REVIEW

Primary care providers' cancer genetic testing-related knowledge, attitudes, and communication behaviors: A systematic review and research agenda

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BACKGROUND: Primary care providers (PCPs) can play a critical role in helping patients receive the preventive health benefits of cancer genetic risk information. Thus, the objective of this systematic review was to identify studies of US PCPs' knowledge, attitudes, and communication-related behaviors regarding genetic tests that could inform risk-stratification approaches for breast, colorectal, and prostate cancer screening in order to describe current findings and research gaps.

METHODS: We conducted a systematic search of six electronic databases to identify peer-reviewed empirical articles relating to US PCPs and genetic testing for breast, colorectal, or prostate cancer published in English from 2008 to 2016. We reviewed these data and used narrative synthesis methods to integrate findings into a descriptive summary and identify research needs.

RESULTS: We identified 27 relevant articles. Most focused on genetic testing for breast cancer (23/27) and colorectal cancer risk (12/27); only one study examined testing for prostate cancer risk. Most articles addressed descriptive research questions (24/27). Many studies (24/27) documented PCPs' knowledge, often concluding that providers' knowledge was incomplete. Studies commonly (11/27) examined PCPs' attitudes. Across studies, PCPs expressed some concerns about ethical, legal, and social implications of testing. Attitudes about the utility of clinical genetic testing, including for targeted cancer screening, were generally favorable; PCPs were more skeptical of direct-to-consumer testing. Relatively fewer studies (9/27) examined PCPs' communication practices regarding cancer genetic testing.

DISCUSSION: This review indicates a need for investigators to move beyond descriptive research questions related to PCPs' knowledge and attitudes about cancer genetic testing. Research is needed to address important gaps regarding the development, testing, and implementation of innovative interventions and educational programs that can improve PCPs' genetic testing knowledge, assuage concerns about the appropriateness of cancer genetic testing, and promote open and effective patient-provider communication about genetic risk and genetic testing.

KEY WORDS: primary care; knowledge; health beliefs; communication; cancer; genetic testing.

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INTRODUCTION

Genetic testing for individuals at hereditary risk for mutations in known cancer susceptibility genes is an important component of preventive medicine.[1,2] Recommendations exist for the use of genetic testing to identify pathogenic variants in high-penetrance genes including *BRCA1/2*, associated with hereditary breast and ovarian cancer,[3–5] and mismatch repair genes associated with hereditary nonpolyposis colorectal cancer (i.e., Lynch syndrome).[6–8] Such genetic testing can inform personalized cancer risk management strategies, including the use of cancer screening tests.

Although screening tests including mammography, colonoscopy, and sigmoidoscopy have established population-level health benefits,[9–12] these tests also carry risks; as such, there is a desire to identify individuals who will derive the greatest benefit and least harm. Genetic testing for cancer susceptibility may have utility for risk stratification, thereby allowing for a more focused application of screening tests to those at greatest disease risk.[13–16] For example, estimating an individual's cancer risk based on genetic markers and traditional risk factors may lead to a more refined use of currently recommended screening tests (e.g., mammography).[17,18] Information derived from genetic testing could also shift the

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balance of harms and benefits for some individuals such that screening tests like the PSA test, which is not recommended for the general population, may have utility in those with greatest genetic risk for prostate cancer.[19–21] Those at higher genetic risk may also be candidates for more sensitive screening approaches (e.g., breast MRI).

For the promise of risk-stratified cancer screening to be realized, at-risk individuals will need to be identified and obtain genetic testing. Although nearly half of Americans are aware of genetic testing for cancer susceptibility,[22] test use is low in the general population[23] and among individuals with increased family history.[24] Primary care providers (PCPs) play a critical role in addressing patients' preventive healthcare needs and are in a position to assist patients with making informed decisions about the appropriate use of genetic testing. Prompted by patient inquiries or the collection and interpretation of family history data, PCPs can refer patients to genetic professionals for risk assessment and testing. Alternatively, PCPs can directly order genetic tests for patients, which may be particularly important when access to genetic professionals is limited. PCPs may also be tasked with interpreting the results of direct-to-consumer genetic tests purchased without physician involvement. Furthermore, PCPs can use patients' genetic test results to inform recommendations for primary (e.g., prophylactic surgery, chemoprevention) and secondary (e.g., use and timing of screening tests) cancer prevention efforts.[5,6]

However, research suggests that PCPs may be unprepared for the task of helping patients gain access to benefits of cancer genetic risk information. According to a systematic review of studies regarding genetic services for common diseases including cancer published from 2000–2008,[25] PCPs noted limitations in their knowledge about basic genetics and confidence in collecting and interpreting family history data, and generally felt underprepared for integrating genomic medicine into patient care. Similarly, a systematic review of research published from 2001–2012 concluded that a lack of knowledge and skills, along with attitudes and concerns about patient distress, were common barriers to PCPs integrating genetic services into patient management.[26] This past work confirms that PCPs' knowledge and attitudes are critical to their genetic testing utilization.

With the present study, we sought to describe US PCPs' knowledge, attitudes, and communication-related behaviors regarding genetic tests that could inform risk-stratification approaches for breast, colorectal, and prostate cancer screening. To inform a future research agenda in this area, we aimed to both describe the scope of research questions addressed in past studies and summarize their findings. For this review, we defined *knowledge* as PCPs' level of subjective or objective understanding about genetic services; basic genetic concepts; treatment and management options for patients with genetic mutations; and their self-efficacy, confidence, and comfort in discussing genetic testing with patients. *Attitudes* included PCPs' personal opinions or views about topics including the validity or utility of clinical and direct-to-consumer genetic tests for hereditary cancer risk; incorporation of genetic testing into practice; and ethical, legal, or social implications of testing for patients. *Communication-related behaviors* reflected the extent, frequency, and outcomes of PCPs' discussions of genetic testing for cancer risk with patients. We limited the review to genetic testing related to breast, colorectal, and prostate cancer because these represent cancers of high incidence and mortality,[27] and for which genetic information may be particularly useful in risk-stratification decisions for the frequency and/or modality of screening tests.[13–15,28]

METHODS

Literature search

A health science librarian (MLF) comprehensively searched six databases (PubMed, EMBASE, Web of Science, PsycINFO, CINAHL, Cochrane Library) to identify peer-reviewed articles relating to PCPs and genetic testing for breast, colorectal, or prostate cancer. The search strategy was developed to retrieve articles assessing PCP knowledge; attitudes; provider- or patient-initiated discussions of genetic testing; referral behaviors; and interventions to improve these domains (for details see Figure 1 and Online Appendix 1). Additional articles were identified through searching reference lists of selected papers.

Inclusion/exclusion criteria

Studies were included if they addressed genetic testing related to breast, colorectal, or prostate cancer and involved PCPs (operationalized as internists, family practitioners, obstetrician/gynecologists, nurse practitioners, and physician assistants, consistent with IOM[29] and CMS[30] definitions). Studies involving other provider types were included if relevant outcomes for PCPs were reported separately and/or if PCPs represented a majority of the sample. Studies were limited to English-language articles set in the US and published from January 2008 (the last year of data included in a related systematic review[25])–August 2016 in peer-reviewed journals, excluding commentaries, editorials, proceedings, dissertations, book reviews, and meeting abstracts. Gray literature was not searched.

Record review

The combined searches produced 2377 results (Fig. 1). After removing duplicates, one team member reviewed the remaining 1822 records and excluded 1725 based on irrelevant article titles and abstracts (e.g., bench science studies, international data). Another team member verified these decisions. Two team members screened the full texts of the remaining 97 articles, eliminating an additional 55 that did not address genetic testing or a cancer of interest.

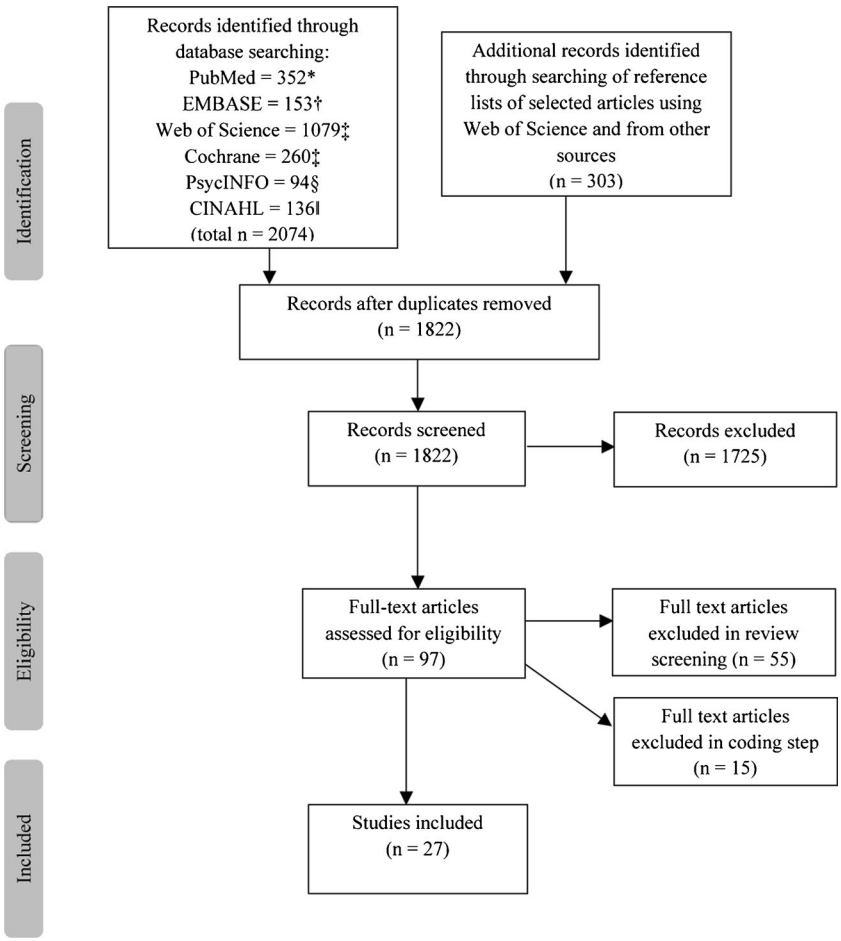

Fig. 1 PRISMA flow diagram describing the literature search and record review process for the present study. *PubMed was searched on September 5, 2016, using MeSH terms and title/abstract search; †EMBASE was searched on August 3, 2016, using a combination of EMTREE and title/abstract search; ‡Web of Science and Cochrane Library were searched on August 3, 2016, using keyword search; §PsycINFO was searched on August 13, 2016, using keyword search; ‖CINAHL was searched on August 18, 2016, using a combination of CINAHL headings and keyword search

Article audit

Five team members double-coded the full text of the remaining 42 articles using an audit form and data dictionary developed for this review (Online Appendix 2), and they determined that 15 articles did not meet the study criteria. Team members coded the final 27 articles for provider type; cancer type; data source (e.g., survey); and PCPs' knowledge, attitudes, and communication-related behaviors. Relevant research questions, measurement approaches, and results were documented. Coding differences were discussed and reconciled by coding pairs; when disagreement arose, final decisions were reached by group consensus. The team reviewed these data and used narrative synthesis methods to integrate findings into a descriptive summary.

RESULTS

Table 1 shows characteristics of the reviewed studies; Table 2 summarizes the research questions, methods, and relevant outcomes of each study. Most commonly, studies included family practitioners (n = 17), internists (n = 14), and obstetricians/gynecologists (n = 12), with sample sizes ranging from 7–1500 participants. Studies focused on genetic testing for breast (n = 23) and colorectal cancer risk (n = 12); only one study examined testing for prostate cancer risk. Surveys were the most commonly used data collection method (n = 21), and a minority of studies (n = 3) evaluated an intervention.

Knowledge of genetic tests for cancer risk

We identified 24 (of 27) articles addressing research questions relevant to PCPs' knowledge.[33–53,55,56,58] Of these, 13 studies evaluated PCPs' objective knowledge.[34,35,38,39,41,42,44,47–50,55,58] In seven studies, objective knowledge was assessed with a survey utilizing a scale or test to measure accurate understanding of basic genetic principles, clinical practice guidelines, and/or features of hereditary cancer syndromes.[35,38,39,42,47,48,50] In the other six studies, case scenarios or standardized patients were used to measure PCPs' abilities to identify high-risk patients and appropriate genetic testing situations.[32,34,41,44,49,55] Aside from articles reporting on the same sample,[38,48] there was no overlap in measures. Across

Table 1 Characteristics of the 27 Reviewed Articles

Characteristic	*n* (%)*
Primary care provider type†	
Family practitioners	17 (63)
Internists	14 (52)
Obstetricians/gynecologists	12 (44)
Nurse practitioners/nurses	9 (33)
Physician assistants	8 (30)
Study sample size	
≤50 participants	4 (15)
51–100 participants	7 (26)
101–500 participants	11 (41)
>500 participants	5 (19)
Cancer site	
Breast	23 (85)
Colorectal	12 (44)
Prostate	1 (4)
Other (e.g., ovarian, endometrial)	11 (41)
Study topic	
Primary care provider knowledge	24 (89)
Primary care provider attitudes	11 (41)
Primary care provider communication-related behaviors	9 (33)
Data collection method	
Survey	21 (78)
Qualitative (e.g., interviews, focus groups)	1 (4)
Mixed methods (i.e., qualitative and quantitative approaches)	2 (7)
Other (e.g., abstraction from medical records, standardized patient)	4 (15)
Study evaluated effects of an intervention (yes)	3 (11)

**Values do not total 27 (100) because articles could be characterized with multiple response options*
†A total of 14 studies also included non-primary care providers (e.g., surgeons, oncologists, unspecified MDs) as participants. Additional details are provided in Table 2

these studies, PCPs were reported to have incomplete or inaccurate knowledge about the inheritance and characteristics of hereditary cancer syndromes and interpretation of genetic test results. For instance, in a cross-sectional survey of 176 family practitioners, the modal score on a 10-point scale of factual hereditary breast and colorectal cancer knowledge was 6, with none of the sample accurately answering all items.[42] In addition, three studies examined objective knowledge pertaining to legal genetic discrimination protections.[39,44,53] Knowledge in this domain was concluded to be suboptimal; for instance, in a cross-sectional survey of 1120 family practitioners and internists, 9% had accurate knowledge about legal insurance protections related to genetic test results.[53]

Ten studies examined PCPs' self-reported subjective or perceived knowledge.[33,37,42–45,47,50,51,55] Although there was no overlap in measures, studies consistently reported a lack of PCPs' confidence in their genetic testing-related knowledge. For example, in a cross-sectional survey of 1311 family practitioners, 54% were not confident in their knowledge of genetic testing in primary care including testing for breast cancer risk.[45] In another cross-sectional survey of 1209 Oregon clinicians, 83% of primary care providers and 76% of obstetricians/gynecologists reported that they were "not at all" or "somewhat" (vs. "moderately" or "very") confident in their knowledge of colorectal cancer genetics.[37] Seven studies also evaluated PCPs' knowledge in terms of their comfort level with discussing aspects of cancer genetic testing with patients,[33,40,43,44,46,53,56] with results suggesting that PCPs rarely felt prepared for the task of counseling patients about genetic testing. For example, a survey of 50 obstetricians/gynecologists found that 74% did not feel comfortable counseling patients about available genetic testing for Lynch syndrome, and 76% did not feel comfortable counseling patients about such testing criteria.[40]

One study evaluated an intervention designed to influence PCPs' genetic testing knowledge and clinical behaviors.[52] Scheuner and colleagues used a mixed-method study design and medical record abstraction to evaluate the feasibility and effectiveness of a multicomponent cancer genetics toolkit in the context of women's primary care clinics at a Veterans Administration medical center. Among seven primary care physicians, nurse practitioners, and physician assistants, toolkit use improved knowledge about cancer genetics [mean total correct responses increased from 59% (range: 26%–77%) pre-implementation to 73% (range: 52%–90%) post-implementation].

Attitudes regarding genetic tests for cancer risk

We identified 11 (of 27) articles including research questions regarding PCPs' attitudes.[33,34,41–45,51,52,55,56] Of these, five studies examined PCPs' views about ethical, legal, or social implications of testing.[33,41,43,44,55] These studies noted varying beliefs about the effect of genetic testing on patients' anxiety. Specifically, whereas a cross-sectional survey of 351 family practitioners, internists, and obstetrician/gynecologists found that only 14% of respondents believed that information about breast cancer risk creates unnecessary anxiety for many women,[41] a mixed-method assessment of 24 family practitioners, urologists, and urology residents found that 74% of participants were concerned that increased-risk results could unnecessarily increase patient anxiety.[33] Concerns about privacy issues and discrimination were also documented; for instance, a cross-sectional survey of 220 family practitioners and internists found that 27% were "very concerned" about genetic privacy,[43] and qualitative data collected from 24 family practitioners, urologists, and urology residents highlighted concerns that test results could put patients at risk for discrimination by insurance companies and employers.[33] Furthermore, a cross-sectional survey featuring a vignette of a hypothetical low-risk patient found that among 284 family physicians, 65% believed that refusing to refer the patient to genetic services would harm the provider-patient relationship.[55]

Three studies evaluated PCPs' perceptions regarding the validity or utility of clinical genetic tests, all in the context of breast cancer risk.[34,41,45] One reported on coded interactions between 86 family practitioners and internists with standardized patients and noted that many providers expressed skepticism about the value of genetic testing.[34] Through the analysis of survey items, the other studies reported predominantly positive provider opinions about the accuracy and health benefits of genetic testing.[41,45] Two studies examined PCPs' perceptions

Table 2 Summary of 27 Studies of Primary Care Provider Knowledge, Attitudes, and Communication-Related Behaviors Regarding Cancer Genetic Testing

Article	Provider type(s) and (sub)sample size	Cancer type(s)	Relevant research questions	Data collection method, relevant outcomes, and measurement
Bell et al. 2015[31]	Primary care physicians (n = 121)	Breast	Does an educational intervention elicit appropriate provider communication behaviors related to HBOC genetic counseling and testing?	Standardized patient case scenarios. C: Standardized patient visits audio-recorded, transcribed, and coded for 69 physician behaviors by 2 coders (average kappa = 0.91)
Bellcross et al. 2011[32]	Family practitioners (n = 515), pediatricians (n = 250), internists (n = 485), OB/GYNs (n = 250)	Breast, ovarian	What are providers' awareness and use of *BRCA1/2* testing? How aware are providers of family history patterns appropriate for *BRCA1/2* testing referral?	Survey. K: Providers asked to select indications for *BRCA1/2* testing from 7 clinical scenarios representing increased and low-risk situations consistent with USPSTF guidelines
Birmingham et al. 2013[33]	Primary care physicians (n = 10), urologists (n = 9), urology residents (n = 5)	Prostate	What are providers' attitudes, knowledge, and behavioral intentions regarding DTC genomic testing for prostate cancer risk?	Mixed methods. K, A, C: Single items and multi-item scales*; focus groups coded with grounded theory approach by 2 coders (kappa = 0.77)
Burke et al. 2009[34]	Family practitioners and internists (n = 86)	Breast	What are Seattle/King county providers' family history-taking and genetic risk assessment skills related to breast cancer?	Standardized patient case scenarios. K, A, C: Standardized patient visits audio-recorded, transcribed, and coded by 2 coders (disagreement resolved by a third coder in ≤2.1% of cases)
Chan et al. 2014[35]†	Family practitioners and internists (n = 116), other specialties (e.g., surgery) (n = 24)	Colorectal	What is the impact of provider academic affiliation and training on hereditary colorectal cancer knowledge?	Survey. K: 6 true-false questions consistent with national guidelines and general knowledge about hereditary colorectal cancer derived from literature, vetted by multidisciplinary team
Cohn et al. 2015[36]†	Family practitioners and internists (n = 116), other specialties (e.g., surgery) (n = 24)	Breast, ovarian	What is the impact of provider training on HBOC knowledge?	Survey. K: 6 true-false questions consistent with national guidelines about HBOC and genetic testing, vetted by multidisciplinary team
Cox et al. 2012[37]	Family practitioners, internists, NPs, and PAs (n = 363), OB/GYNs (n = 333), naturopaths (n = 216)‡, specialists (n = 297)‡	Breast, colorectal, ovarian	What are Oregon providers' levels of knowledge and use of 8 cancer genetic tests?	Survey. K: Single-item measures included in an investigator-designed survey piloted with Oregon healthcare providers
Cragun et al. 2013[38]§	Physicians (n = 53), NPs (n = 25), nurses (n = 2), PAs (n = 1)	Breast, ovarian	What are the *BRCA1/2* knowledge, interest, preferences, and facilitators/barriers to cancer genetics education among Florida non-genetics community providers offering *BRCA1/2* testing?	Survey. K: 8 multiple-choice and clinical scenario items used to assess knowledge and adherence to national clinical practice guidelines*; survey developed by multidisciplinary team and evaluated for face and content validity
Cragun et al. 2016[39]	NPs (n = 27), OB/GYNs (n = 26), oncologists (n = 22), general surgeons (n = 6), nurses (n = 4), PAs (n = 1), other specialties (n = 5)	Breast, ovarian	What are the HBOC knowledge, practices, and adherence to pretest counseling elements among Florida non-genetics providers offering HBOC genetic testing?	Survey. K, C: Multiple items that were used in a previous survey* along with additional investigator-designed items
Frey et al. 2014[40]	OB/GYNs (n = 50), general surgeons (n = 62)‡	Colorectal	What is providers' cancer screening knowledge for women with HNPCC?	Survey. K: Questionnaire developed by the investigators
Guerra et al. 2009[41]	Family practitioners (n = 144), internists (n = 140), OB/GYNs (n = 67)	Breast	What are the prevalence and determinants of the adoption of breast cancer risk assessment by providers?	Survey. K, A, C: Multi-item scales to assess identification of high-risk clinical scenarios, risk assessment beliefs, and physician behaviors as part of an investigator-designed survey
Kelly et al. 2009[42]	Family practitioners (n = 176)	Breast, colorectal	What are the awareness of hereditary cancers and genetic testing experiences of providers in rural and Appalachian practice?	Survey. K, A: Multi-item, internally valid scales and clinical scenarios used as part of an investigator-designed survey
Klitzman et al. 2013[43]	Family practitioners (n = 8), general medicine internists (n = 42), other internist specialties (n = 170)	Breast, colorectal	Do providers order genetic tests for cancer and other disorders for their patients? What factors are associated with these decisions?	Survey. K, A: 44-item questionnaire developed by the investigators based on published literature and clinical experience
Lowstuter et al. 2008[44]	Family practitioners, internists, OB/GYNs, NPs, and medical oncologists (n = 1181)	Breast, colorectal, endometrial	What are Californian non-genetics providers' perceptions and knowledge of cancer genetics and genetic discrimination laws, attitudes, and referral practices?	Survey. K, A: 47-item survey; items based on a previously validated pilot study*
Mainous et al. 2013[45]	Family practitioners (n = 1311)	Breast	What are providers' perceptions and experiences with clinical and DTC genetic testing?	Survey. K, A, C: Investigator-designed items developed from a literature review

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Table 2. (continued)

Article	Provider type(s) and (sub)sample size	Cancer type(s)	Relevant research questions	Data collection method, relevant outcomes, and measurement
Menzin et al. 2010[46]	OB/GYNs (n = 289)	Breast, gynecologic	What are providers' practice, training, and knowledge of breast health maintenance and cancer screening?	Survey. K: 18-item investigator-designed survey
Nair et al. 2015[47]	Family practitioners (n = 89), internists (n = 51), OB/GYNs (n = 74), NPs and PAs (n = 61)	Breast	What is Georgia providers' existing HBOC knowledge?	Survey. K: 31-item survey; items adapted by experts from prior national surveys, and beta tested with healthcare providers*
Pal et al. 2013[48] §	OB/GYNs (n = 28), NPs (n = 24), nurses (n = 3), PAs (n = 1), genetic counselors (n = 5)‡, general surgeons (n = 12), medical oncologists (n = 6), other specialties (including internists) (n = 7)	Breast, ovarian	What are the knowledge and practices related to identifying, testing, and managing individuals at risk for *BRCA1/2* mutations among Florida providers offering genetic testing?	Survey. K: Multiple-choice and clinical scenario items*; survey developed by multidisciplinary team and evaluated for face and content validity
Plon et al. 2011[49]	OB/GYNs (n = 51), family practitioners (n = 50), internists (n = 48), general surgeons (n = 45), hematology/oncology physicians (n = 31)	Breast	How would Texas non-genetics providers use a cancer patient's *BRCA1/2* results to make genetic testing and risk management recommendations for a healthy at-risk relative?	Survey. K: Investigator-designed case-based descriptions and follow-up questions
Ready et al. 2010[50]	OB/GYN residents (n = 65)	Breast, colorectal, ovarian	What are OB/GYN residents' family history documentation practices and knowledge of HBOC and HNPCC?	Survey. K: Test adapted from previous studies and evaluated for comprehension at a professional conference*
Salz et al. 2012[51]	Family practitioners (n = 115), internists (n = 20), NPs (n = 13), PAs (n = 6), other (n = 2)	Colorectal	What are providers' needs for information about colorectal cancer survivorship care?	Survey. K, A: Investigator-designed questionnaire informed by an Institute of Medicine framework for survivorship care plans
Scheuner et al. 2014[52]	Physicians (n = 5), PAs (n = 1), NPs (n = 1)	Breast, colorectal	What is the feasibility and effectiveness of a multi-component cancer genetics toolkit in the women's primary-care clinics at a large Veterans Administration medical center?	Mixed methods and abstraction from medical records. K, A, C: Investigator-designed survey items developed through literature review, expert opinion, and cognitive testing, plus medical record review
Shields et al. 2008[53]	Family practitioners (n = 616), internists (n = 504)	Breast, colorectal, ovarian	What are providers' genetic testing experiences, and do experiences differ among minority-serving providers?	Survey. K: Investigator-designed survey informed by focus groups and interviews with healthcare providers, expert feedback, and literature review
Vadaparampil et al. 2015[54] §	Physicians (n = 53), NPs (n = 25), nurses (n = 2), PAs (n = 1)	Breast, ovarian	Do non-genetic providers perform guideline-based intake and informed consent before *BRCA1/2* testing?	Survey. C: Multiple items*; survey developed by multidisciplinary team and evaluated for face and content validity
White et al. 2008[55]	Family practitioners (n = 284)	Breast	Are providers' referral patterns consistent with USPSTF recommendations when a patient requests *BRCA1/2* testing? Do patient characteristics influence referrals?	Survey. K, A: Investigator-designed items following a vignette of a hypothetical patient not appropriate for referral to *BRCA1/2* genetic services based on USPSTF guidelines
Wood et al. 2013[56]	Family practitioners (n = 14), internists (n = 17), OB/GYNs (n = 9)	Breast, colorectal	What are providers' perceptions of their role in managing cancer risk based on family history?	Interview/focus groups. K, A: Interviews were audio-recorded and transcribed; data analyzed by 1 coder in collaboration with investigator team
Zazove et al. 2015[57]	Family practitioners (n = 89)	Breast, colorectal	Can tailored electronic medical record prompts about patient family history improve providers' test/referral ordering?	Abstraction from medical records. C: Manual chart audits conducted by trained auditors who reviewed visit notes, test orders, and referrals

Note: Providers = Primary care providers; K = knowledge; A = attitudes; C = communication-related behaviors; OB/GYNs = obstetricians/gynecologists; NPs = nurse practitioners; PAs = physician assistants, HBOC = hereditary breast and ovarian cancer; DTC = direct-to-consumer; HNPCC = hereditary nonpolyposis colorectal cancer; USPSTF = United States Preventive Services Task Force

**Relevant outcomes were assessed with measures used and/or validated in prior research; see original publication for details*

†Studies report on the same sample

‡Findings from this group excluded for the purposes of the present study

§Studies report on the same sample

Although this study included respondents from Canada, the majority of respondents (98%) were from the US

regarding the validity and utility of direct-to-consumer tests for cancer risk, documenting generally unfavorable attitudes.[33,45] For example, a cross-sectional survey of 1311 family practitioners found that 58% of participants believed this testing would do more harm than good for patients.[45]

Five studies investigated PCPs' attitudes about the implications of genetic tests for cancer screening and other risk management behaviors.[33,41,44,51,56] For instance, consistent with the goals of risk-stratified cancer screening, 65% of 1181 surveyed non-genetics providers agreed that genetic testing may reduce unnecessary cancer screening.[44] Similarly, a mixed-method assessment of 24 family practitioners, urologists, and urology residents found that 86% believed direct-to-consumer genomic testing could inform the age at which to start prostate cancer screening and 76% believed such testing could inform screening frequency.[33] A cross-sectional survey of 156 PCPs also found that 78% of participants perceived that it is very important to know about genetic counseling and testing for cancer survivors in order to identify high-risk individuals who could benefit from more comprehensive surveillance.[51]

Communication-related behaviors regarding genetic tests for cancer risk

We identified 9 (of 27) articles examining aspects of provider-patient communication.[31,33,34,39,41,45,52,54,57] Of these, four studies provided descriptive information about PCPs' experiences discussing genetic risk and testing (2 relying on surveys in the breast cancer context[41,45] and 1 using mixed-methods in the prostate cancer context[33]). The other used a novel study design featuring standardized patients to evaluate communication behaviors among 86 providers and found that in only 21% and 3% of encounters did providers express opinions suggesting that patients at high maternal or paternal breast cancer risk, respectively, were candidates for genetic testing.[34] Two additional studies used cross-sectional surveys to assess the extent to which PCPs adhered to clinical guidelines in discussions about consenting to hereditary breast and ovarian cancer testing[39,54] and observed that many were non-adherent (e.g., among 81 providers, 39% did not always discuss implications for family, and 64% did not always discuss the possibility of another hereditary cancer syndrome[54]). Furthermore, two studies reported limited instances of discussions between PCPs and patients regarding direct-to-consumer testing (e.g., with 71%[45] to 92%[33] of providers not having had a patient ask questions about direct-to-consumer testing).

Three studies involved the use of interventions designed to influence PCPs' communication behaviors.[31,52,57] For instance, as noted, Scheuner and colleagues evaluated a multicomponent cancer genetics toolkit targeted at providers that improved their discussion and documentation of cancer family history and appropriate patient referrals for genetic consultation.[52] Another study evaluated an educational intervention among 121 primary care physicians and found that some communication behaviors improved (e.g., 78% of intervention physicians explored genetic counseling benefits with a standardized patient versus 61% of controls); however, the intervention did not lead to significant differences in the offering of a genetic counseling referral or recommending of genetic testing.[31]

DISCUSSION

PCPs represent the front line of screening for inherited disease risks and are increasingly involved in delivering genetic services.[59–61] As gatekeepers, PCPs need to know how to interpret genetic test results, understand when to refer and ask for second opinions of genetic professionals,[59,62,63] and address complex personal, cultural, ethical, legal, and social issues associated with genetic testing.[59]

This review confirmed relatively low levels of objective and subjective genetic testing-related knowledge among PCPs. Such trends have serious implications for the integration of genetic testing technologies into routine patient care; feeling unqualified to manage tasks surrounding test ordering and counseling[46] and uncertain about guidelines[43] inhibits PCPs' genetic test adoption. Although basic genetic knowledge appears to be stronger among younger, more recent medical graduates, specialists, and providers in academic medical centers,[25,64] there is a clear need for educational interventions that can improve genetics-related knowledge among all PCPs. Evidence suggests that PCPs are generally open to additional genetics education, such as brief training programs that contain continuing web-based education modules accessible outside of a classroom.[38,43] Opportunities to develop skills to interpret genetic tests and understand how to maintain genetic privacy and confidentiality appear to be of interest to these providers.[43] Furthermore, access to clearinghouses that provide a comprehensive listing of available tests, information on test sensitivity and specificity, the pros and cons of testing for specific conditions, and professional society guidelines would enhance PCPs' ability to keep up with developments in genetic testing[65] and address the challenge of understanding which tests are worth doing on which patients. Such knowledge will be vital for PCPs as genetic testing approaches and decisions become more complex (for instance, as reflected in the complexity of ordering and interpreting multi-gene panel tests, which are cost-effective, efficient tests that allow for the simultaneous analysis of numerous moderate- and high-penetrance cancer susceptibility genes[66]).

Across studies, there was substantial variation in the measures used to assess objective genetic testing-related knowledge. This suggests a need for future research to develop validated, standardized measures that assess critical factual knowledge necessary for PCPs to integrate genetic testing into patient management. Such measures may reflect knowledge related to core competencies (e.g., principles of Mendelian inheritance, characteristics of high-risk family histories) and professional society guidelines regarding testing. Data from these measures could be coupled with patient-level data (derived from electronic health records and learning health systems[67]) to assess the efficacy of provider-directed educational interventions in improving knowledge and testing-related behaviors.

PCPs' attitudes, driven by beliefs about test validity or clinical utility, are complex and evolving. Although genetic

testing may have value for risk stratification, some skepticism exists among PCPs;[34] such skepticism may be ameliorated through future research aimed at improving provider knowledge and developing clinical decision support tools to guide providers to order the correct tests and use genetic data for patient management. Two of the reviewed studies also found generally unfavorable attitudes regarding the utility of direct-to-consumer genetic tests.[33,45] As public awareness and use of direct-to-consumer testing grows,[68,69] PCPs are likely to be confronted with patients seeking clarity, interpretation, or reassurance about these results.[70,71] Research is needed to develop and evaluate educational and communication interventions that prepare PCPs to navigate these situations and to help their patients recognize the limitations of these tests.[71–73]

We identified fewer studies examining aspects of communication. Yet, because of their influential role, PCPs will need to address the complexities of risk communication in the genomics era. Ideally genetic professionals will offer genetic counseling; however, the need to provide referrals, supplement limited access to genetic professionals, and integrate genetic risk information into the holistic care of a patient will nonetheless require PCPs to collect a detailed family history and discuss nuanced aspects of individual risk with patients (e.g., the incremental risk for chronic disease attributable to genetic versus behavioral factors).[74]

Research to develop and test innovative communication-focused interventions is warranted. Consistent with suggestions from IOM,[75] PCPs may benefit from training that includes opportunities to directly learn about communication preferences from patients with an inherited predisposition to disease. To evaluate these programs, researchers should assess the perspectives of PCPs and their patients along with patient-level care delivery (e.g., satisfaction) and health outcomes (e.g., knowledge, adherence to appropriate screening tests). PCPs could benefit from genetics-related electronic health record tools, and those in community settings could particularly benefit from a team-based approach that includes assistance of a genetic professional trained in risk assessment and communication. Future research to evaluate technology-based approaches that extend the reach of a limited genetics workforce, such as telegenetics,[76,77] telephone counseling,[78,79] and web-based case conferencing,[80] may address this need.

Limitations

Although we used a comprehensive search strategy, all relevant literature may not have been captured in this review. We aimed to focus on genetic testing for breast, colorectal, and prostate cancers; however, the vast majority of identified studies addressed *BRCA1/2* testing, and we did not examine potential differences in outcomes across cancer types. Future studies will need to assess PCPs' experiences with more diverse forms of genetic testing. Furthermore, this review focused predominantly on broad research trends and gaps from the perspective of PCPs, thereby not addressing patient perspectives, social factors (e.g., racism), or healthcare system factors (e.g., clinical decision support) that can influence care delivery.[81] This review did not focus on how provider characteristics (e.g., training, age) may contribute to differences in the outcomes of interest; future research could explore such differences. Finally, this review does not offer insight into PCPs' experiences with novel applications of genomic testing currently being introduced into cancer care (e.g., multi-gene panel testing, although a reviewed article noted that 42% of providers have ordered such testing[39]).

Conclusions

We have found a need to move beyond research questions related to describing PCPs' cancer genetic knowledge and attitudes about the utility of testing. Important knowledge gaps exist in the development, evaluation, and implementation of interventions and educational programs designed to improve PCPs' understanding of fundamental genetic principles, genetic test characteristics, and professional guidelines regarding testing; assuage PCPs' concerns about clinical, ethical, legal, and social implications of testing; and promote open and effective communication about genetic risk and testing. Interventions and programs that capitalize on innovative methods for reaching and engaging PCPs and that allow them to gain access to and develop partnerships with genetic professionals are needed. Such efforts could ultimately help PCPs to embrace genomics into their practices and effectively address their patients' preventive health needs.

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