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Use of eConsult to enhance genetics service delivery in primary care: A multimethod study



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ABSTRACT

Purpose: Electronic consultation (eConsult) is a freely-available secure online platform connecting primary care providers (PCPs) to geneticists. Our purpose was to determine whether eConsult is effective in improving genetics service delivery in primary care.

Methods: PCP questionnaires regarding eConsult's utility, geneticists' tracking form assessments of eConsult type and appropriateness, and geneticists' interviews on implementing eConsult were carried out.

Results: In 2 regions of Ontario, Canada, from January 2019 to June 2020, there were 305 genetics eConsults. For 169 (55%), PCPs indicated receiving good advice for a new course of action; for 110 (36%), referral was now avoided; and for 261 (86%), eConsult was perceived valuable for patient management. Of the 131 geneticist-completed tracking forms, cancer questions were most common (68, 52%). For 63 (48%), geneticists disagreed/strongly disagreed PCPs should know the answer to the referral question. From the interview data, it was observed that geneticists described eConsult positively and suggested how it might improve access and efficiencies if integrated into genetic service delivery. Dealing with eConsults virtually could reduce waitlists, and suggesting appropriate investigations for PCPs could improve efficiencies.

Conclusion: eConsult offers a potential solution for receiving timely genetics advice and avoiding unnecessary patient referrals, however, greater effect on access and wait times will need systematic integration into PCP and geneticist practice.

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Introduction

The benefits of genetic and genomic information are increasingly being recognized, particularly in the areas of personalized diagnosis, prognosis, risk assessment, and management. The integration of genomic medicine into primary care has been challenging owing to barriers, including lack of awareness of genetics services, lack of knowledge such as when and how to refer to genetics, perceived lack of clinical utility of some genetic tests, and lack of time to incorporate genetics into clinical practice.¹⁻⁵ This is compounded by the finding that primary care

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providers (PCPs) express unfamiliarity with genetics specialists and clinics, many having never referred to or connected directly with them.^{1,5,6} Genetics clinics declining PCP referrals, often owing to unclear referral guidelines, has left PCPs uncertain of the referral process,¹ with both underand over-referring to genetics by PCPs being described.⁵ Geneticists have outlined a key role for PCPs in genomic medicine both now and in the future, which requires genetics health professionals and PCPs to work together more effectively.⁵

In addition to this is a shortage of genetics professionals internationally to meet the increasing demand for genomics services,⁷ resulting in long wait times⁸ and inequity in service delivery with PCPs reporting that genetic centers are often too far away for patients to access.^{3,6,9} Wait times for nonurgent, nonprenatal genetic referrals in Ontario, Canada, range from 1 month to 2 years.¹⁰ Wait times for other specialties (not including genetics) in Ontario were reported in 1 study to be a median of 42 days with 75% of all patients seen within 80 days and all within 760 days.¹¹

Health care systems have been described as lacking the "structures and processes to facilitate genetic medicine into practice"² with a need for "structured and targeted implementation strategies for genetic services."²

Many roadmaps have been proposed for implementing genomic medicine into practice.^{4,12-14} Development of referral and testing guidelines, point of care tools, enhancement of the relationship between geneticists and PCPs, and use of telehealth are among the suggestions.

We are proposing that electronic consultation (eConsult) might be a way to help address excessive wait times for genetics consultation, problems of geographic access, and scarce genetics specialist resources, providing answers to PCPs' questions about their patients' genetic concerns and potentially also with educational benefits. eConsult is defined as an "asynchronous, directed communication over a secure electronic medium that involves sharing of patientspecific information and (seeks) clarification or guidance regarding clinical care."15 Genetics questions are rarely urgent and can therefore be answered when convenient for the geneticist. A systematic review of eConsult systems between PCPs and specialist physicians included 36 studies, most in the United States and focused on single-specialty services (most commonly dermatology). They found that referrals were avoided in between 12% and 84% of eConsults with satisfaction ranging from 78% to 93%.¹⁵ A pilot study of the use of eConsult specifically for genetics questions showed that approximately one-third resulted in a planned patient referral being avoided.¹⁶

Our objective was to determine whether eConsult is an effective method for improving genetics service delivery in primary care. We wanted to determine PCPs' perceptions of the utility and value of genetics/genomics eConsults and geneticists' assessment of the type and appropriateness of the eConsults they received from PCPs. We also aimed to explore geneticists' experiences with the implementation of an eConsult program, their satisfaction and concerns, and views on the value of the program and its sustainability.

Materials and Methods

This study built on work previously carried out in the Champlain health region of Ontario, Canada, using the Champlain Building Access to Specialists through eConsultation (BASE) eConsult Service.^{15,17-19} This eConsult service is freely available to any health care provider and patient in the province of Ontario. The Champlain BASE eConsult Service is currently used by 3786 PCPs across the province.²⁰

Description of eConsult

eConsult is a form of asynchronous communication in which PCPs and specialists, in this case geneticists, can communicate directly about a patient. PCPs submit a patient question via a secure, web-based portal. They can attach additional information (eg, photo, test results). The case is assigned to a specialist on the basis of availability. The specialist receives an email notification prompting them to access the case via the secure site. They are expected to provide an answer within 1 week. They can reply to the question, request additional information or recommend a patient referral, and advise the PCP on other matters such as additional tests or actions to be completed before a face-toface specialty care appointment. PCPs ultimately decide how to apply the specialist's suggestions to the care of their patients. In the Ontario eConsult program used in this study, geneticists are compensated for their time on each eConsult through a provincial program.¹⁷⁻¹⁹

Study setting

This study took place in 2 regions of Ontario, Canada (the Champlain and Mississauga Halton health regions). The Champlain region has a population of approximately 1,292,639, of which 51% are female, 49% are male, with a median age of 41.5 years.²¹ This region has 1644 family physicians, of whom 57% are female and 45% are aged 50 years or above,²² and 9 clinical geneticists at the time of the study. The Mississauga Halton region has a population of approximately 1,164,740, of which 51% are female, with median age of 40 years.²³ This region has 1204 family physicians, of whom 53% are female and 51% are aged 50 years or above,²² and 4 clinical geneticists.

Study design

This study used quantitative and qualitative components. Quantitative methods enabled evaluation of the eConsult genetics service from all users over the study time frame and location. Interviews offered the opportunity to gain a deeper understanding from geneticists about the operation of the service and its benefits and limitations. The first step was to raise awareness of the availability of eConsult for genetics consultations to PCPs (family physicians and nurse practitioners) in the 2 regions. This was carried out by notifying PCPs of the availability of genetics eConsult through the usual channels of communication between the Champlain BASE eConsult Service and enrolled PCPs (ie, emails, newsletters).

Quantitative component

Quantitative data were provided by the Champlain BASE eConsult program from the PCP eConsult close-out survey (Appendix A), which was a regular quality improvement component routinely completed after an eConsult and included (1) a description of the outcome of the eConsult for the patient, (2) the referral outcome, (3) the overall value in management, (4) whether the eConsult addressed an important problem that should be incorporated into continuing medical education (strongly disagree to strongly agree), and (5) an open-ended question for additional feedback. The data set also included provider and patient demographics and eConsult response times.

In addition, geneticists who were participating in the Champlain and Mississauga Halton eConsult programs received an emailed invitation to participate in the study. Participating geneticists answered PCPs' questions about genetics concerns related to their patients' care through eConsult. After each eConsult, they were asked to complete an eConsult checklist (Appendix B) to identify the type of question asked and whether they would have expected the PCP to have known the answer to the eConsult question. Participating geneticists received \$300 Canadian dollars at the end of the study to compensate them for time spent completing these data sheets. Ontario geneticists are compensated through provincial level funding for the eConsult itself.

Qualitative component

A qualitative research assistant conducted individual interviews with participating geneticists at 2 points in time: 3 months after the commencement of the study to capture their early experience and between 9 and 12 months after the commencement of the study. A semistructured interview guide was developed on the basis of the literature and experiences of the research team including questions exploring satisfaction with, value of, and implementation of the service and suggestions for change. The 9 to 12 month interview guide was modified to elicit feedback on several themes found in the earlier interviews with participating geneticists to confirm or further refine these themes. (interview guides—Appendices C and D). The interviews, which were about 30 minutes in length, were conducted by phone, audio recorded, and transcribed. At the end of the study, participating geneticists received \$150 Canadian dollars to compensate them for participating in the 2 qualitative interviews.

Data analysis

Quantitative

Quantitative data were analyzed using SPSS (IBM) statistical software. Data from surveys were analyzed descriptively using frequency distributions and means analysis.

Qualitative

Transcripts from geneticists' interviews were imported using NVivo software to aid in data organization, review, coding, and analysis and to facilitate an exploration of trends and themes that emerged from the data. A thematic analysis was conducted using the constant comparative method.²⁴ Codes were developed both from the topic domains in the interview guide and inductively from the data. Data were analyzed, and themes were elicited within and across interviews. The thematic analysis involved a process of initially reading each transcript to gain an overall sense of the data, then rereading the transcripts to identify major topics or issues.²⁵ The process of identifying codes was initially carried out by 2 team members (T.M. and J.C.C.), with 1 team member (T.M.) coding the remaining transcripts once codes were agreed upon. After the transcripts were coded, the working group met to ensure that the emerging themes were supported by the data. The process of coding also involved discussions of the issues identified in the data and was iterative, adding new interview data as they were received. As the data analysis moved to the analytic level, relationships among the themes and issues were identified. The research team met regularly to discuss the data and came to a consensus on the major themes.

Results

PCP portion of the study

PCP demographics

Of the 205 PCPs who submitted a genetics eConsult during the study period, we were able to find demographic information on 179 physicians registered with the College of Physicians and Surgeons of Ontario (CPSO). The remaining 26 were nurse practitioners. Participating physicians had a median of 14 years in practice (range 2-40), 142 of 179 (79.3%) were female, and 157 of 178 (88.2%) practiced in an urban community.

PCP assessment of the genetics eConsult service

Over the 18 months of the study (January 1, 2019 to June 30, 2020), there were 305 genetics eConsults submitted by 205 PCPs. Of these, 89.2% (272) were by physicians and 10.8% (33) by nurse practitioners. Most PCPs (143/205, 69.8%) submitted 1 eConsult, with 62 of 205 (30.2%)

submitting between 2 and 5 eConsults. The majority, 97.4% (297/305), were answered by geneticists in the Champlain health region, with 2.6% (8) by geneticists in the Mississauga Halton health region. Most eConsults concerned adult patients (257, 84.3%) with 48 (15.7%) eConsults in patients under age 18. About two-thirds (207, 67.9%) of eConsults were sent by PCPs to general genetics and 32.1% (98) to cancer genetics. Geneticists spent a median of 10 minutes answering the question (SD = 7.3) and were paid a median of 33.33 Canadian dollars (SD = 24.2) per eConsult. Most eConsults (265, 86.9%) required only 1 response, meaning no further information was needed. The median time between the eConsult submission by the PCP and their receiving of their first response was 1.8 days (SD = 3.5). In 16.4% (50) cases, the responding geneticist included an attachment to their eConsult response. Table 1 shows the outcome of the eConsult for patients as reported by PCPs on the eConsult close-out survey. For more than half of the genetics eConsults, the PCPs indicated "got good advice for a new/additional course of action that I will be implementing" (169, 55.4%). More than a third PCPs (110, 36.1%) indicated that "referral was originally contemplated but now avoided at this stage." For an additional 18% (55) cases, the eConsult confirmed that referral was still not needed. In 5.9% (18) cases, referral was not originally contemplated but now initiated following the geneticist's advice. The vast majority of PCPs (261, 85.6%) indicated that the eConsult response was valuable/very valuable in guiding their ongoing evaluation or management of the patient. More than half PCPs (160, 52.4%) agreed/strongly agreed that the specific eConsult question they posed addressed an important clinical problem that should be incorporated into upcoming continuing education events.

Geneticist portion of the study

Geneticist demographics

There were 7 clinical geneticists participating in the Champlain BASE eConsult Service at the time of the study and all participated. They were all female with a median age of 42.5 (range 38-67) years and median years in practice of 11 (range 4-25) years.

Geneticists' eConsult tracking

Table 2 shows the clinical disorder that best matched the eConsult question as assessed by the participating geneticists. Data were collected by geneticists from January 1, 2019 to March 31, 2020 (early suspension due to COVID-19). Of the 131 eConsults for which we have data, 84.7% (111) were for adult problems and 15.3% (20) were pediatric. Most eConsults (68, 51.9%) were questions regarding cancer, with more than half of those being questions about breast/ovarian cancer. In 48.1% (63) of the eConsults, the geneticist disagreed/strongly disagreed that they would expect a family physician to know the answer to the

Table 1eConsult PCP close-out survey responses (N = 305)

Question: Which of the following best describes the outcome of the eConsult for your patient?

Outcome	п	%
1. Able to confirm course of action that I originally had in mind	106	34.8
2. I got good advice for a new/additional course of action that I will be implementing	169	55.4
3. I got good advice for a new/additional course of action that I am not able to implement	5	1.6
4. None of the above	25	8.2
Question: As a result of this eConsult would	you say th	at
Response	п	%
1. Referral was originally contemplated but now avoided at this stage	110	36.1
2. Referral was originally contemplated and is still needed	89	29.2
3. Referral was not originally contemplated and is still not needed	55	18.0
4. Referral was not originally contemplated but eConsult resulted in a referral being initiated	18	5.9
5. Other ^a	33	10.8
	• 1	

eConsult, electronic consultation; PCP, primary care provider.

^aA total of 24 comments, including 14 eConsults redirected (mainly from general to cancer genetics), 2 PCPs will consider referral, 2 patients declined referral, 1 PCP will discuss with patient, 1 geneticist helped triage the already made referral, 1 updated previous recommendation, 1 PCP unsure of next steps, 1 eConsult not helpful, 1 not applicable.

question posed. (2 [1.5%] strongly agreed, 25 [19.1%] agreed, 41 [31.3%] neutral, 46 [35.1%] disagreed; 17 [13%] strongly disagreed).

Findings from interviews with geneticists

In general, the eConsult service was viewed positively by the participating geneticists. Many described deriving satisfaction from completing them.

"I find generally speaking that it's a gratifying process because the providers that are asking the questions are usually inserting a note of thanks or...a positive comment about how the response was helpful." (P2)

The geneticists described many ways in which using eConsult might be of benefit to genetics service delivery, which is by increasing access and improving efficiencies. They described that eConsult has the potential to reduce some of the inequities in access to genetics consultations that might occur because of geography, either lack of availability of genetics specialists or travel challenges to genetics clinics.

"...there's a lot of cities in Ontario where...it's very difficult for a patient to get to a genetics clinic, even an outreach clinic and...there might be some doctors who just don't know where the genetic specialists are located. So it might be easier for them to just send an eConsult." (P6)

Table 2 Clinical disorder category that best matches the eConsult question as completed by participating geneticists (N = 131)

Category ^a	n	%
Cancer	68	51.9
Breast/ovarian cancer	38	29.0
Syndromes	11	8.4
Ethnicity	7	5.3
Genetic tests/technology	6	4.6
Connective tissue dysplasia	6	4.6
Mental health	5	3.8
Cardiovascular disease	3	2.3
Isolated congenital anomaly	2	1.5
Pharmacogenomics	2	1.5
Metabolic disorder	2	1.5
Direct-to-consumer personal genomic testing	2	1.5
Hemochromatosis	0	0
Other	20	15.3

eConsult, electronic consultation.

^aThree respondents selected >1 category.

The geneticists spoke of how eConsult has the potential to increase efficiencies, by determining early on if the referrals are inappropriate, potentially freeing up time on the referral wait list, if a referral is indicated possibly for an indication that was not recognized by the PCP, and if the eConsult could be answered very quickly and easily.

"...given our tremendous pressures in terms of resources and our long wait times, rather than having a patient wait for two years only to find out that their referral would have been better directed elsewhere, or that they really didn't need to be seen, it's so much better for those types of questions just to be answered in this type of way." (P6)

"I think there were a couple [of eConsults] who needed a referral to genetics, but the other ones were very straightforward answers that you could get through email from us rather than sending a referral and then waiting for us to decline their referral...so it's overall a very good use of resources." (P5)

"We've had some feedback that there are some patients that would not have been referred but because we had said...You know, here's another aspect that maybe you haven't thought of, but this person really should be referred." (P3)

Some participants suggested that eConsults could improve efficiency by suggesting investigations to be carried out by the PCP while waiting for an in person genetics consultation or perhaps instead of that consultation.

"I think the other thing it does, even for the referrals that do turn out to be necessary, it allows the specialists to recommend investigations upfront to make use of the wait time." (P2)

In addition, participating geneticists perceived that eConsult had educational value.

"And so I think it's good for learning for the physicians who are asking questions and potentially for me when I need to figure out how to answer a person's question...yes, an educational tool. And I think it should mean that they will remember and will have a better sense when to refer to genetics." (P4)

Participants expressed that they appreciated the secure platform for eConsults and that they were compensated for their time in contrast to more informal email or hallway consultations.

Challenges identified by participating geneticists included current underutilization of the eConsult service likely owing to the lack of awareness and perhaps PCPs' habit of using email to ask questions. However they also identified potential capacity issues if it became very popular. Over the year of the study, they did not perceive any effect on wait times for appointments in genetics clinics. They expressed some frustration with PCPs' lack of knowledge, expressing that "...many of the questions...were questions that I actually was surprised that I was getting in some ways, because I felt like they were things that the family physician should have known or should have been able to figure out..." (P1) They suggested planning continuing education events to answer the common eConsult questions. In addition to raising awareness of the service, this might also limit repeat questions and elevate the complexity of the questions being asked as this participant says "If there are exceptional cases or more complex cases, then this is a great service." (P3)

We asked participating geneticists at the 9 to 12 month interview to reflect on their experience with eConsult over the study duration and views about its future utility. Participants expressed that it worked well but was limited in its ability to affect triage and wait times. To really address the challenges of genetic service delivery, eConsult needed to be offered in combination with other innovative virtual and in person care modalities. "I still think that the help that it [eConsult] provides is limited in scope, that for it to be really, really helpful, it would have to be really vamped up to replace our triage system and be provided by other health care professionals like our genetic counselors." (P3) They voiced concerns about the accuracy of family history on which eConsult advice was given, on the volume of eConsults that would need to be triaged/answered to make any difference to wait lists and who would do this kind of work, genetic counselors and/or clinical geneticists. See Appendix E for additional quotes supporting these themes.

Discussion

This study showed that eConsult provides timely, efficient answers to PCPs' genetics questions, perceived by them to be clinically useful and importantly, that it is acceptable to clinical geneticists. As a result of the eConsult, in more than half cases, PCPs planned to implement a new course of action for that patient and in more than half cases, referral that was originally planned was now avoided or referral was confirmed as not needed. This is similar to the performance of multispecialty eConsult services and the genetics eConsult pilot project reported previously.^{15,16} This study adds the perspective of participating geneticists that in their view, eConsult offers the potential to increase access to genetics consultation for PCPs and patients in more remote or inadequately served areas, to achieve equity in access, and to potentially increase efficiencies and possibly decrease wait times by responding to some questions that can be answered quickly and easily. Research is needed to further explore these benefits.

This study offers new insights into the genetics educational needs of PCPs. Most of eConsults were questions regarding cancer, often breast/ovarian cancer. In about 20% of eConsults, participating geneticists indicated they would have expected PCPs to know the answer to the question posed, and in interviews, they expressed some frustration at this. PCPs' lack of knowledge regarding genetics, including genetic testing and referral guidelines, has been reported widely.⁴ Continuing education informed by the knowledge gaps identified through eConsult questions is likely to be of interest to PCPs and may over time change the nature of eConsults, increasing their complexity. Archibald et al²⁶ comment on the use of eConsult as "an innovative opportunity to enhance continuing professional development activities," addressing learning needs at the point of care and providing skills or tools to meet those needs. Geneticists may be using the eConsult as an educational opportunity. In 16% of eConsults, they included an attachment to the eConsult, perhaps outlining genetics clinic referral guidelines or resources specifically related to the question. This could be encouraged as a method of building primary care knowledge of genetics because initiatives that are interactive, case-based, and skill focused have been most successful.²⁷ Links to disease-specific primary care genetics resources could be included in the eConsult response (examples include: Genetics Education Canada—Knowledge Organization [www.geneticseducation.ca], Genomics Education Programme [https://www.genomicseducation.hee.nhs. uk/], Gen-Equip Project [www.primarycaregenetics.org]).

Clearly the challenges of genetic service delivery are more complex than can be solved by eConsult alone as described by this study. Increasing numbers of genetic tests, with resulting demand for genetic services is challenging the system and workforce capacity.²⁸ Many suggestions have been put forward to address this. Some have commented that as genetic testing becomes more mainstream, less expensive, and has higher utility, PCPs will need to provide genetic counseling and testing for some conditions.²⁹ For example, PCPs are now ordering genetic testing for hemochromatosis⁵ and first line genetic tests for the investigation of children with global developmental delay and intellectual disability (chromosomal microarray analysis and fragile X DNA analysis). In the past, these tests would have been ordered after consultation with or by a geneticist.^{5,29,30} As described by our participating geneticists, eConsult would enable geneticists to advise and educate PCPs on investigations before a genetics consultation thereby increasing efficiencies or on appropriate investigations with referral needed only if positive test results or certain criteria were met. Genetic counseling assistants could be considered to perform some of the basic clinical and administrative functions to support some of the questions emerging from eConsult such as sending resources, testing, or referral guidelines.²⁸ PCPs will need guidance on ordering genetic tests and interpreting and communicating their results.

Greenhalgh et al³¹ in their systematic review of diffusion of innovations in service organizations describe key attributes required for innovations in health service delivery to be successful: a clear, unambiguous advantage in effectiveness or cost-effectiveness; compatibility with perceived needs; simplicity in use; trialability; observability of benefits; and adaptability. Findings from this study indicates that eConsult meets most of these attributes.

Limitations

This study looked at eConsults provided by a small number of geneticists in only 2 regions of Ontario, Canada; however, its strength is that we used different methods of data collection describing eConsult outcomes as well as geneticists' perceptions of the service. Participating PCPs were mostly female and urban, a higher percentage of female PCPs than in the regions. This may reflect that more outreach about the availability of genetic eConsults is needed. However, 1 study that looked at who used eConsult showed that neither sex nor proximity to specialists were found to explain usage.³² Only length of time in practice was predictive, with being out of medical school and an additional 10 years estimated to decrease the probability of ever using eConsult by 5% points.³² The payment system for eConsult in Ontario may be somewhat unique, however payment models are being explored in countries such as the United States to expand the adoption and use of eConsult.^{33,34} More research is needed to determine if over time, the eConsult service has an effect on wait times and results in more appropriate referring.

Conclusion

eConsult offers a potential solution to receiving timely genetics advice given the increasing demand for genetic consultation and testing, as well as avoiding unnecessary patient referrals, however, its benefits are not yet fully realized. Overall, geneticists are positive about eConsult and PCPs generally appear to be asking appropriate questions. Our findings highlight the importance of further work including raising awareness of the eConsult service and its value, monitoring geneticists' workload generated from the service, planning appropriate continuing education on the basis of identified needs, looking at cost-effectiveness, measuring whether expected change outcomes are realized such as increasing equity and access, and reducing wait times for genetic consultation and increasing appropriate consultations, making use of wait times to initiate first line investigations and acceptability to PCPs and geneticists. For greater effect on access and wait times, it will need more systematic integration into referral workflow of PCPs and geneticists.

Data Availability

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

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Ethics Declaration

This study was approved by the Sinai Health Research Ethics Board (REB). All institutions involved in human participant research received local REB approval. Informed consent was obtained from all participants as required by the REB.

Conflict of Interest

C.L. and E.K. are cofounders of the Champlain BASE eConsult Service, but they have no commercial interest in the service and do not retain any proprietary rights. As Coexecutive Directors of the Ontario eConsult Centre of Excellence, they receive salary support from the Ontario Ministry of Health. E.K. answers occasional electronic consultations (less than 1 per month) as a specialist through the service for which she is reimbursed. All other authors report no conflicts of interest.

Additional Information

The online version of this article (https://doi.org/10.1016/j. gim.2022.07.003) contains supplementary material, which is available to authorized users.

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