Anticipating the primary care role in genomic medicine: expectations of genetics health professionals

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Abstract

Our purpose was to explore genetics health professionals' (GHPs) expectations of primary care providers' (PCPs) role in genomic medicine now and in the future. Focus groups/interviews were conducted with GHPs in Ontario, Canada. Recordings were transcribed and analysed using qualitative descriptive analysis. Five focus groups (6 clinical geneticists, 24 genetic counselors, 1 nurse, 4 laboratory staff, 3 genetics program administrators) and 3 interviews (nurses) were conducted. GHPs described a key role for PCPs in genomic medicine that could be enhanced if GHPs and PCPs worked together more effectively, making better use of GHPs as a scarce specialist resource, improving PCP knowledge and awareness of genomics, and increasing GHPs' understanding of primary care practice and how to provide PCPs meaningful education and support. Health system change is needed to facilitate the GHP/PCP relationship and improve care. This might include: PCPs ordering more genetic tests independently or with GHP guidance prior to GHP consultations, genomic expertise in primary care clinics or GHPs being accessible through buddy systems or virtually through telemedicine or electronic consultation, and developing educational materials and electronic decision support for PCPs. Our findings highlight need for change in delivering genomic medicine, which requires building the relationship between GHPs and PCPs, and creating new service delivery models to meet future needs.

Keywords Genetics · Genomic medicine · Primary care · Genetic counselling · Genetic service delivery

Introduction

Developments in genomic medicine are widely expected to impact primary care. Primary care providers (PCPs) have described their role as taking a complete family history, identifying individuals at increased risk for genetic conditions and referring appropriately, discussing the benefits and limitations of genetic testing and providing post-test care for patients, including psychosocial support and coordination of screening and management (Carroll et al. 2016b, 2019; Houwink et al. 2011). Despite the potential for this expansive role, the integration of genomic medicine into primary care has proceeded slowly. A systematic review of barriers to the uptake of genetics in primary care identified factors related to genetic services, such as limited evidence of clinical utility, and ethical, legal and social challenges (e.g. potential for patient anxiety and distress, insurance discrimination), as well as challenges related to primary care services (e.g. lack of time, lack of access), and limitations of knowledge and skills among PCPs, as consistent concerns (Mikat-Stevens et al. 2015).⁴ In addition to these, PCP lack of awareness of

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genetic services, inadequate coordination of referral, and lack of genetics workforce are frequently reported (Delikurt et al. 2015; Haga et al. 2013; Hamilton et al. 2014; Houwink et al. 2011).

Many “roadmaps” for addressing these limitations and advancing the integration of genomic medicine into primary care highlight the importance of improved linkages between PCPs and genetics health professionals (GHPs) (David et al. 2015; Houwink et al. 2013; Manolio et al. 2013; Pearce et al. 2019). David et al. (2015) argue that “Any vision for comprehensive primary care redesign should include a strategy for integrating genomic medicine with primary care in academic and nonacademic settings and engage genomic medicine leaders in developing a clinical implementation strategy”. Similarly, Haga et al. (2013) note that PCPs will play an important role in the delivery of genomic medicine, including the role of gatekeeper to services, and screening and disease prevention, but that they will need “ready access to genetics experts” and that the best practice standards will come from “a collaboration between medical genetics and other clinical specialties”.

Despite the anticipated importance of linkages between PCPs and GHPs, most research to date has explored only the views of PCPs regarding genomic medicine. Little is known about the views of GHPs regarding the role of PCPs in genomic medicine, and how they see themselves working with PCPs. The purpose of this project was therefore to explore GHPs’ experiences with PCPs including the role they see for PCPs in genomic medicine now and in the future.

Materials and methods

Study design and participants

Given the project’s exploratory nature, we used a qualitative approach employing a descriptive analysis, informed by the principles of grounded theory (Charmaz, 2006; Patton 2002; Sandelowski, 2000). Qualitative research is well suited to exploration of interactions or processes among individuals or groups, and the beliefs and expectations that inform action. Thus, we conducted in-person focus groups (FGs)

to gain insight into collective social interpretations among GHPs regarding experiences with and expectations of the primary care delivery of genomic medicine. Individual phone interviews were used in centres with too few staff for a focus group. Interviews offer the potential of deeper understanding of a topic, while focus groups enable more wide ranging discussion. Our research was conducted in Ontario, Canada, where the provincial health insurance system provides universal, comprehensive access to physician and hospital services to all residents, free at point-of-care, including primary care and an array of specialist services. PCPs are first point-of-care for all medical services, with non-emergency access to specialist care typically requiring PCP referral. Genetic services are delivered through regional centers (10 in the province), situated in academic and community hospitals staffed by clinical geneticists and genetic counselors, with specialized genomic lab services. In addition, there are 5 northern outreach clinics staffed by genetic counselors and/or nurses with visiting geneticists providing 3 to 6 clinics/year. Urgent referrals, for example for prenatal indications, may be seen immediately, but wait times for non-urgent referrals can sometimes exceed a year (Cancer Care Ontario, 2018).

Focus group/interview guide

After reviewing the literature about the PCP role in genomics, seven open-ended questions were incorporated into an FG/interview guide, summarized in Table 1. The guide was revised as the FGs and interviews progressed.

Data collection

We recruited GHPs from 5 of 10 Ontario regional genetics centers and all 5 northern outreach clinics. Sites were chosen to reflect diverse locations and range of genomic services. Email invitations describing the project were sent to genetics program managers at each selected center, followed by a call from the research coordinator (SM) to answer questions. If program managers expressed interest, project information and invitations were sent to them for distribution to

Table 1 Focus group/interview guide

1. What comes to mind when you think of genetics in primary care?
2. What has been your experience with PCPs and genetic service delivery?
3. What are your thoughts on the role of PCPs in genetic service delivery?
4. What are your thoughts about and experiences of educating PCPs about genetics?
5. What are your thoughts about the future of genetics and genetic service delivery?
6. Are there any other initiatives or changes that you would implement with respect to genetic service delivery and education within the next 5 years?
7. Are there any other issues about primary care genetics education that you would like to raise?

the GHPs in their clinics. Eligible participants were clinical geneticists, genetic counselors, nurses, laboratory staff and genetics program administrators. FGs/interviews each lasted about an hour and were audiotaped. We collected demographic data using baseline questionnaires prior to FGs/interviews.

Data analysis

Recordings were transcribed verbatim, anonymized and analysed using techniques informed by grounded theory including coding, interpretations of patterns in the data, and use of the constant comparative method (Charmaz, 2006). Co-investigators (JCC, SM, JEA) read the same two transcripts independently and developed a coding manual in collaboration with other team members. Following each FG, three of the authors (JCC, SM, JEA) met to review tentative findings and adjust future FG/interviews in light of emerging themes. We asked participants about these emerging themes in order to support or challenge our evolving understanding. Remaining transcripts were coded by SM. Codes were developed, grouped and re-grouped into descriptive categories, which were refined through discussion into three themes that were agreed upon by all researchers. It is these themes that are present in the Results section and in the Discussion. See Fig. 1 for the analysis pathway. The researchers came from

diverse professional backgrounds to help mitigate bias, including a family physician (JCC), clinical geneticist (JEA), genetic counselor (SM), public health physician and health services researcher (BW) and health policy researcher (FM).

Results

Sample characteristics

Staff from four regional centers and three northern clinics agreed to participate. Five mixed professional FGs (6 clinical geneticists (MD), 24 genetic counselors, 1 nurse, 4 laboratory staff, 3 genetics program administrators) and three interviews (nurses) were conducted. See Table 2 for participant demographics.

Focus group and interview findings

GHPs described key roles for PCPs in genomic medicine that could be enhanced if GHPs and PCPs worked together more effectively. By doing so, they saw great potential for improving delivery of genomic services in the future and improving patient outcomes. Three main themes emerged: the need for PCPs and GHPs to work together more effectively to make better use of GHPs as a scarce specialist resource, the critical

Fig. 1 Qualitative data analysis pathway

Table 2 Participant demographics ($n=41$)

Age*	Mean age 40 years (range 24–60)
Sex	Male: 3 Female: 38
Years in practice ⁺	0–4: 10 5–9: 2 10–14: 4 15–19: 3 20 or more: 6
Profession	Clinical geneticist (MD): 6 Genetic counselor: 24 Nurse: 4 Laboratory staff (laboratory heads, genetic counselors, managerial staff, excluding technicians): 4 Genetics program administrator: 3

*Data from 21 participants

⁺Data from 25 participants

importance of a PCP role in genomic medicine now and in the future and how GHPs can support that role, and strategies to improve delivery of genomic medicine.

Working together more effectively

GHPs argued that delivery of genetic/genomic services could be enhanced if PCPs and GHPs worked together more effectively to make best use of limited numbers of genomic specialists. This might be accomplished through changes by PCPs to improve their awareness of existing genetic/genomic services and how to use them, and to improve their knowledge of genomic medicine, as well as through changes made by GHPs to meet some of the challenges of primary care practice.

Making best use of already scarce genomic specialists Participants were concerned about the long wait times at genetic clinics and pressure for appointments, saying that PCPs have to get involved to help ameliorate this. They wanted PCPs to be part of the genomic medicine team, ordering initial investigations as part of the genetics workup, to make genetic consultations more efficient and meaningful. As one respondent explained, “I think that they will and should be taking over some of the more routine genetic tests...and then they can use the scarce genetic resources as a consult.” (Int 2 Participant 1\RN).

Improving PCPs’ awareness of genetic/genomic services To improve availability and efficiency of genomic services, participants described the need to improve PCPs’ awareness and use of existing services. “They do not have any kind of a comprehensive understanding of what our service pro-

vides and all the different types of genetic services that are available to the whole range of patients at different ages for different reasons.” (FG 1 Participant 2/MD) They described lack of clarity around when and how to make referrals as a potential barrier. “The other barrier that I see is even if they ask family history they need to know the processes...from a logistics perspective—now I have the family history but what do I do with it, is it an adult or child referral, cancer referral. Where does it go?” (FG 3 Participant 1/GC).

Improving PCPs knowledge of genomic medicine GHPs identified PCP lack of knowledge, demands of practice (time, scope of practice), and practical limitations (access to genetics centers and unclear indications for genetic tests) as barriers to delivery of genomic medicine by PCPs. Lack of knowledge was reflected in participants’ reports of both under- and over-referring by PCPs. “I think there is a bit of uncertainty about appropriate referrals... sometimes we wonder why some people were not referred and why other ones are. (FG2 Participant 2/GC) GHPs recognized the educational challenges facing PCPs with rapid advances in genomic medicine, “the level of knowledge that they require to interpret the information...which itself is becoming more and more specialized and detailed, the threshold is going to be raised. That is what is so difficult for them.” (FG 1 Participant 2/MD).

“The hub”: PCPs’ role in genomic medicine now and in the future

GHPs described the critical importance of PCPs in providing genomic medicine now and in the future and how GHPs could support that role. They described multiple components to the PCPs’ role, including taking a family history, identifying patients who would benefit from genetic consultation and referring appropriately. They had generally positive expectations of PCPs based on their interactions such as phone calls and referrals and described PCPs as key to patients’ access to specialty care. “When I think about the role of primary care ...there is a bit of the gate-keeper... to do the assessment and make sure those individuals who might benefit the most get there... The family physician is in an amazing position to not only educate, but...to influence the attitudes of individuals toward genetic testing. So, it is a really pivotal role in terms of encouragement or discouragement or providing realistic expectations about what will happen and to address the barriers that patients have.” (FG 1 Participant 8/MD).

GHPs were enthusiastic about a prospective expanded role for PCPs in genomic service delivery that would result in improved patient outcomes. “I also see them playing this huge role in the future with all of these new genetic tests coming because...they’re going to be the first one the

patient goes to and says, ‘Can I have this test?’ (FG 5 Participant 5/GC) Some GHPs wondered about their future role in genomic medicine. “I still see the primary care provider at the center or hub of the wheel...and all of us as spokes. I mean, for me the bigger question is not what the primary care provider’s role is going to be, but what our role is going to be.” (FG 1 Participant 2/MD).

Reciprocity: increasing GHPs’ understanding of primary care practice GHPs acknowledged they were unsure how genomic medicine fit into PCPs’ practice. Participants predicted this lack of understanding could impact the content of consultation letters and their value to PCPs. “It feels to me like we are losing touch with each other to a certain extent...so many of our referrals come from primary care and because we see a gap between the information we receive from the care provider and what we need to provide good care. Similarly, they may see when we write back to them, a gap in what information we are providing back or assumptions that they understand things that we think are second-nature.” (FG 1 Participant 2/MD).

Practical strategies to improve delivery of genomic medicine

GHPs identified that for the “hub” model to be successful in genomic medicine, the relationship between genomic services and primary care is key. They described how the relationship seems to work currently and how it could be improved in the future.

Current relationship between GHPs and primary care Personal communication and connection were described as important, through phone or email, informal conversations, consultation letters, or presence in specialty clinics. They described the benefits of this positive relationship. “I think there’s a lot of value in establishing connections between genetics professionals and primary health care providers... as genetic professionals, we play a huge role in educating primary health care providers, and the more they’re educated and know what services we provide, the better service or patient care is achieved.” (FG 5 Participant 2/GC) They described that PCPs were more likely to refer if they knew a genetics clinician personally or had spoken with them about a referral, highlighting the need for more personal contact through outreach, rounds, etc. “knowing where to get the information, who to go to and developing that relationship and making it really strong. They are overwhelmed and will be, but there is a support network in place.” (FG 3 Participant 6/Lab).

Possible ways to improve the relationship between PC and genetic/genomic services GHPs spoke of the importance of consultation letters as an opportunity for in-context education. “They’re getting...a nice snapshot of that particular condition that the patient was referred for. They’ve got some direction as far as screening recommendations. They’ve got information about...possibly referring other relatives. So, I think every time a consultation letter goes back, it’s...a teachable moment for that doctor.” (Int 1 Participant 2/RN) Participants said they welcomed calls from PCPs but received them infrequently and thought this was a service they could encourage. “I am very happy a family doctor or midwife calls with a question because it means they know that there is something that has piqued their awareness that they need to deal with, but are just unsure how...when they are in that situation again they...know what to do...or at least have the phone line to the genetics clinic.” (FG 2 Participant 4/GC).

Several GHPs identified strategies for improving the connection between GHPs and PCPs. Some suggested the integration of genetic counselors into primary care practices. From the experience of having a presence in a specialty clinic, e.g. a genetic counselor in oncology, they noted this had offered opportunities for teaching and improved appropriateness of referrals. “...having a genetic counselor in a family practice office where there are multiple doctors—a genetic counselor could support all of them to help improve and facilitate taking family history and seeing who is at risk or who deserves a referral to go see a geneticist.” (FG 2 Participant 4/GC) Others described reaching out to PCPs. “We’ve been talking about finding out when groups are having meetings and—not necessarily doing a formal presentation, but being there, being available to answer questions about our service, increase awareness and teach them about what it is that we do so they can do some of it themselves” (FG 5 Participant 3/GC).

See Tables 3, 4 and 5 for additional illustrative quotes.

Discussion

GHPs in this study described a vital role for PCPs in addressing the genetic/genomic service needs of patients now and in the future. They were also very clear about the need for PCPs and GHPs to work together more effectively, making the best use of already scarce genetics healthcare providers, improving PCP awareness of existing genetic/genomic resources and improving their knowledge of genomic medicine. GHPs described being challenged by their lack of understanding of primary care practice, what knowledge gaps exist and how to best provide meaningful education for PCPs. Our findings

Table 3 Theme #1: working together more effectively to deliver genomic medicine

Theme	Illustrative quote
Working together more effectively to deliver genomic medicine: Making best use of already scarce genomic specialists	<p>“We cannot see every sickle cell carrier in Ontario! The genetics clinic will be bogged down and really there is a step that can be done on the primary care level which is to test the partner and then see”. (FG 2 Participant 2/GC)</p> <p>“Thinking about how it would work if someone’s cousin had muscular dystrophy. Now, they may not be able to track everything down, but they should kind of prep the patient to find out what type of muscular dystrophy. You know, just to even kind of start to get them working on getting the information we are going to need would be really helpful.” (FG 2 Participant 2/GC)</p> <p>“They may take on more of a genetics role than they have in the past... especially if they have an interest in it they may, you know, take more responsibility for ordering some tests that traditionally people that have come to the clinics for, say for microarray, but the understanding would be that if they got result that, you know, then they would have to refer back to us. But, we have been encouraging physicians to take on more responsibility for some things, like hemochromatosis. And, you know, to be starting the work up for those kinds of patients before sending them directly to us.” (Int 1 Participant 1/RN)</p>
Improving PCPs’ awareness of genetic/genomic services	<p>“I think we have a sense that not all physicians are fully aware of our services and, umm, therefore, make use of us.” (FG 5 Participant 1/GC)</p>
Improving PCPs knowledge of genomic medicine: Under-referring	<p>“I think it comes up from time-to-time in prenatal where a patient will be referred for something on ultrasound or prenatal screen and as we are doing the family history something else comes up that is potentially much more relevant than the actual reason they were seen when they really should have been referred prior to pregnancy...it could have been a benefit to the patient.” (FG 1 Participant 3/GC)</p>
Improving PCPs knowledge of genomic medicine: Over-referring	<p>“Sometimes, I definitely see referrals in neurology where it is a single individual with one grandparent with Alzheimer’s disease and is being referred for a genetic assessment.” (FG 1 Participant 6/GC)</p>

underscore the need to build a robust relationship between GHPs and PCPs.

This adds to findings of a recent study in which PCPs reported generally positive experiences with genetics clinics and appreciated consultation letters for both patient care and their own education (Carroll et al. 2016a). However, few had ever connected with a genetics specialist. They described that, in contrast to other specialties, genetics referrals were often declined, perhaps because they did not meet referral criteria, but when that occurred they felt criticized and unsure of how to proceed with future referrals. GHPs in our study acknowledged their role in educating and supporting PCPs and the need to expand that role. They recommended increased outreach through rounds, targeted consult notes, and making providers aware of availability of phone call advice from genetic specialists. These strategies would build the relationship and enhance communication between PCPs and GHPs.

This study adds to the literature outlining the challenges to genetic service delivery, however, from the perspective of GHPs. Lack of access, inequity and variability of provision of genomic services are significant barriers to

integration into primary care (Mikat-Stevens et al. 2015; Pearce et al. 2019). PCPs have reported that genetic testing is not readily available, genetic centers are too far away for patients to access, and genetic consultations are difficult to obtain or have long wait times (Haga et al. 2012, 2013; Mikat-Stevens et al. 2015). As identified in this study, lack of awareness of genetic/genomic services has been identified as a barrier to appropriate referral by non-genetics health professionals. (Delikurt et al. 2015) Insufficient numbers of geneticists and genetic counselors to meet the present and future demands of genomic medicine will only compound these problems (Flannery, 2018; Mikat-Stevens et al. 2015; Stoll et al. 2018). There is discussion of genomic medicine moving from “genetic exceptionalism” to being “mainstreamed into routine preventive, diagnostic and interventional health care” to help ameliorate some of the issues described above, although there are challenges inherent in its delivery (Pearce et al. 2019). Our study showed that participating GHPs were in agreement with some of these changes, and for example, wanted PCPs to order genetic tests including both those that are “routine” (e.g. hereditary hemochromatosis) or

Table 4 Theme #2: PCPs' role in genomic medicine now and in the future

Theme	Illustrative quote
PCPs' role in genomic medicine now and in the future:	<p>"I see them as a client care hub or the coordinator of individual client's care—health care." (Int 2 Participant 1/RN) I see at this point in time we are faced with increasing numbers of patients who have potential genetic diagnoses or increasing numbers of genetic tests that can be done and we cannot possibly see all of those cases; therefore, we are starting to say to send it back to the family doctor to have testing. (FG 4 Participant 6/MD)</p>
Increasing GHPs' understanding of primary care practice	<p>"I would have thought that prenatal and the genetics piece of prenatal was a big part of family practice and that was a bit of a wake-up call for me. I recognize that what we have to offer, as valuable as we think it is, is only a small piece of a primary practitioner's day-to-day existence." (FG 1 Participant 2/ MD)</p> <p>"I would kind of feel that I do not really know how much they are going to know about the condition and how much I need to tell them in the letter. Do I need to send them information or I am insulting them if I give them references. I kind of do not really know how much they know, how much they are prepared to take on and how much time they have to spend in their own practice looking up some rare condition that, you know, we might have only ever seen once or twice here. It would be nice to kind of know what they need from us." (FG 3 Participant 7/GC)</p>

first tier investigations for a patient waiting for genetics consultation (e.g. chromosomal microarray for autism spectrum disorder).

Hamilton et al. (2014) argue that it is unlikely genetic services will be taken up into the health care system by simple diffusion. They state it "will require development of targeted organizational supports to strengthen the likelihood of adoption and implementation". New methods of service

delivery promise the most impact. These might include patient care plans, genetic counselors in primary care clinics, and electronic consultation. Development of patient care plans giving PCPs clear direction about investigations they might undertake while awaiting a genetics consultation may improve timeliness as well as quality of referrals. Having a genetic counselor in a primary care clinic or as a "buddy" for questions by email or phone (Carroll et al. 2019), electronic consultation and telemedicine may increase access and enable more appropriate referrals (Flannery, 2018; Stoll et al. 2018). In one study, PCPs highly valued electronic consultations with geneticists and avoided a third of planned referrals (Bhola et al. 2019).

Our study described the critical role GHPs saw for PCPs in providing genomic medicine now and in the future and the need to support that role and work together more effectively. Lack of coordination between genetics and non-genetics services was described by Pearce et al. (2019) in their systematic review of delivering genomic medicine. Battista et al. (2012) also speak of the importance of coordination of genetic services and suggest that the future may call for the "reconfiguration of professional roles and responsibilities" and "forging of new relationships". They suggest that new "enhanced primary care models" are needed such as situating genomic services close to or in primary care, perhaps delivered by general/nurse practitioners with special skills in genetics or genetic counselors, with communication assisted by information technology (Cohen et al. 2009; Elwyn et al. 2005; Kromberg et al. 2006; Westwood et al. 2006). These models would enhance PCPs' knowledge, and likely ensure more effective case-management (Cohen et al. 2009; Battista et al. 2012). Harding et al. (2019) have proposed a framework for bridging primary and specialty genetic care through a progressive stepped model, whereby clinicians provide genetic services to the extent of their knowledge and comfort, progressing to genetic consultation only when necessary. New service delivery models and collaborative working arrangements such as these might enable the expanded roles and competencies needed for the future.

Table 5 Theme #3: relationship between GHPs and primary care

Theme	Illustrative quote
Relationship between GHPs and primary care:	
Possible ways to improve genomic services	<p>I think a [GHP] presence is useful, but I do not think from a resource perspective how useful it is. I actually sat in an oncology clinic for a year trying to educate. I think just being there they can ask about this or that. Because you are sitting there present, it actually has an impact. Now I really think the oncologists at least at "X" hospital are really on the ball who to refer and not to refer.(FG3 Participant 1/GC)</p> <p>I think it would be nice if they knew that they could call us and ask us before they did something like send a sample, send a referral or something like that. You know, we are supposed to be a resource; that is what we are here for. (FG1 Participant 1/ Lab)</p> <p>But, to just have that sort of built into it so once a week there is a genetic counsellor who is available for consultation and they can refer patients to them or even have them available for themselves to ask questions about patients they have seen in that week. (FG3 Participant 6/Lab)</p>

Establishing new models of genetic service delivery will not be without challenges. For primary care, to see increased clinical uptake, more evidence for the clinical relevance, cost-effectiveness and improved patient outcomes from genomic medicine will be needed (David et al. 2015; Hamilton et al. 2014; Manolio et al. 2013). In addition, innovative PCP educational initiatives will be needed such as “pushing out” primary care relevant e-resources (Carroll et al. 2016a), primary care specific evidence-based practice-friendly point-of-care tools (Carroll et al. 2011, 2014), up-to-date web resources (American Academy of Pediatrics, 2020; Genetics Education Canada: Knowledge Organization, 2020; Genomics Education Programme, 2020; National Institutes of Health, 2020; The Jackson Laboratory, 2020), educational on-line modules and courses (Houwink et al. 2014; Jackson et al. 2018; Paneque et al. 2016, 2017; Reid et al. 2016; Rubanovich et al. 2018), and clear practice guidelines (Carroll et al. 2016a; Pearce et al. 2019). Practice redesign will be important to fully integrate family history and genetic information into the electronic health record with risk alerts and clinical decision support tools (David et al. 2015; Flannery, 2018; Houwink et al. 2013; Manolio et al. 2013; Pearce et al. 2019). It will be important to use the principles of implementation science in integrating genetic medicine into primary care (Greenhalgh et al. 2004), choosing disorders with relevance for primary care and with the strongest evidence for benefit (Manolio et al. 2013).

On the genomics side, new models of service delivery are being developed including use of genetic counselor extenders, genetic counseling assistants, and implementation of technology, on-line education, decision tools and results, telephone and group counseling, to improve efficiency and reach more patients (Flannery, 2018; Stoll et al. 2018). Increased human resources in genetics will be needed, including genetic counselors, clinicians with genetics training, and clinicians capable of “interpreting patients’ genetic information and advising clinicians on appropriate actions to be taken” (Manolio et al. 2013). New genetics inter-professional health teams will be needed to support PCPs and meet patient needs as new genetic tests and technologies appear (Battista et al. 2012; David et al. 2015). Patient and community engagement will be needed in this health system change. GHPs will be vital in this changing landscape of genomic medicine but will likely face their own challenges in role redefinition and responsibility.

Strengths and limitations

To our knowledge, this is one of the first studies reflecting the views of GHPs about PCPs and their role in genomic medicine now and in the future. A limitation is that the participating GHPs were from one province in Canada, where there is a publicly funded health care system. However, our

findings were consistent across the focus groups and interviews, covering a diverse group of professionals and locations. We did not return our findings to participants for validation, which potentially could have modified our results.

Future research

Future research is needed to determine if the findings of this study have relevance in other health care systems. Additionally, health services research is needed to determine if some of the new models of genetic service delivery proposed in this paper can be implemented successfully, such as elements of “mainstreaming” genetic testing by PCPs, improving PCPs’ genetic knowledge through an improved consultation process (consultation letters and/or electronic consultation) and genetic counselors/nurses either embedded in PCP clinics or serving as designated resources.

Conclusions

GHPs perceived a critical role for PCPs in genomic medicine. While they acknowledged PCPs’ lack of knowledge and practice challenges, they recognized the importance of a PCP “hub” model for delivery of genomic medicine, and the corollary need for some adaptations from GHPs to meet PCPs’ needs. New, coordinated, policy direction is needed for a truly transformative change to occur in the provision of genomic medicine. The challenge will be to mandate and enable better communication, collaboration and different models of practice, perhaps embedding GHPs in primary care.

Author contribution JC: conceptualization, formal analysis, funding acquisition, methodology, writing—original draft.

SM: conceptualization, data curation, formal analysis, methodology, project administration, writing—review and editing.

FM: conceptualization, formal analysis, methodology, writing—review and editing.

BW: conceptualization, formal analysis, methodology, writing—review and editing.

JP: conceptualization, methodology, project administration, writing—review and editing.

JA: conceptualization, formal analysis, funding acquisition, methodology, writing—review and editing.

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Data availability Data will be provided upon request.

Code availability Not applicable.

Declarations

Ethics approval and consent to participate Ethics approval was obtained from the Children's Hospital of Eastern Ontario Research Ethics Board (REB). Informed consent was obtained from all participants as required by the REB.

Conflict of interest The authors declare no competing interests.

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