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ARTICLE

Processes and outcomes from a clinical genetics e-consultation service managed by a primary care physician champion

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ABSTRACT

Purpose: Timely access to clinical genetics consultations remains a barrier to timely genomic medicine services, which new service delivery models might help address.

Methods: We implemented a genetics electronic consultation (eConsult) service staffed by a primary care physician (PCP) champion, supervised by genetics specialists. Chart reviews from July 2018 to January 2022 examined categories of questions received, e-consultant's recommendations, and outcomes of any conventional genetics referrals. Providers were surveyed on likelihood of ordering a conventional genetics consultation before eConsult and satisfaction with eConsult responses.

Results: Conventional genetics consultation was recommended for 338 out of 462 (73%) eConsults received, among whom 254 out of 338 (75%) had an order placed for a conventional consult by the provider requesting the eConsult. Among all 462 eConsults, including in cases which conventional consult was not recommended, 279 (60%) were referred for conventional genetics consultation, of which 171 out of 279 (61%) completed a consult. Of these, 122 out of 171 (71%) were recommended for genetic testing, and 84 out of 122 (69%) completed testing. The genetic testing of 23 out of 84 (27%) patients identified informative actionable findings. Supervising genetics specialists made substantive revisions to PCP draft responses for only 8% (36/462) of eConsults.

Conclusion: This case series demonstrates that a PCP champion eConsult model can feasibly triage and respond to genetics questions with PCP-relevant content and yield high provider satisfaction. Such a model warrants further evaluation as an addition to the genetic services of health care systems.

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Introduction

Electronic consultations (eConsults) are emerging as an important tool for primary care providers (PCPs) to gain direct access to expert specialty care for their patients.[1–6] eConsults are defined as asynchronous, consultative, provider-to-provider communication within a secure web-based platform, which allows for specialty responses to focused questions regarding patient clinical management.[4] eConsults offer quick access to recommendations for diagnosis and treatment that can often be completed by the PCP with guidance from specialty care.[2,7–9] eConsult services are seen as a tool that health systems can use to achieve the desired outcomes of efficient consultant response time, decreased clinical wait times for patients, avoidance of unnecessary conventional in-person consultations, and increased PCP and specialty provider satisfaction rates.[2,3,10–13] eConsult services for specialties such as endocrinology, psychiatry, urology, and cardiology have previously been described as successful in achieving these outcomes.[2,3,7,9,13,14]

At the same time, there is little information on the processes and outcomes of genetics eConsult services. There are not enough medical geneticists or genetic counselors available in the workforce to meet the growing demand for clinical genetics referrals.[15–18] Given this worldwide shortage of trained genetics experts, coupled with the growing understanding of and interest in the impact of human genetic variation on health, there is an increased need for an efficient and effective method for non-genetics professionals, especially PCPs, to access timely clinical genetic advice for their patients. Very few health systems have described their experiences with genetics eConsult services managed by trained genetics professionals.[19–21] We have implemented a genetics eConsult service managed by a PCP champion. Here, we describe the outcomes and impact on patient care of the first 462 eConsults received since the initiation of this service.

Methods

Setting

Mass General Brigham (MGB), formerly Partners HealthCare System, is a large integrated health care system in eastern Massachusetts, which cares for 1.5 million patients annually. Brigham and Women's Hospital (BWH) is a founding member of MGB and treats 700,000 patients annually. The BWH Division of Genetics, along with subspecialty genetics clinics in other divisions within the Department of Medicine, provides care for patients in need of genetic risk assessment, genetic testing, genetic diagnosis, genetic counseling, and management of genetic disease. The clinical genetics program sees approximately 10,000 patients annually in total including the Adult Genetics Clinic, along with distinct programs and clinics for pharmacogenomics, pulmonary, endocrine, cardiovascular, preconception/prenatal, and preventive genetics.

Description of eConsult service

In 2014, MGB launched an eConsult program through which PCPs could seek specialist expert opinion virtually through chart review.[22] Since April 2016, eConsults have been managed with electronic health record (EHR) software from the Epic Systems Corporation. The BWH Division of Genetics launched its eConsult service in July 2018. Since its inception, the service has used a general internist PCP with additional training and particular interest in genetic and genomic medicine to respond to eConsults. This PCP has pursued additional self-education in clinical genetics, including a year-long fellowship lecture series, online educational offerings, including the American College of Medical Genetics and Genomics review course, and clinical observations in multiple BWH genetics clinics. In responding to eConsults, this PCP champion is supervised by a medical geneticist, cancer geneticist, clinical pharmacist, and/or genetic counselor, as appropriate. As of January 31, 2022, this service had received 462 consults, described below. The MGB Institutional Review Board determined that this work did not constitute human subjects research.

Figure 1 illustrates the processes by which genetics eConsults are initiated, handled, and completed. The referring provider enters an eConsult order indicating genetics as the specialty requested and states the specific patient question for the consultant (Supplemental Figure 1). The PCP champion receives the request through the EHR as an in-basket message and is expected to respond within 48 business hours. The PCP champion reviews the clinical question and drafts a response, drawing on clinical genetic resources, including GeneReviews, National Comprehensive Cancer Network guidelines, his own medical knowledge, and health-care-system-based information, including appropriate referral patterns and scheduling processes within the MGB system. For each eConsult, the PCP champion makes a statement whether conventional genetics consultation is recommended, is not recommended, or could be considered at the discretion of the referring provider. Decisions on whether to recommend a conventional face-to-face consult are based on various factors, including professional guidelines for genetic testing. The eConsult responses also included recommendations on additional information gathering, such as additional family history gathering, family member genetic testing results, labs, imaging, or other subspecialty consultation that would be informative. After drafting the eConsult response, the PCP champion sends it to a medical geneticist, cancer geneticist, clinical pharmacist, and/or genetic counselor for review, as appropriate. The genetics specialist(s) may revise the response as needed before the PCP champion completes the eConsult (Supplemental Text 1). The completed eConsult appears as

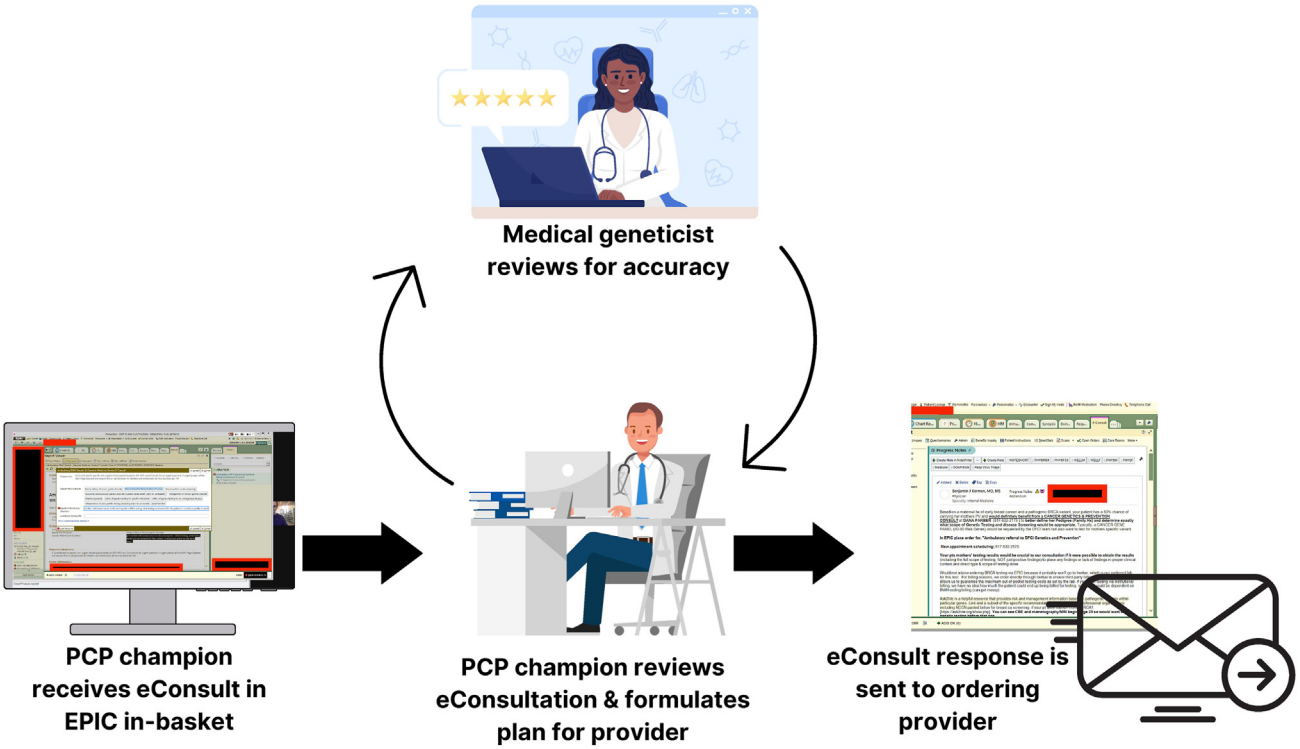

Figure 1 Workflow of genetics eConsult service. The referring provider enters a genetics eConsult order in the EHR, stating the question for the consultant. The PCP champion receives the request through the EHR as an in-basket message, reviews the clinical question and drafts a response. The draft response is reviewed and revised as needed by a genetics specialist before the PCP champion's completion of the eConsult. The completed eConsults appear as an alert in the referring provider's EHR in-basket and are documented in the patient's record.

an alert in the referring provider's "in basket" and is documented in the patient's EHR (Supplemental Figure 2).

Data collection

Data for the present report were collected in the following ways. First, whether and the degree to which the supervising genetics specialist revised the PCP champion's draft response to the eConsult was recorded. Second, retrospective chart review was conducted on all eConsults received from inception from July 2018 through January 2022 (n = 462, Table 1). Chart reviews were conducted a median of 16.9 months after each eConsult entry (mean 18.9 months, range 5.5-30 months) to allow for adequate time for completion of conventional consults and genetic testing, if ordered. Abstracted data included the clinical question asked, whether the PCP champion recommended a conventional genetics consult, whether the referring provider ordered a conventional genetics consult, and whether the patient attended a genetics appointment. If the patient attended a genetics appointment, data were collected regarding recommended clinical management changes, whether genetic testing was ordered, and genetic testing results (if testing was completed). Third, a survey was sent through an Epic message to a convenience sample of 125 providers after they ordered an eConsult but before a response was sent. Providers were asked how likely they would have been to request a conventional genetics consult as an alternative if the eConsult service had not been available; answer choices given were 0% to 20%, 20% to 40%, 40% to 60%, 60% to 80%, or 80% to 100%. Fourth, another survey was sent via Epic message to a different convenience sample of providers after completion of 366 eConsults, asking providers to rate their satisfaction with the service on a scale from 1 to 5, with 5 being the most satisfied.

Data analysis

Data are presented descriptively as medians with ranges or proportions, as appropriate. Clinical questions in the eConsults were categorized as shown in Table 2. A 3-category kappa statistic was used to compare provider-reported pre-consult intention for conventional genetics consult and the PCP champion's recommendation for conventional consult.[23] For this comparison, provider intentions of 0% to 20% or 20% to 40% were considered concordant with a PCP champion recommendation not to seek conventional consultation, intentions of 40% to 60% were considered concordant with a recommendation discretionary consideration of a conventional consultation, and intentions of 60% to 80% or 80 % to 100% were considered concordant with a recommendation for conventional consultation (Table 3). For patients who completed a conventional genetics consultation, management recommendations from

Table 1 Demographics for 462 unique patients who had a genetics eConsult completed

Characteristic	Total (%)
Sex[a]	
Male	153 (33)
Female	309 (67)
Ethnicity	
Hispanic	39 (8.5)
Not Hispanic	398 (86)
Unavailable	23 (5)
Decline	2 (0.4)
Race	
White	366 (79)
African-American	27 (5.8)
Asian	26 (5.6)
American-Indian/Alaska Native	4 (0.9)
Decline	3 (0.7)
Other	27 (0.9)
Unavailable	9 (1.9)
Age	
0 to 4	12 (2.5)
5 to 11	2 (0.4)
12 to 19	7 (1.5)
20 to 39	240 (51.9)
40 to 59	119 (25.7)
60 to 79	78 (16.8)
>80	4 (0.8)
Mean	41
Median	37

[a]Sex was extracted from administrative clinical data predominantly entered by staff and not necessarily reflective of gender identity.

that consultation were categorized as genetic testing, augmented disease screening, cascade testing, personal phenotyping, specialty referral, diagnostic testing of relatives, medication changes, preconception testing, and others (Table 4). For patients who ultimately underwent genetic testing, results were considered "informative" if they resulted in any changes in management, including pathogenic or likely pathogenic variants in genes associated with autosomal dominant, recessive, or X-linked diseases and pharmacogenomic variants predicting atypical drug response associated with Clinical Pharmacogenetics Implementation Consortium guidelines (Table 5).[24] In describing the genetics specialist supervision of the PCP champion's eConsult responses, substantive revisions were defined as a change in whether and which subspecialty clinic recommended for referral or major change in wording, emphasis, or approach of the response.

Results

Overall eConsult service utilization

Between June 2018 and January 2022, the genetic eConsult service received 462 consults from 232 unique providers, including 390 out of 462 (84%) consults from adult PCPs, 17 out of 462 (4%) from pediatricians, and 62 out of 462 (13.4%) from specialists. Provider types requesting consults included 379 out of 462 (82%) attending physicians, 30 out of 462 (6%) of consults were from residents in training, 34 out of 462 (7%) were from nurse practitioners, and 23 out of

Table 2 Categories of questions among the first 462 patients for whom a genetics eConsult was ordered

Category of Questions	Consults within the Category $n = 462$
Family history (non-cancer)	50 (11)
Family history of cancer	221 (48)
Preconception	21 (5)
Cardiovascular	23 (5)
Known genetic disease	8 (2)
Pharmacogenomics	17 (4)
Genetic testing is indicated	46 (10)
Genetic testing to diagnose unknown disease	38 (8)
Genetic test results interpretation	26 (6)
Pediatrics	12 (3)

Table 3 Comparison of referring providers' initial intentions and PCP champion's recommendations for conventional genetics consultation

	PCP Champion's Recommendation for Conventional Genetics Consult		
Referring Providers' Intention to Consult	Consult Recommended ($n = 62$, 70%)	Discretionary Consult Recommended ($n = 14$, 16%)	Consult Not Recommended ($n = 12$, 14%)
Intention to consult $n = 57$ (65%)	**46 (81%)**	7 (12%)	4 (7%)
Uncertain intention $n = 21$ (24%)	10 (48%)	**4 (19%)**	7 (33%)
No intention to consult $n = 10$ (11%)	6 (60%)	3 (30%)	**1 (10%)**

Data from 88 of 125 referring providers (response rate 70%) who completed survey questions about their intention, before receiving the genetics eConsult response, to order a conventional genetics consult. Data shown in the first column are counts (proportions) of referring providers' intention to order a conventional genetics consult, in which reported likelihood of consult of 0% to 40%, 40% to 60%, and 60% to 100% were considered no intention, uncertain intention, and intention, respectively. Data shown in the first row are counts (proportions) of patients for whom the PCP champion recommended, recommended at discretion, or did not recommend formal genetics consultation. Data in the remaining cells are counts (proportions within rows). Bold cells indicate patients for whom the referring provider's intention and PCP champion's recommendation were concordant (overall kappa 0.14 [95% CI −0.01, 0.29], indicating only slight agreement).

Table 4 Clinical recommendations among 171 patients completing conventional genetics consultation

Clinical Recommendation	Patients with Recommendation (%)[a]
Genetic testing	121 (71)
Augmented disease screening for patient	40 (23)
Cascade testing	28 (16)
Personal phenotyping	18 (11)
Specialty referral	10 (6)
Diagnostic testing of relatives	8 (5)
Medication changes	7 (4)
Preconception testing	6 (4)
Other[b]	5 (3)
Total recommendations	**243**
Mean number of recommendations per patient	**1.9**
Median number of recommendations per patient (range)	**2 (1-5)**

[a]Total number of recommendations is greater than the number of patients with informative genetic findings because of the fact that some patients received a >1 clinical recommendation.

[b]Other recommendations include augmented disease screening of family, disease surveillance, lifestyle modification, preconception testing of a partner, and variant of uncertain significance re-evaluation.

462 (5%) were from physician assistants. Among the 462 patients, mean age was 41 (SD of 17) years, 309 (67%) were women, and 153 (33%) were men. Racial categories included 366 (79%) people identified as White, 27 (6%) as African-American, and 26 (6%) as Asian. Thirty-nine (8.5%) identified as Hispanic (Table 1). Among 232 unique providers, 145 providers placed only a single consult, whereas 87 providers placed more than one eConsult (range 2-13), with an overall mean of 2 consults per provider. All eConsults responses were sent within 48 business hours from the eConsult request order.

Intentions and recommendations

Of the 462 eConsults received, the eConsult service recommended a conventional genetics consult for 73% (338/462), at consultee provider discretion for an additional 12% (54/462), and no conventional genetics consult was recommended for 15% (70/462, Figure 2). Eighty-eight (70%) of the subset of 125 referring providers responded to the survey about their pre-consult intention to place a conventional genetics consult. Table 3 shows the comparison between the referring providers' intentions to place a genetics consult (10/88 [11%] no, 21/88 [24%] uncertain, and 57/88 [65%] yes) and the eConsult service's recommendations for conventional consultation (12/88 [14%] no, 14/88 [16%] discretionary, and 62/88 [70%] yes). Comparison between the intentions and recommendations indicated only slight agreement (kappa 0.14 [95% CI −0.01, 0.29]). The eConsult service drove a small number of new conventional genetics consults (6/88 [7%]) in which the eConsult recommended consultation when the referring provider did not otherwise intend to consult and averted a small number of conventional consults deemed unnecessary (4/88 [5%]) in which the eConsult recommended against a consultation the referring provider intended to place. Of the patients for whom the providers were uncertain about whether to order a conventional genetics consult, the eConsult recommended a consult or discretionary consult for 67% and recommended against a consult for 33%.

eConsult outcomes

Figure 2 shows the numbers of patients progressing from eConsult to conventional genetics consult and genetic testing. Among the 338 out of 462 (73%) eConsults for which the PCP champion recommended conventional genetics consultation, the referring providers for 254 out of 338 (75%) ordered a consult. When the PCP champion recommended a discretionary genetics consultation 54 out of 462 (12% of the time) or did not recommend a genetics consultation 70 out of 462 (15% of the time), the referring provider ordered a consult for 18 (33%) and 7 (10%), respectively. Of the overall 279 referred for genetics consultation, 171 (61%) completed a consult after a median 3.1 months (range 1 day to 28 months). By year, this conventional consult completion rate was 79% in 2018, 63% in 2019, 60% in 2020, 56% in 2021, and 63% in 2022. In comparison, the conventional consult completion rate for adult genetics at our institution overall was 44% (2310/5192) during the same time period: 39% in 2018, 41% in 2019, 50% in 2020, 45% in 2021, and 44% in 2022. Completed genetics consults after eConsult included 155 out of 254 (61%) of those recommended and 14 out of 18 (78%) of those discretionarily recommended by the PCP champion for consultation. Among the largest topic categories of consults, cancer genetics (221/462, 48%), 173 out of 221 (78%) contained educational resources intended to inform and educate requesting providers. Resources included National Comprehensive Cancer Network guidelines,[25] GeneReviews articles,[26] Familial Cancer Database,[27] and risk calculators for breast and colon cancer, as well as patient-facing resources such as MedlinePlus: Genetics and Genetic and Rare Diseases[28] and National Organization for Rare Disorders websites.[29] In the same cancer genetics group, 113 out of 221 (51%) had recommendations made for testing or obtaining available clinical information on relatives with the goal to increase efficiency of face-to-face consults.

Outcomes of conventional genetic consultation

Table 4 shows the conventional genetics consultants' clinical recommendations for the 171 patients who completed a genetics consult, including 40 recommendations for augmented disease screening and 28 recommendations for cascade testing of family members. Among the 171 patients, 122 (71%) were recommended to undergo

Table 5 Informative results among 84 patients completing genetic testing after conventional genetics consultation

Patient Age and Gender	Consult Category	Reason for eConsult	Test Type	Gene with Informative Finding	Management Recommendations
Cancer ($n = 7$)					
36 F	FHx of known disease	FHx pancreatic, prostate, breast cancers	Cancer panel	*BRCA1*	NCCN recommendations
56 F	FHx of known disease	Insurance coverage for FHx *BRCA1*	Cancer panel	*BRCA1*	NCCN recommendations
83 F[a]	FHx of known disease	Daughter with *BRCA2* variant	Cancer panel	*BRCA2* *CHEK2*	NCCN recommendations
57 M	Personal history of known disease	Prostate cancer with FHx of prostate and breast cancer	Cancer panel	*HOXB13*	Prostate cancer screening, cascade testing
43 M	FHx of known disease	Mother with known *APC* variant; maternal aunt with pancreatic cancer	Cancer panel with specific *APC* variant analysis	*APC*	NCCN recommendations
31 F	Familial variant testing	Mother with *BRCA1* variant	*BRCA1* familial variant testing	*BRCA1*	NCCN recommendations
31 M	FHx of known disease	FHx of breast cancer	Cancer panel	*BRCA2*	NCCN recommendations
Preconception ($n = 6$)					
29 M	Family planning	Preconception carrier testing for Ashkenazi patient with known Lynch syndrome variant	Expanded carrier screen (176 genes)	*DLD* *CYP27A1*	Reproductive partner testing
64 M[b]	FHx of known disease	FHx of spinal muscular atrophy	*SMN1/2* sequencing	*SMN1* coding sequence deletion	Carrier testing of adult offspring
30 F	Family planning	Ashkenazi Jewish ethnicity	Ashkenazi Jewish Carrier Screening (22 genes)	*GBA*	Reproductive partner testing
24 F	Familial variant testing	FHx alpha-1 antitrypsin (A1A) deficiency and personal history of low A1A levels	*A1AT* targeted testing	*A1AT*	Pulmonary and liver function testing; reproductive partner testing; avoid hepatotoxins
32 F	Interest	Patient expressed interest in carrier testing	Expanded carrier screening (176 genes)	*CP2T* *NPHS2*	Reproductive partner testing
38 F	FHx of known disease	FHx of hypohidrotic ectodermal dysplasia	Expanded carrier screening (284 genes)	*DHCR7*	Reproductive partner testing
General Genetics ($n = 6$)					
35 M	Diagnostic testing for suspected disease	Concerns for hypophosphatasia	Targeted gene sequencing	*ALPL*	Hypophosphatasia management; reproductive partner testing
32 M	Diagnostic testing for suspected disease	Suspected velocardiofacial syndrome; ventricular septal defect; hyperparathyroidism	Chromosomal microarray testing	22q11.2 deletion syndrome	Refer to 22q clinic for extensive surveillance
21 F	Diagnostic testing for suspected disease	Rhabdomyolysis	Neuromuscular panel	*PYGM*	Type V glycogen storage disease; light exercise; TTE; DEXA; caution with anesthesia and statins
32 F	Familial variant testing	FHx Alport syndrome	Familial variant testing	*COL4A5*	Cascade testing; renal, audiological, and ophthalmologic monitoring

(continued)

Table 5 Continued

Patient Age and Gender	Consult Category	Reason for eConsult	Test Type	Gene with Informative Finding	Management Recommendations
33 F[b]	FHx of known disease	FHx of myotonic dystrophy	Targeted gene sequencing	*CNBP*	Type 2 myotonic dystrophy management/surveillance
40 F	Familial variant testing	FHx of MODY	Familial variant testing	*HNF1A*	Cascade testing; screen for MODY
Pharmacogenomics (n = 3)					
88 M	PGx	Statin ADR	PGx panel	*SLCO1B1* (poor metabolizer)	Change current statin treatment (simvastatin to pravastatin)
36 M[b]	PGx	Many ADR to SSRIs	PGx panel	*CYP2D6* (poor metabolizer) *CYP2C9* (intermediate metabolizer)	Explanation for prior ADR given; recommendations for future use of opiates, SSRIs, and TCAs provided
66 F	PGx	DRESS syndrome to vancomycin, amiodarone, and ceftriaxone	PGx panel	*CYP2C19* (Intermediate metabolizer)	No change to current Rx; recommendations for future use of clopidogrel, PPIs, TCAs, and SSRIs provided

Of "positive" results, eConsult indications, genetic testing results, and management recommendations.
ADR, adverse drug reaction; *DEXA*, dual x-ray absorptiometry; *DRESS*, drug rash with eosinophilia and systemic symptoms; *FHx*, family health history; *MODY*, maturity-onset diabetes of the young; *NCCN*, National Comprehensive Cancer Network; *PGx*, pharmacogenomics; *PPI*, proton pump inhibitor; *Rx*, prescription; *SSRI*, selective serotonin reuptake inhibitor; *TCA*, tricyclic antidepressant; *TTE*, transthoracic echocardiography.
[a]Indicates patient for whom consulting provider responded with intention not to consult genetics in pre eConsult survey.
[b]Indicates patient for whom consulting provider responded with intention to consult genetics in pre eConsult survey.

genetic testing, and 84 (69%) of these completed testing (Figure 2). Table 5 shows the 23 of 84 (27%) patients whose genetic testing identified informative findings, including 7 variants in genes associated with cancer syndromes, 6 variants with relevance for preconception testing, and 3 pharmacogenomic results consistent with prior or current drug response phenotypes.

Supervision and performance of PCP champion

Overall, the genetics specialists made substantive revisions to the PCP champion's draft responses for 8% (36/462) of the eConsults completed: decreasing from 18% (17/92) in the first year of the service (July 20, 2018 to July 19, 2019), 6% (8/140) in the second year (July 20, 2019 to July 20, 2020), 5% (7/152) in the third year (July 21, 2020 to July 20, 2021), and 4% (4/78) for the remainder of the observation period (July 21, 2021 to January 31, 2022, Figure 3). Referring providers for 184 of 370 eConsults (49.7%) completed the survey and rated their satisfaction with the service highly (mean 4.93 [SD 0.26], median 5 on a 5-point scale). Providers were asked to leave open-ended responses related to their satisfaction or dissatisfaction with the eConsult service. Many providers mentioned their satisfaction with the response timeliness and educational material provided to them by the PCP champion (Supplemental Table 1).

Discussion

Outcomes from the first 462 patients demonstrate that a novel PCP champion-operated, genetics eConsult service is feasible, provided timely access to specialty recommendations, and identified substantial proportions of patients recommended for genetic testing. The service promoted the identification of informative genetic findings and management changes that might not have otherwise occurred. This service delivery model warrants further evaluation as a potentially useful addition to the genetic services of health care systems.

Traditionally, eConsult services are maintained by specialty care providers responding to PCP requests.[2,3,7,9,13,14] Other groups in Ontario and Massachusetts have described their experiences of eConsult services managed by trained genetics professionals.[19–21] Other institutions have employed non-geneticists as clinical genetics champions, such as the PCP pilot DNA-10K population genetic testing program at the Northwestern University Health System.[30] To our knowledge, ours is the first description of a specialty care eConsult service operated by a PCP, with guidance and supervision from genetics professionals, and the first reporting of genetic testing and results following the eConsults. There is a recognized shortage of clinical geneticists and genetic counselors.[19–21] A PCP champion with professional interest in genetics and familiarity with local health care system referral processes may improve timely access to specialty knowledge and recommendations while

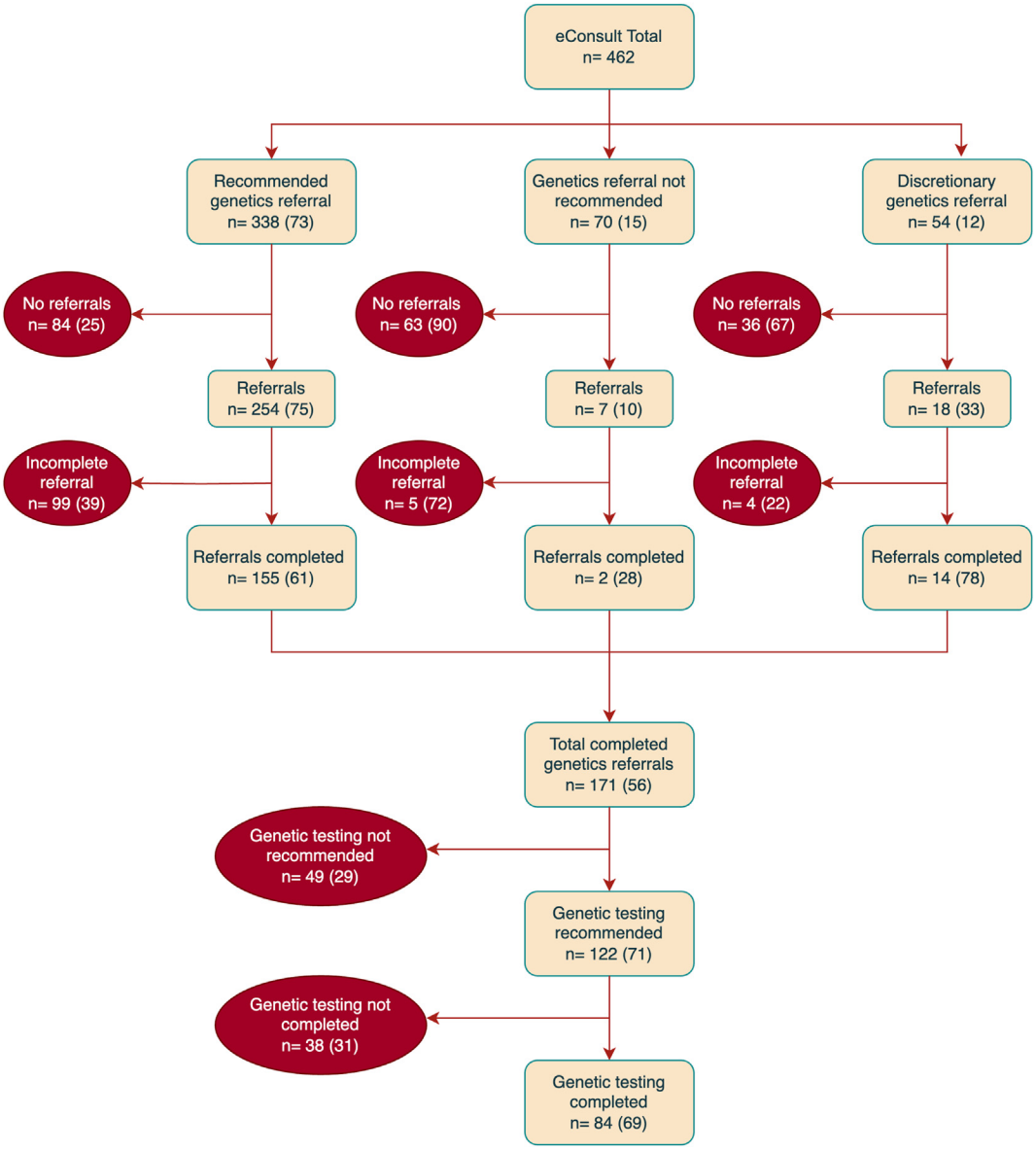

Figure 2 Outcomes of 462 genetics eConsults completed by PCP champion, July 2018 to January 2022. For each eConsult, a statement is made whether conventional genetics consultation is recommended, is not recommended, or could be considered at the discretion of the referring provider. The figure illustrates whether the 462 patients were referred for conventional genetics consultation, completed conventional genetics consultation with median wait time of 3.1 months (range 1 day to 28.1 months), and were recommended for and completed genetic testing, along with the results of such testing. Data shown are counts and proportions relative to the counts in the preceding boxes.

also providing eConsult responses that account for the larger primary care context of provider genetics knowledge gaps, population health and disease prevention, clinical and system workflows, and multiple competing demands. Referring providers expressed high satisfaction with the service, although we cannot exclude the possibility that survey respondents were more likely to have favorable opinions than non-respondents. Our case series cannot directly compare the PCP eConsultant model with a geneticist eConsultant model to measure these potential benefits.

Access to genetics services in the United States and globally is inadequate.[15–18] Access to health care includes both the availability of services and the ability of patients to access those services in a timely way.[31] This case series is limited to a single PCP eConsultant at a single academic medical center with existing genetics services. Even in this setting, the eConsult resulted in timelier genetics services because referring providers received guidance within 48 business hours. However, this case description cannot demonstrate whether a PCP-led genetics eConsult service can improve availability and timeliness of genetics services to systems with even poorer access at baseline, including non-academic and rural settings. A PCP-led eConsult model has the potential to improve access in these settings because

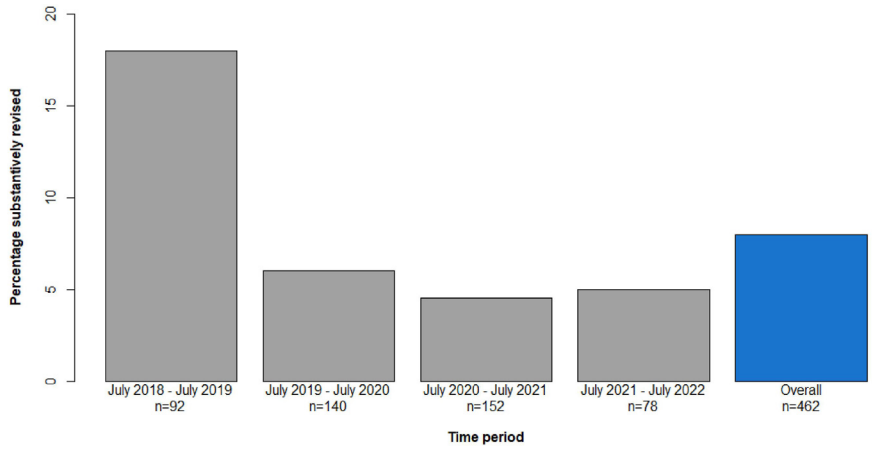

Figure 3 PCP champion performance over time. Data shown are proportions of PCP champion's initial eConsult draft responses (out of total *n* in a given period) substantively revised by the supervising genetics specialist, defined as a change in the recommended subspecialty clinic for referral or major change in wording, emphasis, or approach of the response.

it does not rely on geographic proximity or the timely availability of genetics professionals, if the PCP eConsultant can be trained to respond to most consults independently. Further work is needed to evaluate whether this model can improve availability and timely access to genetics services in currently underserved areas.

In some health system contexts, eConsult services often have the goal of decreasing the number of face-to-face consultations, and previous reports have focused primarily on "unnecessary" consults avoided.[2,7,12,22] In contrast, we observed clinical benefit of the eConsult service in identifying patients who should be referred for appropriate care where 73% of eConsults resulted in conventional consult recommendation, whereas a conventional consultation was not recommended for only 15% of eConsults.[4–6] There was only slight agreement between the referring provider's initial intention and the PCP champion's eConsult recommendation about whether the patient should undergo a conventional genetic consultation. In this sense, eConsults seemed to serve a triage function to the referring provider in deciding which patients to refer formally. Referring providers had the option to place a conventional consult before eConsultation. The fact they placed an eConsult instead also intrinsically demonstrates their uncertainty regarding need for conventional consultation. The data suggest a small increase in necessary and decrease in unnecessary conventional consults as a result of the eConsult service. The eConsult service may have also increased the efficiency of eventual face-to-face consultations by recommending that relevant clinical data or genetic testing be obtained before the conventional consult in about half of cases. In the end, the eConsult service contributed to genetic diagnosis and new medical management changes that may not have otherwise occurred.

We observed drop-off at each step in the clinical pathway between eConsult and ultimate conventional consultation and genetic testing. When the eConsult recommended a conventional referral, the referring provider ordered a conventional referral 75% of the time, and overall, 61% of patients for whom a referral for conventional consult was placed completed the conventional consult. Similarly, only 69% of patients for whom genetic testing was recommended completed testing. This may speak to logistical, financial, administrative, and patient factors. However, for comparison, approximately 44% of ordered conventional genetics consults at our institution were completed (in the same time period), compared with an average of 57% (range of 30% to 77%) across all specialties. In each of the first 5 years of the genetics eConsult period (2018-2022), rates of completion of conventional genetic consults were higher among those patients for whom the referring provider first ordered a genetics eConsult. Further investigation should examine the possibility that an eConsult such as ours increases the likelihood of completion of a conventional genetics consult, if ordered.

Safety and scalability are appropriate questions to consider for a PCP-staffed genetics eConsult service. Only 8% of the PCP champion's draft eConsult responses required substantial revision from supervising geneticists, and performance improved over the 4 years observed, suggesting the delivery of appropriate care. Because genetics providers, a limited resource, were still involved in the supervision of the PCP champion, future work would need to evaluate the appropriateness and safety of a model in which the PCP has discretion to respond to some questions independently and escalate only challenging or complex questions to the specialists. Future work should also examine the generalizability of this model to other PCP champions. The PCP champion in our program had pursued additional didactic and practice-based self-education in clinical genetics. The experience and metrics needed to train and evaluate such a PCP champion have yet to be defined. A formal standardized, appropriately scoped program for genetics education and certification of non-geneticist providers could be envisioned to create a pipeline of PCP genetics champions in the future. A small minority of PCPs can or will want to be clinical genetics champions, but some will welcome the opportunity for professional development and rejuvenation in an era of

rampant provider burnout. This was very much the case for the PCP champion for this genetics service (B.J.K.) for whom it led to involvement as lead physician for the BWH Pharmacogenomics Clinic and other professional opportunities in clinical implementation and research.

In conclusion, this case series demonstrates that a PCP champion eConsult model can feasibly triage and respond to genetics questions in a timely manner and provide PCP-relevant genetics information and high provider satisfaction. Future work is needed to evaluate the generalizability of this model to other PCP champions and to underserved health systems and its impact on patient access to timely, high-quality genetics services.

Data Availability

Deidentified data are available upon written request to the corresponding author.

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Ethics Declaration

The study was deemed exempt from human subject research by the Massachusetts General Brigham Institutional Review Board.

Conflict of Interest

Robert C. Green has received compensation for advising the following companies: Allelica, Atria, Fabric, Genomic Life and Juniper Genomics; and is co-founder of Genome Medical and Nurture Genomics. All other authors declare no conflicts of interest.

Additional Information

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