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ARTICLE

Utility of eConsult to enhance delivery of cancer genetic services and identify hereditary cancer knowledge gaps in primary care

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ABSTRACT

Purpose: This study analyzed the utility of electronic consultation (eConsult) for hereditary cancer (HC) and aimed to identify primary care practitioner (PCP) knowledge gaps.
Methods: A retrospective mixed-methods study was used to evaluate 200 randomly selected PCP eConsult cases submitted to cancer genetics specialists in Ontario, Canada.
Results: In 65% (129/200) of eConsults, PCPs indicated they received clear advice for a new course of action. In 34% (68/200), referral was contemplated but now avoided. In 8% (16/200), referral was advised when not originally planned. For 89% (177/200), eConsult was considered valuable. For most, (63%, 125/200), PCPs agreed eConsult addressed a clinical problem that should be incorporated into continuing medical education. PCPs' questions were mainly about cancer screening (114), genetic testing (107), or genetics referral (76). Geneticist recommendations were mainly about cancer screening (154), genetics referral (104), and the High-Risk Ontario Breast Cancer Screening Program (41). PCP knowledge gaps identified included cancer screening guidelines (112), genetics referral criteria (100), High-Risk Ontario Breast Cancer Screening Program screening criteria (71), and understanding of genetics principles (237).
Conclusion: eConsult is an effective tool for PCP access to HC specialists. Identifiable knowledge gaps emerge that could be used to enhance continuing medical education and drive innovative HC service delivery.

Introduction

Advances in genomic medicine have led to an increasing demand for genetic assessment and testing for hereditary cancer (HC). This is driven in part by novel gene discovery, expanded understanding of associated risks, personalized cancer treatment options, and implementation of new screening models for high-risk patients.[1,2] Genetics health care professionals (GHPs) expect that primary care practitioners (PCPs) will be an integral part of recent genomic advances.[3] Although PCPs also recognize participation in cancer genetic assessment as an important role, they often

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report limited genetics knowledge including lack of skills in taking an appropriate family history, knowledge about guidelines and services and lack of confidence to engage in such services.[4-7] Genetics clinic wait times can be upward of 2 years owing to shortage of GHPs and expanded need for personalized assessments.[8] This has led to implementation at many centers of innovative models of care, such as mainstreaming genetic testing to the oncology clinic and electronic consultation (eConsult).

eConsult to a Medical Geneticist has been freely available to health care practitioners in Eastern Ontario, Canada, since 2013 and offers an option for receiving timely and accessible advice, has the potential to avoid unnecessary patient referrals, and has been well received by both health care practitioners and consultants.[9-11] Over an 18-month period in 2019 to 2020, geneticists providing eConsults to PCPs in the Champlain Building Access to Specialists through eConsultation (BASE) region of Ontario indicated that most of the questions they received (52%) were regarding cancer, predominantly breast/ovarian cancer.[9] To our knowledge, analysis of eConsult specifically for HC genetics has not previously been completed. Our primary aim was to identify common themes emerging from the type of HC questions asked of geneticists by PCPs that could direct future innovations and educational initiatives. We also explored the utility of eConsult as a model for enhancing the delivery of cancer genetic services.

Materials and Methods

Design

This retrospective mixed-methods study evaluated 200 randomly selected eConsult cases, from a total of 380, submitted by PCPs to the cancer genetics specialist group on the Champlain eConsult BASE Service between October 1, 2016, and December 31, 2022. This study was approved by the Ottawa Hospital Research Ethics Board.

Champlain eConsult BASE service

The Champlain eConsult BASE Service was first established in 2010 and allows PCPs to communicate asynchronously with a specialist about a patient's care on a secure web-based platform. PCPs can send an eConsult request to a specialist from one of over 150 specialties by creating a case in the web browser with a specific question accompanied by case details and the option to add supplemental information as attachments (eg, imaging and laboratory reports). The specialist can then offer advice regarding medical recommendations and assess if further referral is necessary. After the closure of the eConsult case, PCPs were required by the eConsult service to complete a close-out survey. This survey evaluated whether the specialist response confirmed their initial course of action or supplied them with new or additional information. Additionally, it assessed the impact of the eConsult on the necessity of an in-person referral. The service is funded by the Ontario Ministry of Health and is offered to PCPs at no charge. PCPs receive a flat rate per case by submitting a fee code for $16 CAD, and specialists are compensated at an hourly rate of $220 CAD per hour prorated to the time (median 15 minutes) it takes them to complete an eConsult case.[12]

Setting

In Ontario genetic health care for patients is covered by the provincial insurance program. PCPs are not able to order provincially funded HC genetic testing and must refer their patients to a genetics clinic. The referral may be accepted or declined depending on whether it meets criteria for assessment. Information regarding HC genetic assessment referral criteria and criteria for genetic testing eligibility is available on provincial and often local genetic clinic websites. After genetic counseling and testing (if offered and accepted), posttest consultation occurs with a genetic professional, and cancer screening recommendations are provided back to the PCP according to the genetic test result and family cancer history. There is also a special program for high-risk breast cancer screening. Patient eligibility for this program is determined after genetic risk assessment.

The Champlain eConsult BASE Service is based in the Champlain region of Ontario, Canada, encompassing Ottawa and its surrounding communities. This region comprises a linguistically and culturally diverse population of over 1.3 million residents in Eastern Ontario. The area consists predominantly of rural landscapes with a single large urban center. The service is additionally offered to PCPs in Nunavut, Canada's northernmost and rural territory.

Data collection

All routine service utilization data, including type of referring PCP, specialist response time, close-out survey responses, rurality index for Ontario score, and a log of all exchanges between the PCP and specialist, were securely collected and stored in the Champlain eConsult BASE system using robust methods, as detailed elsewhere.[13]

Data analysis

eConsult utilization data

Summary statistics and frequencies were used to analyze the 200 cancer genetics eConsult cases to describe patterns such as response time, total time spent by the specialist, patient demographics, and the responses from the PCP close-out survey.

eConsult content

Content analysis of the PCP eConsult questions and geneticist recommendations was retrospectively completed to gain insight into common themes. Four investigators including geneticists (A.R. and M.C.), genetic counselor (S.M.), and family physician (FP, J.C.) independently reviewed the clinical questions and recommendations using modified versions of 2 existing validated taxonomies: the International Classification for Primary Care, version 3[14] and the Taxonomy of Generic Clinical Questions.[15] The International Classification for Primary Care, version 3 was used to classify the clinical topics, and the Taxonomy of Generic Clinical Questions was used for PCP question type and specialist recommendation type classification (eg, diagnosis, management, and screening). Knowledge gaps were classified by the investigators. The established taxonomy was piloted through duplicate coding by the investigators on a subset of 24 eConsult cases (ie, 4 random cases from each year from 2017 to 2022). The investigators discussed their independent assessments using an iterative approach to come to a consensus on a final list of codes. Taxonomies were further refined and piloted on an additional set of 24 random eConsult cases, which resulted in general agreement of the codes between the 4 investigators. In total, 48 eConsults were assessed before no new codes were emerging and coding agreement was achieved. Once codes were identified and the taxonomy was finalized, the 4 investigators analyzed a new subset of 200 random eConsult cases, in which each case was coded by 2 alternating investigators, in which the pair of investigators reached a consensus on each code. With many eConsult cases encompassing multiple questions and answers, the eConsult was not restricted to only 1 code to prevent any loss of information. No new codes emerged when coding the final 200 eConsult cases.

Results

Cohort characteristics

For the random 200 cancer genetics eConsults analyzed from the total of 380, there were 150 requesting PCPs and 3 answering medical genetic specialists. Of the 150 PCPs, 135 were FPs, whereas 15 were nurse practitioners (NPs). Nearly one-quarter (35/150 [23%]) sent in more than 1 eConsult request (maximum 4 cases per provider, with a median of 1 case per provider). Demographic information was available for 133 requesting FPs. Most (104/133 [78%]) were female. The time from medical school graduation ranged from 4 to 45 years (median 17). Most eConsult cases (182/200 [91%]) were submitted by an FP vs 18 of 200 (9%) by a NP and 22 of 200 (11%) cases were identified as rural, including 5 from Nunavut. Mean patient age was 44.1 years (median 41.4, range 0.1-77.9 years), and they were predominantly female (74%, 148/200). The majority of patients, 91% (181/200), did not have a personal history of cancer, meaning that most questions were about the patient's family history. A known cancer gene pathogenic variant was mentioned in 7% (14/200), and a family history of a known cancer gene pathogenic variant was mentioned in 12% (23/200) of eConsults.

The data analysis captured any cancer that was mentioned in the eConsult case (either directly in the question or in the reported family history, when provided) (Figure 1). The type(s) of cancer(s) mentioned in 187 eConsults were predominantly breast (100 cases) and ovarian (38 cases), followed by colon (30 cases) and pancreas (22 cases). Other common cancers, including prostate, were mentioned in 15 or fewer eConsults. About a third of the eConsults (39% [73/187]) involved more than 1 cancer type in the individual or family, with 9% (16/187)

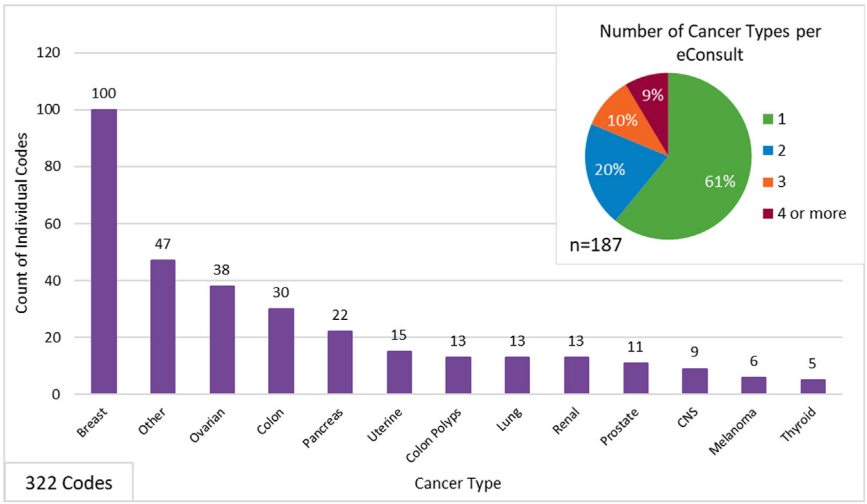

Figure 1 **Cancer type mentioned in eConsult.** eConsult, electronic consultation.

having 4 or more cancers. A specific cancer gene (eg, *BRCA1*) or HC syndrome (eg, Lynch syndrome) was mentioned in 35 eConsults.

PCP close-out survey results

The PCP survey questions and results about clinical impact and outcome from the eConsult are shown in Table 1. In about two-thirds of eConsults (65%, 129/200) PCPs indicated that they received clear advice for a new or additional course of action, and in 31% (62/200), PCPs indicated that they were able to confirm a course of action that they originally had in mind. For about one-third of eConsults (34%, 68/200), PCPs indicated that referral was originally contemplated but now avoided. Typically, this was when the PCP had a question about whether their unaffected patient, based on family cancer history, was eligible for genetic testing or enhanced screened and learned from the eConsult process that the patient was not eligible for testing. The eConsult answer would also address their screening question. Conversely, a referral was initiated after the eConsult recommendation when it had not originally been contemplated in 8% (16/200). An example of this scenario was a PCP whose patient had a paternal cousin with a *BRCA1* pathogenic variant. The PCP stated they did not think it was likely of concern for the patient given the degree of relation but was seeking insight through the eConsult. The eConsult provider recommended referral to genetics because the patient was eligible for testing.

The PCP survey questions and results about perceived education value of the eConsult response and potential for future continuing medical education (CME) topic are shown in Table 2. For the majority of eConsults (89%, 177/200), the PCP considered the eConsult valuable in guiding evaluation/management of the patient. Most (63%, 125/200) agreed that the eConsult case addressed an important clinical problem that should be incorporated into upcoming CME events. We also compared the survey results according to PCP type (whether submitted by an FP or NP) and PCP region (urban vs rural) (Tables 1 and 2). Although the numbers were too small for meaningful statistical analysis, there were some differences between FP and NP response, with 12 of 18 (67%) cases submitted by NPs originally contemplating a referral but being able to avoid it after eConsult vs 56 of 182 (31%) cases submitted by FPs. In a higher percentage of NP submitted cases the eConsult result was considered valuable (18/18 [100%]) compared with FP submitted cases (159/182 [87%]). There were no obvious differences noted between responses from urban vs rural PCPs.

Question type

Figure 2 shows the types of question asked by the PCPs, as coded by the researchers (not limited to 1 per case). Questions asked were predominantly identified as being about cancer screening (114 cases), genetic testing (107 cases), or referral to genetics (76 cases). For example, the most common eConsult was for an unaffected patient with a family history of cancer, for whom the PCP wanted to know if there was any genetic testing available, if any increased cancer screening should be initiated, and if the patient should be referred to the genetics clinic to have these questions answered. An example of a one-off question included a PCP asking about any increased cancer risk in their patient with 47,XXX syndrome. There were also questions around surgical or medical management (27), such as use of hormone replacement therapy or

Table 1 Survey results regarding eConsult clinical impact and outcome among PCP type (FP and NP) and PCP region (urban and rural)

Survey Topic (Bold) and Response Option	PCP Type		PCP Region		
	FP ($n = 182$), *n* (%)	NP ($n = 18$), *n* (%)	Urban ($n = 178$), *n* (%)	Rural ($n = 22$), *n* (%)	All ($N = 200$), *n* (%)
Impact of eConsult on PCP course of action					
Confirmed course of action	57 (31)	5 (28)	57 (32)	5 (23)	62 (31)
Received clear advice for new/additional action that will be implemented	118 (65)	11 (61)	114 (65)	15 (68)	129 (65)
Received new advice that cannot be implemented	2 (1)	0 (0)	2 (1)	0 (0)	2 (1)
Other	5 (3)	2 (11)	5 (3)	2 (9)	7 (4)
Outcome of eConsult					
Referral contemplated but now avoided	56 (31)	12 (67)	60 (34)	8 (36)	68 (34)
Referral contemplated and still needed	64 (35)	3 (17)	62 (35)	5 (23)	67 (34)
Referral not contemplated and still not needed	39 (21)	1 (6)	35 (20)	5 (23)	40 (20)
Referral not contemplated but now initiated	15 (8)	1 (6)	13 (7)	3 (14)	16 (8)
Other	8 (4)	1 (6)	8 (4)	1 (5)	9 (5)

eConsult, electronic consultation; *FP*, family physicians; *n*, number of survey responses. *NP*, nurse practitioner; *PCP*, primary care practitioner.

Table 2 Survey results regarding perceived educational value of eConsult among PCP type (FP and NP) and PCP region (urban and rural)

Survey Topic (Bold) and Response Option	PCP Type		PCP Region		
	FP ($n = 182$), n (%)	NP ($n = 18$), n (%)	Urban ($n = 178$), n (%)	Rural ($n = 22$), n (%)	All ($N = 200$), n (%)
Helpfulness/educational value of eConsult response					
Very valuable	119 (65)	14 (78)	120 (67)	13 (59)	133 (67)
Valuable	40 (22)	4 (22)	38 (21)	6 (27)	44 (22)
Neutral	22 (12)	0 (0)	19 (11)	3 (14)	22 (11)
Less valuable	0 (0)	0 (0)	0 (0)	0 (0)	0 (0)
Minimal	1 (1)	0 (0)	1 (1)	0 (0)	1 (1)
eConsult addresses a clinical problem for future CME events					
Strongly agree	41 (23)	6 (33)	41 (23)	6 (27)	47 (24)
Agree	71 (39)	7 (39)	71 (40)	7 (32)	78 (39)
Neutral	62 (34)	3 (17)	57 (32)	8 (36)	65 (33)
Disagree	5 (3)	2 (11)	7 (4)	0 (0)	7 (4)
Strongly disagree	3 (2)	0 (0)	2 (1)	1 (5)	3 (2)

CME, continuing medical education; *eConsult*, electronic consultation; *FP*, family physicians; *n*, number of survey responses; *NP*, nurse practitioner; *PCP*, primary care practitioner.

contraceptives in patients with HC syndromes or family history of breast cancer. PCPs were also noted to have used eConsult to ask questions around private pay/direct-to-consumer genetic testing (10 cases). Finally, there were 11 cases in which a question was about a known cancer pathogenic variant in a patient. Often, this was in relation to a patient being new to the PCPs practice or having had genetic testing many years ago. There was only 1 case each about interpretation of a germline genetic test result or somatic test result.

Recommendation type

Figure 3 shows the types of recommendations provided by the genetics specialist answering the eConsult (not limited to 1 recommendation per case) as coded by the researchers. Most commonly, a cancer screening recommendation for the patient was provided by the specialist (142 eConsults) and sometimes for the patient's family member (12 cases). Often, genetic referral was recommended for a family member rather than the patient (because the relative would be the more informative person to assess/test) (58 cases). In 46 cases, a referral was recommended to be redirected to a nongenetics specialist. This was most commonly a gastroenterologist in relation to consideration of pancreatic cancer screening or to a gynecologist regarding consideration for risk-reducing oophorectomy or type of oral contraceptive. Less commonly, referral to an oncologist, hematologist, cancer screening specialist (eg, radiologist) or menopause clinic was suggested. There were also single occurrences of suggested referral to thoracic surgery, nephrology, and ophthalmology. Referral to the genetics clinic (for assessment or testing) was recommended for the patient in 46 cases or directly to the High-Risk Ontario Breast Screening Program (HR-OBSP) (for assessment) in 41 cases.

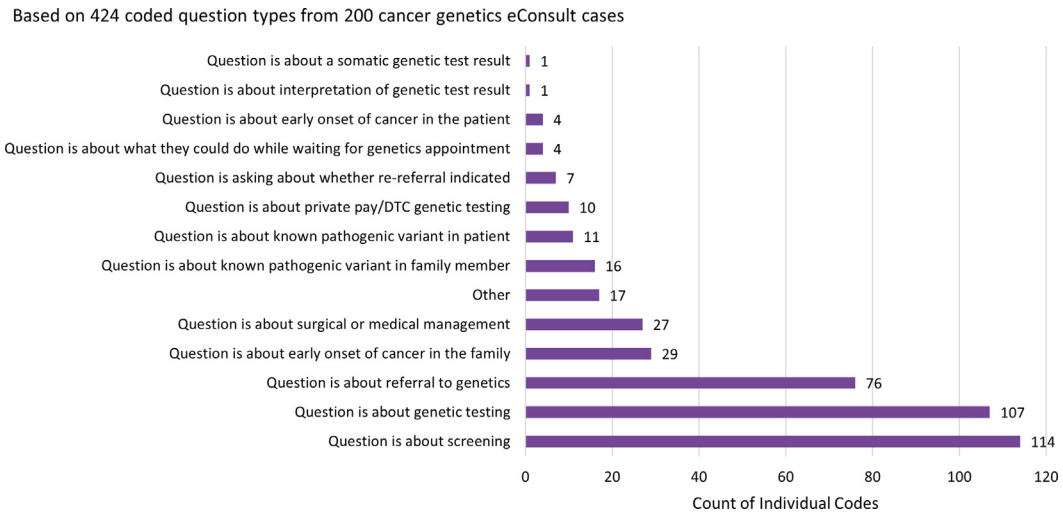

Figure 2 Question type asked by primary care practitioners. DTC, direct-to-consumer.

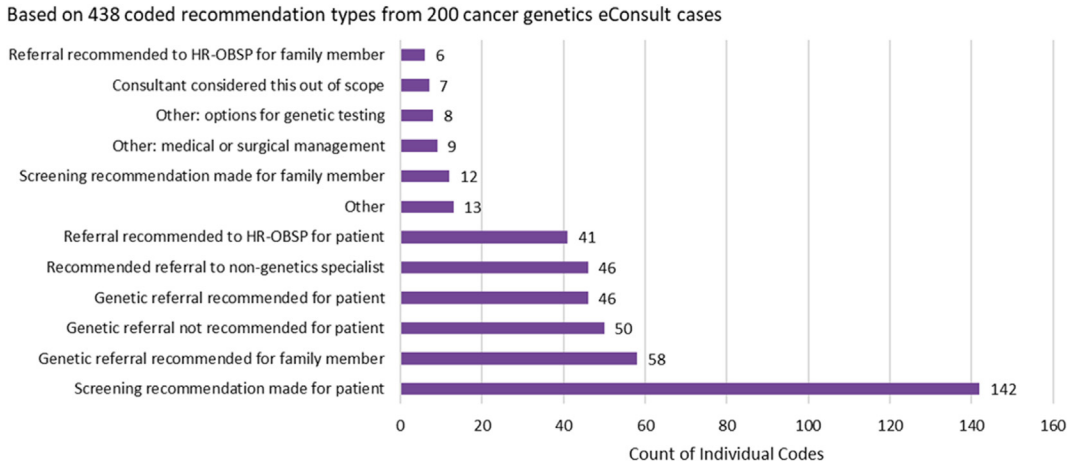

Figure 3 Recommendation type provided by genetics specialist. HR-OBSP, High-Risk Ontario Breast Screening Program.

Knowledge gaps

Three main themes were identified as potential PCP knowledge gaps based on the answers provided by the medical geneticists (not limited to 1 answer) (Figure 4). The first was awareness of available guidelines and referral criteria because the eConsult frequently addressed Ontario cancer screening guidelines, cancer genetics referral criteria, and HR-OBSP referral criteria (112, 100, and 71 cases, respectively). A second theme was around the understanding of genetics principles, such as characteristics of a family at high risk of HC syndrome, the optimal approach of performing genetic testing on an affected individual first, patterns of inheritance, consideration of ancestry, reduced penetrance, and not testing children for adult-onset conditions (94, 83, 28, 25, 5, and 2 cases, respectively). A third theme was recognition that eConsult could be a useful tool for addressing less-common PCP questions that might not typically be considered appropriate for referral by the PCP (or even accepted by the cancer genetics clinic) via the traditional route. Included in this category was explaining when to rerefer to the genetics clinic (19 cases), how patients can access genetic testing if not eligible through provincial funded care (private pay) (11 cases), the meaning of variants of uncertain significance (2 cases), the difference between somatic and germline testing (1 case), and Ethical, Legal, and Social Issues principles, such as insurance concerns around genetic testing (2 cases). The most common

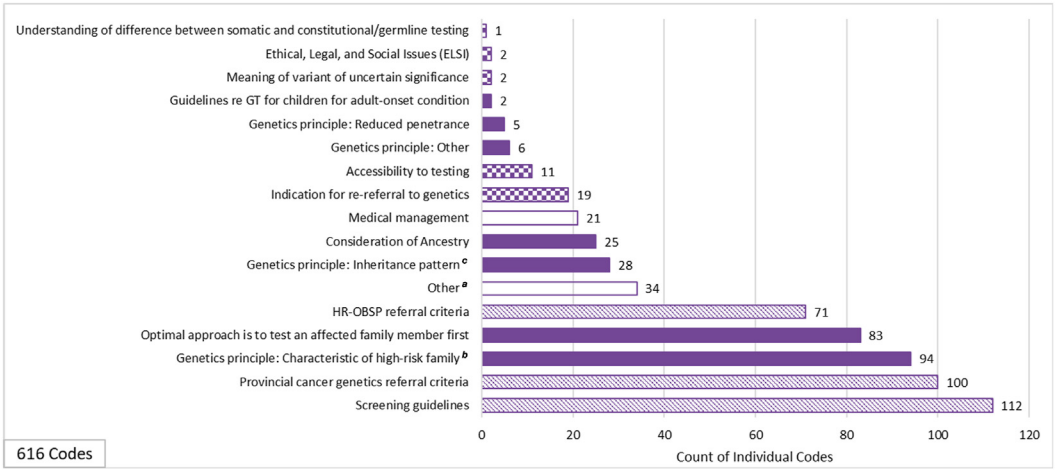

Figure 4 Potential knowledge gaps identified by specialist providing eConsult depicting 3 themes: (1) awareness of available guidelines and referral criteria (hatched), (2) understanding of genetics principles (solid), and (3) less-common primary care practitioner (PCP) questions that might not be asked by typical referral routes (checkered). [a]The most common examples of knowledge/learning gap coded as Other included what information to collect for genetic risk assessment and/or include on referral. Breast cancer risk in male gene carriers and screening for transgendered persons was also mentioned. [b]eg, Multiple generations affected, early onset, multiple primary cancers, bilateral disease, rare cancer types, certain patterns of cancer type. [c]eg, Inheritance of a breast cancer risk gene through males, autosomal dominant vs recessive, reduced penetrance. eConsult, electronic consultation; GT, genetic testing; HR-OBSP, High-Risk Ontario Breast Screening Program.

examples of knowledge/learning gap coded as "Other" (34 cases) included what information to collect for genetic risk assessment and/or include on the referral. Breast cancer risk in male gene carriers and screening for transgendered persons was also mentioned.

Discussion

Implementation of electronic consultations worldwide has been shown to improve access to patient care and decrease the need to refer patients to already limited specialist resources, with an overall trend toward positive impact on access measures, acceptability, cost, and reported PCP and consultant satisfaction.[9,11,16] This study shows similar findings specifically related to HC. Notably, in about one-third of the cases, referral was avoided and in nearly two-thirds, PCPs indicated that the eConsultation led to a new or additional course of action. The demonstrated utility of this service is especially important given the rapidly increasing rates of referral to genetics clinics for questions around HC.[17] This could potentially decrease the need for assessment for referral eligibility at the genetics clinic level in addition to providing education about HC.

Carroll et al[9] previously provided insights into genetics education needs for PCPs and highlighted the importance of planning appropriate CME accordingly. Our study demonstrated that most PCPs considered the HC eConsult response valuable in terms of guiding evaluation/management of the patient and agreed that the eConsult case addressed an important clinical problem that should be incorporated into upcoming CME events. Uniquely, we were able to identify specific common PCP knowledge gaps, such as lack of awareness of available criteria for HC referral, high-risk breast cancer assessment and cancer screening, lack of understanding of genetic principles, and usefulness of eConsult as a tool for addressing questions not typically considered for referral to (or accepted by) HC clinics.

Alternative models of care such as eConsult can be used at multiple stages of the genetic testing process.[18] The eConsult service for HC genetics in our region is open to FPs, NPs, and medical specialists, including oncologists. Our results demonstrated that only FPs and NPs are using this service at present. Lack of specialist use of eConsult could be related to clearer eligibility criteria for genetic testing for patients with cancer and better understanding of HC syndromes as they relate to their area of specialty (eg, breast surgeons and gynecologists who would be expected to have more patients with BRCA gene pathogenic variants in their practice than an FP). Mainstreaming of genetic testing to oncologists began in our region in January of 2022 (encompassing only the final year of the study time frame). As more nongenetics professionals begin to order genetic testing in our region, the eConsult service may have greater uptake from specialists.

Most of the eConsults in our study were initiated by an FP (91%) vs a NP (9%) and from a PCP situated in an urban (89%) setting vs rural (11%). The study question was not designed to address utilization differences between these groups for HC; however, this has been looked at previously for overall Ontario eConsult data.[19,20] In a study looking at 4260 eConsults (3686 from FPs and 574 from NPs), Liddy et al[19] showed that NPs were more likely to report that the eConsult led to a new course of action and reported slightly higher levels of perceived value. Guglani et al,[20] looked at 72,948 eConsults and found that those from rural and urban PCPs had similar results regarding when specialist consult was avoided, and whether PCP felt they had received good advice for a new/additional course of action. Future areas of research could look at similar data specific to HC eConsult services.

Most of the eConsult questions to our service were about whether a patient was eligible for genetic testing or increased cancer screening, and the PCPs appeared to be using eConsult as means of asking whether their patient should be referred to the genetics clinic as opposed just initiating the referral. Another reported use of eConsult is for questions around posttest screening and management options for patients found to harbor a cancer risk variant.[18] In our study, there were only 11 instances out of 200 eConsults in which the eConsult question involved a known cancer gene variant and only 1 instance of a question being about interpretation of a germline genetic test result. Often, this was in relation to a patient new to the PCPs practice. That the eConsult service is not being used more for posttest questions may be due to the current ability of our region to provide timely genetic services to the patient with a meaningful genetic test result and summary letter copied to their PCP. As genetic testing begins to be ordered more by nongenetic specialists and if population genetic testing is implemented, there will likely be increased questions around posttest results, and tools such as eConsult could become even more valuable.

The majority of eConsults in this study were centered around questions regarding breast and ovarian cancer, likely because of the decades-long understanding of hereditary risk for these 2 cancers in association with the relatively well-understood *BRCA1* and *BRCA2* genes. An interesting observation from our work is the small number of eConsults that mentioned prostate cancer. In recent years, the hereditary contribution to this cancer, from genes that overlap breast and ovarian cancer risk, has been demonstrated,[21] and family history of prostate cancer, along with breast and/or ovarian cancer, has been incorporated as an indication for referral for genetic testing for hereditary breast and ovarian cancer gene panels in Ontario and elsewhere.[22-24] Another observation is the number of eConsults regarding pancreas cancer, which may reflect the increasing awareness of heritable contribution to this disease and potential for beneficial screening in high-risk individuals.[25] Similar to prostate cancer, pancreas cancer has been added to genetic testing referral criteria in recent years.[23,24] Finally, there were many

questions addressing more than 1 cancer type in the family. Recognition of the specific type(s) of cancer(s) (beyond breast, ovarian, and colon) included in the current genetic testing referral criteria would be a key focus for PCP education.

Carroll et al[4] showed that PCPs report key tasks of traditional genomic medicine, such as eliciting family history, identifying patients with a genetic condition, deciding who should be offered genetic referral, and knowing where to refer are already part of their practice, but their confidence in performing these tasks is low. Our study suggests similar lack of confidence related to HC with some of the most common questions and recommendations being around eligibility for genetic testing or referral. In the study by Carroll et al,[4] popular suggestions for how to integrate genomic medicine into primary care practice included having contact with a local genetic counselor (66%) or a buddy system with a geneticist (51%). We have shown that eConsult is one tool that can successfully connect PCPs directly with HC genetic specialists, including from rural settings and remote Northern communities.

After lack of awareness of available criteria for HC referral, the next most common knowledge gap identified in this study centered around eligibility for breast cancer risk assessment. The High-Risk Ontario Breast Screening Program[26] organizes screening with annual mammogram and breast MRI for women aged 30 to 69 who are deemed to be at high-risk for breast cancer. This includes known carriers of breast cancer risk genes (eg, *BRCA1* and *BRCA2*) or women who are 8 years post receiving chest wall radiation under age 30. It also includes women who are calculated to have a lifetime risk of developing breast cancer that is 25% or greater by the CanRisk and/or Tyrer-Cuzick (also known as International Breast Cancer Intervention Study) risk models.[27,28] Similar to other cancer genetic referral criteria, eligibility for breast cancer risk assessment is determined based on personal and family cancer history. The program requires this risk to be calculated by a genetics clinic, and women can be referred directly to the HR-OBSP or local genetics clinic for this assessment. Despite the HR-OBSP program being available in Ontario since 2011 and available information, including access to the requisition for referral, on a provincial website,[26] there continues to be a lack of awareness. Our study demonstrates the need for ongoing awareness campaigning about where to access referral criteria, but it is also likely due in part to complex family history-based referral criteria and PCP challenges with taking an appropriate family history.

Obtaining a detailed family health history is the gold standard for risk assessment for HC and other common diseases, but there are well-recognized constraints on PCP time, and patient-facing platforms have been shown to be equal or better quality than even genetic counselor interviews.[29] The potential benefit of incorporation of family-history-taking tools into family medicine practice has been reviewed,[29-31] but further study is needed to provide evidence of direct benefits or recommendation for any particular tool. Ideally, perhaps with AI-driven technology, the electronic medical record (EMR) would be able to pull information from the patient's personal and family health history to flag those who could benefit from a genetic assessment based on local referral criteria. These types of advances are already in process. Kaphingst et al[32] recently demonstrated through the Broadening the Reach, Impact, and Delivery of Genetic Services randomized controlled trial how a chatbot tool can be used to provide pretest genetic services to patients identified through the hospital EMR as eligible for genetic testing. In that study, 3073 patients who were eligible for cancer genetic evaluation were randomized to chatbot group (1554) or enhanced standard-of-care control group (1519). Results suggested equivalence in the primary outcomes, which were completion of pretest cancer genetic services and completion of genetic testing. There were limitations to the Broadening the Reach, Impact, and Delivery of Genetic Services study, with genetic testing procedures not being fully automated. Genetic counseling assistants still contacted every patient, even in the chatbot group, to confirm testing decisions. The study authors also noted that most of their participants were White and female and included few Spanish-speaking patients despite offering service in this language. This type of AI-driven technology will also be limited by human fallibility because it relies on what was originally inputted into the EMR to identify eligible patients and determine what genetic testing to offer and can therefore be biased by missing or improperly labeled data. Special attention will be needed to ensure that the use of AI does not place further barriers to access for racialized, uninsured, or other equity-deserving people.

Another major knowledge gap that emerged from our study was around the potential limitations in the understanding of HC genetic principles with characteristics of a high-risk family and the optimal approach being to test an affected individual first predominating. Recognition of these principles starts with being able to take and document an appropriate family history to identify red flags for HC predisposition. Prior work in this area led to development of a website containing evidence-based resources, including point of care tools on how to identify high-risk families and information on specific HC conditions such as Lynch syndrome and hereditary breast and ovarian cancer associated with *BRCA1* and *BRCA2* genes.[33] Many other online education programs are available for PCPs.[4]

A final theme that was identified was the recognition that eConsult could be a useful tool for answering PCP questions that might not normally be considered for referral by the PCP (or even accepted by the cancer genetics clinic) via the traditional route. This included information on how patients can access genetic testing when not eligible through provincial criteria (private pay) and interpreting prior genetic test results (variants of uncertain significance, explaining the difference between somatic and germline), and a few cases in which questions were answered about Ethical, Legal, and

Social Issues principles and whether genetic testing for adult-onset HC should be offered to children. There were very few eConsults around these types of queries. It could be that PCPs do not think of eConsult as a place to ask such questions, or it may relate to the deeper issue of overall lack of PCP awareness and understanding of genomic medicine. Wider educational initiatives are needed across the spectrum from medical student to practicing physician. Rubanovich et al[34] (2018) reviewed existing education initiatives in the United States and found emerging themes that included immersive and experiential learning, interdisciplinary and interprofessional education, and electronic- and web-based approaches, all of which could also be targeted specifically to HC initiatives.

Future directions

Our study demonstrated that eConsult for HC is an effective tool for connecting PCPs with clinical geneticists in an urban setting but also rurally and as far away as Nunavut, where the major populated center can only be accessed by a 3-hour plane ride from the closest genetics clinic. Recruitment of more PCPs from rural settings or those who serve remote areas has the potential to identify through eConsult those patients who would be most likely to benefit from travel to the larger centers for cancer screening. Recruiting other GHPs, such as genetic counselors, to provide eConsult as the service expands could also be considered. Whether the eConsult service can significantly affect wait times for HC specialists, improve costs to the health care system, or reduce barriers to care for minoritized populations will also be important future areas of research. Although this study identified potential PCP knowledge gaps around HC, future studies looking at outcomes of specific interventions to improve knowledge are necessary.

Potential limitations

Our study focused on eConsults in one region of Ontario, and results might not be generalizable elsewhere, particularly in locations that allow for genetic testing and the use of screening tools to be done outside of genetics clinics or in places that do not have government-funded health care During the time frame of the study (October 2016 to December 2022), there were provincial updates to hereditary testing eligibility (April 2021) and high-risk breast cancer assessment criteria (April 2023) and an addition of a policy statement on the screening of trans people within the Ontario Breast Screening Program (September 2021) with presumed wide circulation to stakeholders. It is unknown whether the dissemination and access to these documents for PCPs would have lessened some of the perceived knowledge gaps identified in our study. Finally, the qualitative analysis of this study was performed by 2 clinical geneticists, 1 genetic counselor, and 1 PCP. It is possible that results would have been different if analysis was performed with a larger PCP presence.

Conclusion

This study showed that eConsult is an effective tool for connecting PCPs with HC specialists and has direct impact on patient management. We also identified knowledge gaps around PCP understanding of HC genetics, which can be used to enhance future CME and drive innovations to better serve this patient population.

Data Availability

The data that support the findings of this study are available on request from the corresponding author.

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Author Contributions

Conceptualization: A.R., J.C.C., S.M., M.C., D.G., E.K., C.L.; Data Curation: A.R., J.C.C., S.M., M.C., D.G., C.L.; Formal Analysis: A.R., J.C.C., S.M., M.C.; Funding Acquisition: E.K., C.L.; Investigation: A.R., J.C.C., S.M., M.C.; Methodology: D.G., A.R., J.C.C., C.L.; Project Administration: D.G.; Validation: D.G.; Visualization: A.R., D.G., J.C.C.; Writing-original draft: A.R., D.G.; Writing-review and editing: A.R., J.C.C., D.G., S.M., M.C., E.K., C.L.

Ethics Declaration

This study was approved by the Ottawa Hospital Research Ethics Board.

Conflict of Interest

Clare Liddy and Erin Keely are cofounders of the Champlain BASE eConsult Service, but they have no commercial

interest in the service and do not retain any proprietary rights. Dr Erin Keely is the Executive Director and Dr Liddy is the Evaluation Lead for the Ontario eConsult Centre of Excellence and both receive salary support from Ontario Health. Dr Alison Rusnak provides eConsult service for hereditary cancer (~6-8 per month) as a specialist for which she is reimbursed. All other authors declare no conflicts of interest.

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