "*Yeah, in our specific clinic, there are intake staff, whether it's the nursing staff or MA. A lot of times, they will ask family history ahead of time. And then I always try to reconfirm that in my interview with the patient as well.*"
>
> Male, urban, high score *Explaining cancer risk* domain

A few interviewees even indicated that these collection methods can change, both over time and from patient to patient. Participant MC-405 stated:

> *"It changes all the time unfortunately. Previously when I was establishing my practice, more of my patients were given paper forms that they could fill out prior to the visit. Currently that seems to have stopped for whatever reason, I think with COVID they don't want to be handing out clipboards, and papers, and pens. Currently it's just an oral history that I ask in the exam."*
>
> Female, urban, high score *Addressing questions* domain

3.2.2.2 ***Do PCPs believe collecting personal and familial cancer history information is important?***

Most PCPs believe collecting personal and familial cancer history information is very important

PCPs were nearly unanimous in expressing that collecting personal and familial cancer history information is very important. The most commonly cited reasons that this information is so important to collect is for disease prevention opportunities and proper long-term care for the patient (Table 17). Participant MC-405 said:

> *"For me personally, I think it's very important… I'm concerned about my patients' risk for colon cancer, breast cancer, ovarian cancer, skin cancers; cancer where I can actively do some preventive health maintenance and order screening."*
>
> Female, urban, high score *Addressing questions* domain

Few PCPs believe collecting personal and familial cancer history information is not so important

On the flip side, a small subset (n=2) of PCPs indicated that collecting personal and familial cancer history information is only of medium importance. Some common ideas among the participants who expressed this type of opinion was a lack of time with the patient, conflicting priorities, and a lack of information about how to proceed with patients who would be identified as high-risk. Participant BC-502 explained:

> *"I would have to say currently in our practice, it's probably medium importance. It's not like we put a high priority on it. Usually if someone's coming in, we want to address the problem that they're coming in with. At their annual examination we try to go over those things…."*
>
> Male, rural, low confidence scores both domains

Interestingly, one participant expressed that while he doesn't currently prioritize the collection of personal and familial cancer history information, he does see that genetic testing is becoming more integrated into primary care settings and expects that these things will become more relevant over time. Participant BC-502 said:

> *“I believe that [collecting a patient’s personal and familial history of cancer] probably is fairly important, particularly going forward into the future as we get a better understanding of these tests. Right now, however, I'm not doing a whole lot of effort to test people. Most of the patients that I have that are tested have been done by specialists, so I don't know. I mean, I think it's important, but I'm not right now, putting forth a great effort in my own patients to screen”*
>
> Male, rural, low confidence scores both domains

3.2.2.3 ***Does a personal or familial cancer history cause a PCP to change how they care for their patients?***

Changes screening recommendations

The interviewed PCPs unanimously report that having a personal or familial cancer history changes how they care for their patients (n=16) (Table 17). The most commonly cited change in practice is that it changes their future cancer screening recommendations for the patient (n=13) including changes in which screening tools they use, the screening intervals they set for the patient, and the age at which they would have them start screening. Participant MC-102 said:

> *“Yeah. Well, I think obviously it depends on the specific diagnosis. In general, I would say we definitely… like let's just say mammograms, for example, if someone has a family history of breast cancer, I encourage them to get a mammogram every year, as opposed to every two years.”*
>
> Female, urban, low confidence scores both domains

Referrals to genetics specialists

The second most commonly cited practice change is PCPs consider, discuss and potentially refer the patient onto a genetics specialist of some kind (n=7) (Table 17). Genetic specialists that PCPs refer their patients to were most commonly genetic counselors (n=5), but PCPs also cited referring patients to oncology departments, cancer clinics/centers, geneticists, and other appropriate specialties. Participant BC-303 explains:

> *"Screening intervals, age at which we would start cancer screening tools, and then whether they need more extensive genetic counseling or referrals. Sometimes if they have a strong family history of cancer, either in general or specific cancers, I'll just oftentimes refer them to genetic counseling or the high-risk breast cancer clinic for further evaluation."*
>
> Female, urban, low confidence scores both domains

Interestingly, one participant even reported that they suspect they miss appropriate opportunities to refer their patients to genetic specialists on a regular basis. Participant BC-108 said:

> *"I suspect I miss opportunities to refer genetic testing all the time and that's an area I'm interested in. It's just so many other competing things. This is one of our greatest challenges. If somebody's having a heart attack right in front of you or falling off the cliff, we're really good at taking care of them. Looking down the line and saying, "Well, we need to treat your high cholesterol for the next 20 years," It's harder for us … with this genetic testing."*
>
> Female, urban, high confidence scores both domains

3.3.3.4 ***Do patients with a personal or familial cancer history receive cancer risk genetic testing from their PCP?***

Most PCPs are not routinely ordering cancer risk genetic testing

Most interviewees report that they never order cancer risk genetic testing (n=11) (Table 17) and cite a variety of barriers to ordering cancer risk genetic testing. The most common theme among the issues cited by the interviewed PCPs were provider-centered concerns about a lack of knowledge and confidence regarding cancer risk genetic testing, both abstractly and logistically. Specific barriers cited by the interviewees include a lack of knowledge about which test(s) to order, how to order the test(s), how to interpret the results, how to relay that information to their patients, and how to get the testing covered by insurance. Participant MC-402 said:

> *“So, I don't [order genetic testing for a patient with a family history of cancer] and that's only because I don't feel like I am not educated enough to know which ones to order, because it's obviously more than just the BRCA. And so that's why I don't. And I also don't know what my current health care system has, like in place for testing, like which labs they use, you know, out of pocket costs, versus insurance coverage, things like that, which is why I usually refer to genetic counseling, because they just know more of the details, but it's certainly something I would do if I felt like I had the resources and the knowledge to do it, if that makes sense.”*
>
> Female, rural, low confidence scores both domains

Another major theme cited by the interviewed PCPs were patient-centered concerns. This theme encompassed issues including, high out-of-pocket costs to the patient, potential issues with insurance coverage for the patient in the future, whether or not the patient wants to know in the first place, and a patient’s willingness to actually act on the result. Participant MC-303 said:

> *“… Let's say it's a cancer screening, right, that would be a genetic screening. What if something comes up as remarkable or concerning or high risk for a certain condition and I can't get ahold of that patient and I'm responsible for that follow through? I need to know if they have stable housing or a reliable address. I need to know if they have a means of being able to be contacted. And then I think the other barrier is, when I talk to patients about the benefits and the risks of screening, if I screened for something, are you going to follow through on working that up, right, because if it doesn't change what you're willing to pursue, if you foresee more risks to doing the screening, then why should I offer it, right, does that make sense? “*
>
> Female, urban, High *Addressing questions* domain

Few PCPs are comfortable ordering a limited range of cancer genetic tests

A minority of PCPs report that they have ordered cancer risk genetic testing at least once during their practice lifetime (n=5). A common theme among these PCPs is that that they really only feel comfortable ordering very well-known cancer genes, like BRCA1/2 (n=3) or other tests specifically recommended by genetic counselors or genetics teams. Participant BC-502 said:

> *“I would send them to the genetic counselor and then they would make recommendations as far as genetic tests to order and then I will order it based upon their recommendation. I just don't know enough about genetics and genetic ties to specific cancers for me to feel comfortable ordering those tests by myself without some sort of input.*”
>
> Male, rural, low confidence scores both domains

4. DISCUSSION

4.1 Beliefs

4.1.1 Beliefs regarding the importance of collecting personal medical history and familial cancer history

The vast majority of PCPs interviewed believe collecting personal medical history and familial cancer history information is very important particularly for the long-term care of their patients. These views are consistent with research that indicates most providers are receptive to using family history for cancer risk assessment and have positive ideas surrounding it (Flynn et al., 2010). This is important because collection of a family cancer history for cancer risk assessment is recommended by numerous professional organizations as previously discussed (Moyer & USPSTF, 2014, Hampel, Bennett, Buchanan, Pearlman, & Wiesner, 2014, EGAPP Working Group, 2009).

In contrast, we also identified a small number of PCPs who reported that collection of familial cancer history just isn’t always a priority in the primary care setting. This is consistent with previous research that found PCPs frequently have to defer preventive health for pragmatic reasons including, managing multiple acute concerns in complex patients and a lack of time with the patients for a variety of reasons (Murugan et al., 2018). This will likely continue to be true so long as the fundamental nature of the primary care profession remains unchanged. There will never be enough time with every patient to address every need. Even among PCPs who indicated they do not believe collecting familial cancer history information is very important right now, one PCP expressed that they believe it will become more important in the future as genetic medicine further penetrates into primary care practice.

4.2 Practices

4.2.1 Collection of personal and familial cancer history information

Both the survey and the interview data revealed that the majority of PCPs collect familial cancer history information from their patients to assess if a patient needs hereditary cancer genetic testing. The survey results indicated that more than 70% of PCPs do not use any cancer risk assessment tools including pedigrees, breast cancer referral screening tools, breast cancer risk assessment tools, or colorectal cancer risk assessment tools. This aligns with the interview data, where most PCPs expressed that they prefer to collect familial cancer history information via direct questioning with the patient in tandem with other collection methods. It is important to note that among the interviewees who indicated they use the direct questioning method, each had their own version and ordering of questions they would ask, and some were considerably more thorough than others. This is consistent with research that shows family history collection among PCPs is quite varied, frequently incomplete to truly detect hereditary cancer risk, and is infrequently updated (Flynn et al., 2010).

4.2.2 Use of personal and familial cancer history information

The interviewees nearly unanimously agreed that collecting personal and familial cancer history information is very important. Both the interview and survey data indicated that PCPs most commonly use this information to change their cancer screening recommendations to allow for earlier, more frequent, and more sensitive cancer screening. The second most commonly cited practice change for these PCPs was that a family history of cancer causes them to consider/refer their patient to a genetics specialist.

In the survey there is a pretty drastic difference between the number of providers who indicate that they change their screening recommendations (n=45) compared to the number of providers who indicated that it makes them consider referring their patient for genetic testing/counseling (n=11). One could conclude that this suggests that PCPs recognize the increased risk their patient is now at due to their positive family history, but they’re not confident enough in the genetic testing process to actually refer those patients onto genetics professionals.

4.2.3 Referrals for genetic testing

Despite the findings in the previous paragraph, the survey results indicate that 80% of PCPs have indeed referred patients for cancer genetic counseling during their time in practice (Table 5). Both the interview and survey data indicate that the PCPs most frequently refer their patients to a genetic counselor and most PCPs indicated that they like to refer their patients to providers within their own health system. This is consistent with literature that indicates that 88% of internal medicine providers feel comfortable referring patients to genetic counselors to assess disease risk (Evenson et al., 2016).

4.2.4 Performing cancer risk genetic testing

Both the survey and interview data indicate that most PCPs are not performing cancer risk genetic testing for their patients, even when it might be appropriate. PCPs cited an extensive list of reasons for not performing cancer risk genetic testing that center on their own lack of knowledge about genetic testing, but also citing issues related to their patient’s decisions surrounding genetic testing. Many of these concerns, particularly those related to the low knowledge levels expressed by PCPs are corroborated in the systematic review performed in 2016 (Hamilton et al.). Overall, our data indicates that PCPs still prefer to simply refer their high-risk patients to genetic specialists, particularly genetic counselors. This is consistent with other research where providers have expressed the opinion that medical geneticists, genetic counselors, and oncologists are the most well qualified to provide cancer genetic testing and counseling services and that they would not have enough time or resources to even incorporate BRCA1/2 testing for female patients over the age of 30 years old (Dekanek et al., 2019).

Unfortunately, genetic counselors are in short supply across the nation, so this is not a sustainable practice (Abaca et al., 2018). This underscores the need to develop appropriate genetics educational opportunities (Freedman et al., 2003) along with other tools and procedures to assist PCPs, specifically by giving them explicit direction on how to handle patients who are identified as high-risk by these screening methods.

4.3 Confidence

4.3.1 PCPs confidence in conducting clinical cancer risk consultation

Surprisingly, very few PCPs rated themselves in the “not confident at all” category for any of the questions on the *Cancer & Genetics : Confidence* scale (Table 2). This is inconsistent with the literature that indicates PCP’s objective and subjective genetics related knowledge scores are typically very poor (Hamilton, 2016). A wide variety of factors could contribute to PCPs having more confidence in various aspects of a clinical cancer consultation, such as increased coverage of genetics topics in medical schools or residency programs as the genetics and genomic fields are maturing, or the fact that more time has passed and providers have had more time to really understand, digest, and practice using the national recommendations.

4.3.2 Quantitative factors associated with confidence domains or lack thereof

4.3.2.1 ***Differences between univariate and multivariate confidence analyses***

For the *Addressing questions* domain, RUCC of clinic reached statistical significance in the multivariate model but didn’t change drastically from the univariate model. Meanwhile, the number of years practicing medicine was significant in the univariate model, but this significance was lost in the multivariate setting. The number of years the PCP had been practicing at their clinic became less significant in the multivariate model. The p-values for the most important variables in the univariate setting for the *Explaining cancer risk* domain*,* sex, RUCC, and the CFIR self-efficacy subscale score, remained fairly similar in the univariate and multivariate analyses, although all variables did become less significant in the multivariate models. This suggests that other variables entered into the multivariate analysis accounted for some of the variance that had previously been attributed to them in the univariate setting.

4.3.2.2 ***RUCC***

The RUCC of the clinic stayed close to statistical significance in the univariate and multivariate settings for both domains, *Addressing questions* and *Explaining cancer risk.* Importantly, all the RUCC estimates are positive, indicating that providers that practiced in rural settings consistently had lower confidence both in their ability to addresses questions about cancer risk genetic testing and their ability to explain cancer risk to their patients.

This is consistent with findings from Harding and colleagues who conducted focus group interviews and found that rural PCPs report a variety of barriers to genetic testing including, less access to continuing education regarding genetics and genetic testing, less follow up communications from specialists, and informal support from colleagues with genetics experience, all of which could easily lead to PCPs having lower confidence levels in these topic areas (2018).

4.3.2.3 ***Length of time at clinic***

The length of time a PCP has been at their clinic was only close to significance in the univariate model for the *Addressing questions* domain. Interestingly, the beta estimate for the *Addressing questions* domain for the length of time at their clinic variable was positive in the univariate setting, but negative in the multivariate setting. This indicates that a longer time at the clinic was associated with higher confidence scores in the univariate setting, but the longer time at the clinic was associated with lower confidence scores in the multivariate setting. This suggests that one or more of the variables that were added into the multivariate model had some kind of interactive effect on these confidence scores.

4.3.2.4 ***Sex***

Sex was not significant in the univariate or multivariate models for the *Addressing questions domain*. However, sex did come close to significance in both the univariate and multivariate models for the *Explaining cancer risk* domain. The estimate for the sex variable in the *Explaining cancer risk* domain was positive in both the univariate and the multivariate models, indicating that females were more confident than males in this domain. The idea that a PCP's sex would influence their ability to conduct a cancer risk consultation was unexpected and in this specific context does not appear to be reported on in the literature. It is important to note that the survey measure was evaluating confidence in these specific abilities and not the ability itself.

4.3.2.5 ***CFIR Subscales***

The *CFIR self-efficacy subscale* reached statistical significance in both the univariate and multivariate regression models for the *Addressing questions* domain and stayed close to significance in both models for the *Explaining cancer risk* domain. These estimates indicate that scoring higher on this subscale is positively associated with being in the higher scoring group, particularly for the *Addressing questions* domain. On the other hand, the *CFIR knowledge and beliefs subscale* did not come close to significance in either domain. This highlights the disconnect between what PCPs know on an abstract level and what they are actually comfortable doing in their own practice.

4.4 Limitations

Limitations of this study include the cross-sectional design of the study and the relatively small sample size obtained for the survey. This small sample didn't give us much power to detect statistical differences between the different groups. In addition, the response rate for the survey was low at 39.2%, but consistent with response rates for other surveys regarding cancer genetics that target medical providers (Dekanek et al., 2019 & Cohn et al., 2015). Further, the sample only included PCPs from Washington, Wyoming, and Montana, was majority white and not Hispanic or Latino, and is therefore unlikely to be representative of all PCPs in the US. Self-report surveys can also be problematic as they are subject to participant's internal biases about their work. Finally, since PCPs had to volunteer for the interview following the survey, they may have been self-selecting in some fashion. The providers who participated in the interview may have had stronger feelings about the EDGE study and wanted to voice them.

5. CONCLUSIONS

Despite the many cancer risk assessment tools designed for use in clinical care, most PCPs in our study report gathering personal and familial cancer history from their patients via a general direct questioning process with the patient. Most PCPs believe that collecting this information is important because it informs their future cancer screening recommendations and a smaller portion report that it makes them consider referring the patient to a genetic specialist of some kind. Most PCPs are not ordering cancer risk genetic testing and those that are report they are really only comfortable ordering a limited range of cancer genetic tests. PCPs who practice in more urban areas and who score higher on the *CFIR self-efficacy subscale* tended to have higher confidence levels with respect to initiating the conversation about cancer risk and responding their patients’ questions about cancer risk based on their family history as well as their confidence in explaining lifetime and age-related cancer risk to their patients.

It is clear that PCPs are not using the family history collection tools that have been designed for assessing hereditary cancer risk and they are not ordering cancer risk genetic testing. We must find creative ways to make these tools and ordering processes more up-to-date, accessible, useful, and intuitive for use in everyday clinical practice. Future strategies for implementing genomic medicine in primary care settings should take note of these results and be prepared to meet PCPs where they are to provide them with tailored resources to help these hereditary cancer screening and genetic testing strategies realize their true population-level potential.

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Appendix

Table 1: Participant Characteristics (n=60)

Characteristic	n (%)
Sex	
Male	22 (36.7%)
Female	38 (63.3%)
Race/Ethnicity	
White, Not Hispanic or Latino	49 (81.7%)
Nonwhite, Not Hispanic or Latino	8 (13.3%)
White, Hispanic or Latino	2 (3.3%)
Nonwhite, Hispanic or Latino	1 (1.7%)
Years in Practice	
< 1 year	3 (5%)
1-5 years	18 (30%)
6-10 years	15 (25%)
11-15 years	9 (15%)
16-20 years	5 (8.3%)
21-25 years	7 (11.7%)
26-30 years	2 (3.3%)
>30 years	1 (1.7%)
Length of time at clinic	
<1 year	6 (10%)
1 year	4 (6.7%)
2 years	6 (10%)
3 years	11 (18.3%)
4 years	2 (3.3%)
5 years	3 (5%)
6 years	5 (8.3%)
7 years	1 (1.7%)
8 years	2 (3.3%)
9 years	0 (0%)
10 years	4 (6.7%)
>10 years	16 (26.7%)
Degree(s) Held*	
MD	31 (51.7%)
DO	9 (15%)
PA-C	5 (8.3%)

ARNP/APRN	16 (26.7%)
PhD	1 (1.7%)
MPH	3 (5%)
MBA	0 (0%)
Other ("DNP")	1 (1.7%)
Primary Practice Specialty	
Family Medicine	41 (68.3%)
Internal Medicine	19 (31.7%)
Other	0 (0%)
Clinic Setting - Rural-urban Continuum Codes (RUCC)	
1	25 (41.7%)
2	0 (0%)
3	23 (38.3%)
4	0 (0%)
5	0 (0%)
6	0 (0%)
7	12 (20%)
8	0 (0%)
9	0 (0%)

* = Because 1 individual can hold more than 1 degree percentages may not add up to 100%

Table 2: PCPs' self-reported screening practices & tools

Screening Tool	Do not use, n(%)	Use with all patients, n(%)	Use only with certain patients, n(%)
Active collection of familial cancer history (n=60)	1 (1.7%)	47 (78.3%)	12 (20%)
Family tree (pedigree) (n=60)	48 (80%)	-	12 (20%)
Breast cancer referral screening tool (B-RST) (n=60)	48 (80%)	1 (1.7%)	11 (18.3%)
Breast cancer risk assessment tools (BCRAT/Gail model; Claus model;Tryer-Cuzick/IBIS model; BOADICEA;BRCAPRO) (n=60)	43 (71.7%)	2 (3.3%)	15 (25%)
Colorectal cancer risk assessment tools (PREMM; MMRpredict;MMRpro) (n=59)	55 (91.7%)	2 (3.3%)	2 (3.3%)
Other Yes = 35 (58.3%), No = 25 (41.7%)			

Table 3: Risk assessment tools specified by PCPs		
Breast cancer risk assessment tools		n
	BCRAT/Gail model	7
	Age and personal and familial history	1
	Tryer-Cuzick	1
Colorectal cancer risk assessment tools		
	Age and family history	1

Table 4 : PCPs' Self-reported familial cancer history collection methods (n=57)	
Method	**n (%)**
Information is included in visit note	29 (48.3%)
Paper form completed by patient	28 (46.7%)
Electronic form completed by clinician	24 (40%)
Electronic form completed by patient	4 (6.7%)
Other methods entered: electronic form filled by nurse or MA, entered in family history in Epic, asked verbally at preventive visits, nursing staff completes form with patient, screened during annual physical (new patient on paper form then entered into Epic family history)	

*Total may add up to more than 100% due to the "select all that apply" nature of the question

Table 5: PCPs' self-reported cancer genetic testing related behaviors

Behavior	Yes n(%)	No n(%)
Considering of familial cancer history when making care recommendations (n=60)	57 (95%)	3 (5%)
Has ever referred patients for cancer genetic counseling or testing (n=60)	48 (80%)	12 (20%)
Ordering cancer genetic testing (n=59)	16 (27.1%)	43 (72.9%)

Table 6: Use of familial cancer history for care recommendations (n=57)	
Use	**n***
PCP changes cancer screening recommendations	45
PCP considers referral for genetic counseling and/or testing	11
PCP may change other treatments or medications	6
Prompts discussion of other cancer risk factors	2
PCP refers patient to other specialty	2
PCP provides cancer risk genetic testing	1

* May add up to more than 57 due to free response nature of the question

Table 7: PCPS' self-reported frequency of cancer genetic testing related behaviors

Behavior	Frequency of Behavior				
	Never	**Rarely** (less than once a year)	**Occasionally** (a couple of times a year)	**Often** (a couple times a month)	**Almost always** (several times a week)
Talking to patients about cancer genetic testing (n=60)	4 (6.7%)	16 (26.7%)	34 (56.7%)	5 (8.3%)	1 (1.7%)
Referring patients for cancer genetic counseling or testing (n=48)	1 (2.1%)	17 (35.4%)	29 (60.4%)	-	1 (2.1%)
Ordering cancer genetic testing (n=16)	9 (56.25%)	6 (37.5%)	1 (6.25%)	-	-

Table 8: Where PCPs refer their patients for cancer genetic counseling or testing

Place patient is referred to	n
Genetic counselor	24
Genetics department	5
Geneticist	5
Oncology department	3
Other appropriate specialist	2
Referral coordinator	1
In office genetic testing	2
Direct to consumer genetic testing	1
Within system	28
Outside system	5

* Numbers will add up to more than 60 because free text responses allow PCPs to express multiple ideas in their response

Table 9: PCPs' self-efficacy in conducting a cancer risk consultation

	Not confident at all N (%)	Not very confident N (%)	Somewhat confident N (%)	Very confident N (%)
Initiate talking to patients about cancer (n=60)	0 (0%)	1 (1.7%)	21 (35%)	38 (63.3%)
Record relevant information on a patient's family history of cancer. (n=60)	0 (0%)	1 (1.7%)	27 (45%)	32 (53.3%)
Respond to patients' questions about cancer risk based on family history. (n=60)	1 (1.7%)	16 (26.7%)	34 (56.7%)	9 (15%)
Respond to patients' questions about genetic testing for cancer risk. (n=60)	11 (18.3%)	30 (50%)	17 (28.3%)	2 (3.3%)
Explain lifetime cancer risk to patients. (n=60)	8 (13.3%)	32 (53.3%)	14 (23.3%)	6 (10%)
Explain age-related cancer risk to patients. (n=59)	3 (5.1%)	14 (23.7%)	32 (54.2%)	10 (16.9%)
Provide support to patients going through cancer risk assessment based on family history and genetic testing. (n=60)	4 (6.7%)	28 (46.7%)	20 (33.3%)	8 (13.3%)

Table 10: Average *Addressing questions* domain scores by demographic factors

Characteristic	Average score (SD) (n=60)
Sex	
Male (n=22)	1.93 (.50)
Female (n=38)	1.87 (.54)
Indexed Race and Ethnicity	
White, Not Hispanic or Latino (n=49)	1.86 (.49)
Nonwhite, Not Hispanic or Latino (n = 8)	2.00 (.65)
White, Hispanic or Latino (n =2)	2.25 (1.06)
Nonwhite, Hispanic or Latino (n=1)	2.00 (N/A)
Years in Practice	
<= 10 years (n=36)	1.84 (.49)
> 10 years (n=24)	1.96 (.57)
Length of time at clinic	
<= 10 years (n=40)	1.81 (.54)
>10 years (n=38)	2.05 (.46)
Degree(s) Held	
MD/DO (n=40)	1.98 (.49)
PA-C (n=4)	2.00 (.58)
ARNP/APRN/DNP (n=16)	1.89 (.52)
Primary Practice Specialty	
Family Medicine (n = 41)	1.89 (.53)
Internal Medicine (n=19)	1.89 (.52)
Clinic Setting	
Urban (n = 48)	1.95 (.52)
Rural (n=12)	1.67 (.49)

Table 11: Average *Explaining cancer risk* domain score by demographic factors

Characteristic	Average score (SD) (n=59)
Sex	
Male (n=22)	1.75 (.75)
Female (n=37)	1.45 (.73)
Indexed Race and Ethnicity	
White, Not Hispanic or Latino (n=48)	1.49 (.75)
Nonwhite, Not Hispanic or Latino (n=8)	1.88 (.79)
White, Hispanic or Latino (n=2)	2.00 (.71)
Nonwhite, Hispanic or Latino (n=1)	2.00 (N/A)
Years in Practice	
<= 10 years (n=35)	1.60 (.77)
> 10 years (n=24)	1.52 (.74)
Length of time at clinic	
<= 10 years (n=39)	1.54 (.82)
>10 years (n=38)	1.63 (.60)
Degree(s) Held	
MD/DO (n=39)	1.69 (.67)
PA-C (n=4)	1.63 (1.03)
ARNP/APRN/DNP (n=16)	1.57 (.84)
Primary Practice Specialty	
Family Medicine (n =40)	1.46 (.73)
Internal Medicine (n=19)	1.79 (.77)
Clinic Setting	
Urban (n=47)	1.61 (.76)
Rural (n=12)	1.42 (.73)

Table 12 : Univariate regression model findings for the prediction of membership to the high scoring *Addressing questions* group

Variable	β	SE	Wald	df	Sig.
Sex	0.189	0.612	0.096	1	0.757
Number of years at current clinic	1.145	0.618	3.437	1	0.064
Specialty	0.318	0.664	0.23	1	0.632
Practice years	0.368	0.181	4.12	1	0.042
RUCC	-0.385	0.198	3.771	1	0.052
Degree held (Reference) (MD/DO)	-				
(PA) 1	0.368	0.0737	0.249	1	0.618
(APRN/ARNP/DNP) 2	1.466	1.188	1.525	1	0.217
Race/Ethnicity (Reference) (White, Not Hispanic/Latino)			1.403	3	0.705
(Nonwhite, Not Hispanic/Latino) 1	19.963	40192.818	0	1	1
(White, Hispanic/Latino) 2	20.692	40192.818	0	1	1
(Nonwhite, Not Hispanic/Latino) 3	21.203	40192.818	0	1	1
CFIR self-efficacy subscale	1.283	0.492	6.789	1	0.009
CFIR knowledge and beliefs subscale	0.567	0.731	0.603	1	0.438

Table 13 : Univariate regression model findings for the prediction of membership to the high scoring *Explaining cancer risk* group

Variable	β	SE	Wald	df	Sig.
Sex	0.993	0.565	3.094	1	0.079
Number of years at current clinic	-0.61	0.568	1.155	1	0.282
Specialty	-0.742	0.575	1.667	1	0.197
Practice years	0.175	0.162	1.159	1	0.282
RUCC	-0.24	0.143	2.789	1	0.095
Degree held (Reference, MD/DO)	-	-	1.452	2	0.484
(PA) 1	0.736	0.663	1.232	1	0.267
(APRN/ARNP/DNP) 2	0	1.291	0	1	1
Race/Ethnicity (Reference, White, Not Hispanic/Latino)	-	-	1.251	3	0.741
(Nonwhite, Not Hispanic/Latino) 1	-21.991	40192.991	0	1	1
(White, Hispanic/Latino) 2	-21.203	40192.991	0	1	1
(Nonwhite, Not Hispanic/Latino) 3	-21.203	40192.991	0	1	1
CFIR self-efficacy subscale	0.538	0.351	2.353	1	0.125
CFIR knowledge and beliefs subscale	-0.207	0.654	0.1	1	0.751

Table 14: Multivariate logistic regression model findings for the prediction of membership to the high scoring *Addressing questions* group

Variable	Step 1					Step 2		
	β	SE	Wald	df	Sig.	β	SE	Sig.
Sex	0.323	0.847	0.145	1	0.703	0.295	1.14	0.796
Number of years at current clinic	-1.347	1.009	1.782	1	0.182	-1.593	1.271	0.21
Specialty	1.243	0.953	1.703	1	0.192	1.178	1.087	0.278
Practice years	0.276	0.267	1.066	1	0.302	0.34	0.353	0.335
RUCC	-0.942	0.411	5.259	1	0.022	-1.159	0.496	0.019
Degree held (Reference) (MD/DO)	-	-	4.911	2	0.086	-	-	0.115
(PA) 1	-0.17	0.996	0	1	0.986	-0.898	1.152	0.436
(APRN/ARNP/DNP) 2	3.857	1.958	3.879	1	0.049	3.916	2.383	0.1
Race/Ethnicity (Reference, White, Not Hispanic/Latino)	-	-	2.287	3	0.515	-	-	0.529
(Nonwhite, Not Hispanic/Latino) 1	17.14	40192.966	0	1	1	16.577	40192.873	1
(White, Hispanic/Latino) 2	18.397	40192.966	0	1	1	17.176	40192.873	1
(Nonwhite, Not Hispanic/Latino) 3	19.143	40192.966	0	1	1	19.841	40192.873	1
CFIR self-efficacy subscale	-	-		-	-	1.89	0.857	0.027
CFIR knowledge and beliefs subscale	-	-	-	-	-	-0.176	1.276	0.89

Table 15: Multivariate logistic regression model findings for the prediction of membership to the high scoring *Explaining cancer risk* group

Variable	Step 1					Step 2		
	β	**SE**	**Wald**	**df**	**Sig.**	**β**	**SE**	**Sig.**
Sex	1.151	0.702	2.689	1	0.101	1.065	0.741	0.151
Number of years at current clinic	-0.734	0.87	0.711	1	0.399	-0.583	0.89	0.513
Specialty	-0.593	0.719	0.681	1	0.409	-0.535	0.747	0.474
Practice years	0.123	0.235	0.274	1	0.6	0.153	0.244	0.532
RUCC	-0.245	0.178	1.891	1	0.169	-0.28	0.188	0.136
Degree held (Reference) (MD/DO)	-	-	0.05	2	0.975	-	-	0.926
(PA) 1	-0.158	0.8	0.039	1	0.843	-0.1	0.86	0.907
(APRN/ARNP/DNP) 2	-0.273	1.578	0.03	1	0.863	-0.616	1.575	0.696
Race/Ethnicity (Reference, White, Not Hispanic/Latino)	-	-	2.495	3	0.476	-	-	0.441
(Nonwhite, Not Hispanic/Latino) 1	-21.519	40192.955	0	1	1	-21.605	40192.837	1
(White, Hispanic/Latino) 2		40192.955	0	1	1	-20.799	40192.837	1
(Nonwhite, Not Hispanic/Latino) 3	-19.463	40192.955	0	1	1	-19.193	40192.837	1
CFIR self-efficacy subscale	-	-	-	-	-	0.567	0.41	0.167
CFIR knowledge and beliefs subscale	-	-	-	-	-	-1.024	0.959	0.285

Table 16: Interviewee Characteristics (n=16)

Characteristic	N(%)
Sex	
Male	8 (50%)
Female	8 (50%)
Indexed Race and Ethnicity	
White, Not Hispanic or Latino	13 (81.3%)
Nonwhite, Not Hispanic or Latino	3 (18.8%)
Years in Practice	
<= 10 years	10 (62.5%)
> 10 years	6 (37.5%)
Length of time at clinic	
<= 10 years	11 (68.8%)
>10 years	5 (31.3%)
Degree(s) Held	
MD/DO	13 (81.3%)
ARNP/APRN/DNP	3 (18.8%)
Primary Practice Specialty	
Family Medicine	12 (75%)
Internal Medicine	4 (25%)
Clinic Setting	
Urban	12 (75%)
Rural	4 (25%)
***Addressing questions* domain score group**	
High	5 (31.3%)
Low	11 (68.8%)
***Explaining cancer risk* domain score group**	
High	5 (31.3%)
Low	11 (68.8%)

Table 17 : Qualitative research question results and associated themes (n= 16)

Research Question	Responses	Frequency (n)
1. What do PCPs bring up in the conversation about family history?	Familial cancer history	12
	Personal medical history	10
Themes identified	*Unusual family/personal history*	*9*
	Collected via direct questioning by PCP	*12*
2. Do PCPs believe collecting personal or familial cancer history information is important?	Very important	14
	Somewhat important	2
Theme identified	*Longitudinal care/Prevention Opportunities*	*12*
3. Does a personal or familial cancer history cause a PCP to change how they care for a patient?	Yes	16
	No	0
Themes identified	*Changes cancer screening recommendations*	*13*
	Referral to genetic specialists	*7*
4. Do patients with a personal or familial cancer history receive cancer risk genetic testing from their PCP?	Never	11
	Sometimes	5
Themes identified	*Provider centered concerns*	*13*
	Patient centered concerns	*7*
	Order super well known tests (like BRCA)	*3*