

# Primary care providers' experiences with and perceptions of personalized genomic medicine

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## Abstract

**Objective** To assess primary care providers' (PCPs') experiences with, perceptions of, and desired role in personalized medicine, with a focus on cancer.

**Design** Qualitative study involving focus groups.

**Setting** Urban and rural interprofessional primary care team practices in Alberta and Ontario.

**Participants** Fifty-one PCPs.

**Methods** Semistructured focus groups were conducted and audiorecorded. Recordings were transcribed and analyzed using techniques informed by grounded theory including coding, interpretations of patterns in the data, and constant comparison.

**Main findings** Five focus groups with the 51 participants were conducted; 2 took place in Alberta and 3 in Ontario. Primary care providers described limited experience with personalized medicine, citing breast cancer and prenatal care as main areas of involvement. They expressed concern over their lack of knowledge, in some circumstances relying on personal experiences to inform their attitudes and practice. Participants anticipated an inevitable role in personalized medicine primarily because patients seek and trust their advice; however, there was underlying concern about the magnitude of information and pace of discovery in this area, particularly in direct-to-consumer personal genomic testing. Increased knowledge, closer ties to genetics specialists, and relevant, reliable personalized medicine resources accessible at the point of care were reported as important for successful implementation of personalized medicine.

**Conclusion** Primary care providers are prepared to discuss personalized medicine, but they require better resources. Models of care that support a more meaningful relationship between PCPs and genetics specialists should be pursued. Continuing education strategies need to address knowledge gaps including direct-to-consumer genetic testing, a relatively new area provoking PCP concern. Primary care providers should be mindful of using personal experiences to guide care.

### EDITOR'S KEY POINTS

- Implementation of genomic medicine within primary care has been limited. Challenges to implementation include limited access to genomic medicine expertise and tests and lack of clinician awareness.
- Primary care providers described profound lack of knowledge about direct-to-consumer genetic testing and felt challenged to support patients in this area. They were unsure about their desired level of involvement in genetic testing but expressed concern that it might be a growing trend and that their involvement might be inevitable owing to the trusting physician-patient relationship.
- Primary care providers' experiences, both professional and personal, as well as their knowledge of personalized medicine and genetic testing, influenced their views on and desired role in personalized medicine. To enhance providers' confidence in managing personalized medicine issues, increased personalized medicine knowledge, closer ties to genetics specialists, and relevant personalized medicine resources accessible at the point of care are required.

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# L'expérience qu'ont les soignants de première ligne de la médecine génique personnalisée et ce qu'ils en pensent

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## Résumé

**Objectif** Vérifier l'expérience qu'ont les soignants de première ligne (SPL) de la médecine personnalisée, ce qu'ils en pensent et le rôle qu'ils voudraient jouer dans ce type de médecine, particulièrement pour le cancer.

**Type d'étude** Étude qualitative utilisant des groupes de discussion.

**Contexte** Des équipes interprofessionnelles de soins primaires de régions urbaines et rurales de l'Alberta et de l'Ontario.

**Participants** Cinquante-et-un SPL.

**Méthodes** On a organisé des groupes de discussion dont les conversations ont été enregistrées sur bande magnétique. Ces enregistrements ont été transcrits et analysés au moyen de techniques inspirées de la théorie ancrée qui comprend un codage, l'interprétation de modèles dans les données et des comparaisons continues.

### POINTS DE REPÈRE DU RÉDACTEUR

- La médecine génique est encore très peu utilisée au niveau des soins primaires. Les obstacles à sa mise en œuvre comprennent un accès limité à de l'expertise et à des examens appropriés, et le fait que les cliniciens connaissent mal ce domaine.
- Les soignants de première ligne (SPL) ont avoué avoir très peu de connaissances sur la façon d'utiliser les tests génétiques offerts directement au consommateur et ils se sentaient donc mal à l'aise d'aider les patients dans ce domaine. Ils ne savaient trop à quel point ils désiraient participer aux tests génétiques, mais se disaient préoccupés par le fait que cette tendance pourrait s'accroître et que leur contribution pourrait devenir inévitable compte tenu de la relation de confiance qui existe entre médecin et patient.
- L'expérience tant professionnelle que personnelle qu'ont les SPL de la médecine personnalisée et des tests génétiques de même que leurs connaissances dans ce domaine influençaient leur opinion sur ce type de médecine et leur intérêt à y participer. Si on veut que les SPL soient plus à l'aise de gérer les issues propres à la médecine personnalisée, on devra améliorer leurs connaissances dans ce domaine de la médecine, resserrer leurs liens avec les spécialistes de la génétique et s'assurer que les ressources appropriées à la médecine personnalisée soient accessibles à l'endroit où les soins sont donnés.

Cet article a fait l'objet d'une révision par des pairs.  
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**Principales observations** Il y a eu 5 groupes de discussion auxquels ont participé 51 SPL; 2 ont eu lieu en Alberta et 3 en Ontario. Les participants ont mentionné leur peu d'expérience dans la médecine personnalisée, précisant que le cancer du sein et les soins prénataux étaient les principaux domaines auxquels ils avaient été confrontés. Ils se disaient préoccupés par leur manque de connaissances, se fiant parfois à leur expérience personnelle pour orienter leur attitude et leur façon de faire. Ils prévoyaient qu'ils auraient inévitablement un rôle à jouer en médecine personnalisée, en raison surtout de la confiance que les patients leur accordent en leur demandant leur avis; toutefois, il y avait une préoccupation sous-jacente à propos de la somme des informations et de la vitesse des découvertes dans ce domaine, en particulier dans le cas des tests génétiques personnels offerts directement au consommateur. De meilleures connaissances, des meilleurs contacts avec les spécialistes de la génétique, et des ressources fiables et adéquates pour la médecine personnalisée, accessibles à l'endroit où sont donnés les soins, ont été mentionnées comme des mesures importantes pour assurer une mise en œuvre réussie de la médecine personnalisée.

**Conclusion** Les SPL sont disposés à discuter de médecine personnalisée, mais ils ont besoin de meilleures ressources. On devrait utiliser des modèles de soins qui favorisent une relation plus significative entre les SPL et les spécialistes de la génétique. Il faudra faire appel à des stratégies de la formation continue pour parfaire les connaissances, notamment dans le cas des tests génétiques offerts directement au consommateur, un domaine relativement nouveau qui suscite certaines préoccupations chez les SPL. Ces derniers ne devraient pas trop se fier à leur expérience personnelle pour guider les patients.

Advances in genomic medicine promise enhanced understanding of disease and individualized, patient-centred care. The terms *personalized medicine* or *precision medicine* are used to signify “prevention and treatment strategies that take individual variability into account,” blending molecular or DNA profiling with clinical or pathological indices.<sup>1,2</sup> To realize its benefits, personalized medicine must be fully integrated into primary care; however, there has been limited implementation of genomic medicine within primary care. Challenges include a paucity of evidence of clinical usefulness, lack of clinician acceptance, limited access to genomic medicine expertise and tests, poor integration of genomic test results and clinical decision support into the electronic medical record (EMR), and limited understanding by patients and clinicians.<sup>3,4</sup> Barriers to integration of genomic medicine frequently mentioned by primary care providers (PCPs) include lack of knowledge about genetics and genetic risk assessment, limited access to genetic services, and time constraints.<sup>5,6</sup> While “road maps” for integration of genomic medicine into primary care have been proposed,<sup>4,7</sup> system issues and knowledge gaps negatively affect their integration and emphasize the value of understanding local circumstances and PCPs’ attitudes toward genomic medicine before designing implementation plans.<sup>3</sup>

The objective of our study was to assess PCPs’ experiences with, perceptions of, and desired role in personalized medicine, with a focus on cancer. We chose cancer because oncology has been identified as a “clear choice for enhancing the near-term impact of precision medicine”<sup>2</sup> given that it is common, a leading cause of death, and there has been good progress in identifying germ-line mutations conferring an increased cancer risk.<sup>2</sup> Our study was conducted as part of the CanIMPACT (Canadian Team to Improve Community-Based Cancer Care along the Continuum) initiative, which has the overall objective of improving integration between primary care and oncology specialist care including responding to the challenges of personalized medicine.

## METHODS

### Research design

A qualitative approach informed by grounded theory was used. We chose grounded theory as it has been widely used in health sciences and uses both inductive and deductive methods, which were helpful in understanding processes and interactions of PCPs in the context of personalized medicine.<sup>8,9</sup> Focus groups (FGs) were used to capture the subjective meaning of the experience of personalized medicine from PCPs’ perspectives.<sup>10,11</sup>

Ethics approval was obtained from the research ethics boards at Mount Sinai Hospital in Toronto, Ont, and

the University of Alberta in Edmonton, with administrative approval from the University of Toronto research ethics board.

### Recruitment

We used purposeful sampling<sup>12</sup> to identify potentially eligible primary care practices. A list of suitable inter-professional primary care team practices in Ontario and Alberta was compiled based on suggestions from primary care provincial cancer leads and the research team. Inclusion criteria were rural or urban practice, full-scope primary care practice, and a minimum of 4 participants per practice including at least 2 FPs. Letters of invitation were sent to the lead PCPs of each practice followed by a call from the research team (J.C.C. or D.P.M.) to answer questions. If the lead PCP expressed interest, project information was sent to the office managers to distribute to their teams’ primary care health professionals.

### Data collection

Focus groups of approximately 60 minutes’ duration were conducted using a semistructured interview guide developed by the research team and were audiorecorded. The interview guide is available from the corresponding author upon request. We collected demographic data from baseline questionnaires administered prior to each group. Following an initial open discussion on personalized medicine, we identified that personalized medicine could include risk stratification on the basis of family history with screening or management tailored to risk, or it could include genetic testing. The interviewer used the broad headings in the interview guide reflecting open-ended questions with more specific probes if those topics had not been discussed spontaneously toward the end of the interview.

### Analysis

Focus group recordings were transcribed verbatim, anonymized, and analyzed using techniques informed by grounded theory<sup>8</sup> including coding, interpretations of data patterns, and constant comparison method.<sup>13</sup> Co-investigators (J.C.C., D.P.M., T.M., M.A.O., R.H.) read the same 2 transcripts independently and met with team members to develop a coding manual. Remaining transcripts were subsequently coded line by line by the research assistant (T.M.). Team members periodically met to review and refine codes, and any discrepancies were resolved through discussion. NVivo 10 software was used for data management.

## FINDINGS

We conducted 5 FGs with interprofessional PCP teams, 3 in Ontario and 2 in Alberta (4 in urban or semiurban settings

and 1 in a rural setting). A total of 51 individuals participated with 46 completing baseline questionnaires. **Table 1** presents participants' demographic characteristics.

Participants' experiences, both professional and personal, as well as their knowledge of personalized medicine and genetic testing, influenced their views on and desired role in personalized medicine. Overall, PCPs were engaging infrequently with personalized medicine, and owing to lack of knowledge were either reverting to personal experiences to inform clinical practice or referring patients frequently to genetics clinics for genetic counseling or testing. Increased personalized medicine knowledge, closer ties to genetics specialists, and relevant, reliable personalized medicine resources accessible at the point of care were cited as important to enhance confidence in managing personalized medicine issues.

### Themes

**Infrequent experience with personalized medicine in primary care.** When asked to share their understanding of personalized medicine, PCPs' descriptions were vague but included phrases such as "wave of the future" and "what medicine will be." Their personalized medicine experiences were mainly in cancer, particularly genetic testing for hereditary breast cancer, with some experiences in prenatal care. They described almost no experience with colorectal or other cancers. Patients were frequently described as the drivers of genetic testing and referrals, and PCPs sometimes believed that

patients knew more about available genetic tests than they did.

I wouldn't say I've got tons of personal experience in my practice, other than that I've seen some of the commonly identified cancer screening, specifically *BRCA 1, 2*, coming back and being investigated within families, that's probably the one being the most prominent .... I'm excited about the prospect of what personalized genomic medicine might offer us down the road, but my knowledge of it is pretty limited. (FP, FG5)

**Primary care providers' lack of knowledge.** A pervasive theme was limited knowledge of personalized medicine. Primary care providers had little awareness of developments in personalized medicine, available tests, and triggers for appropriate referral for genetic counseling and testing. Knowledge affected practice, with some PCPs, based on responses provided, not referring when appropriate nor recognizing the benefits of genetic testing. A common example was lack of awareness of genetic tests for hereditary colorectal cancer and how results might change screening recommendations. Some PCPs acknowledged not knowing enough about personalized medicine and yet, based on responses provided, others were not cognizant of their incomplete or incorrect knowledge.

Primary care providers who were aware of their lack of knowledge expressed concern and anxiety. Moreover, there was apprehension over the magnitude of knowledge that was required before they were confident to discuss genetics with patients.

I think that even to have the conversation about genetic testing with the patient, it's such a big conversation 'cause they're going to ask us these questions that we don't even have the answers to, like what is the benefit for me? ... If you don't know what the potential treatment options are with a positive [result] or the potential treatment options of a negative [result], how can I even have an informed conversation with my patients? (FP, FG4)

Most PCPs expressed an almost complete lack of knowledge about direct-to-consumer genetic testing (DTC-GT). There was much emotion associated with this area, with some PCPs describing it as "scary" and others worried about a potential deluge of patients requesting care after testing privately. Primary care providers with actual experience of patients with DTC-GT results voiced frustration at their delayed involvement and concern for patients who had been tested without prior counseling or thought of implications such as the privacy of their genetic information or the effect on their ability to obtain insurance. There was also concern for

**Table 1. Demographic characteristics of participants (N = 51): Mean age of participants was 44 years (range 23 to 65).**

CHARACTERISTICS	N (%)*
Practitioner type	
• FP	30 (59)
• Registered nurse	11 (21)
• Nurse practitioner	2 (4)
• Physician assistant	1 (2)
• Family medicine resident	4 (8)
• Medical student	1 (2)
• Other	2 (4)
Sex	
• Female	34 (76)
• Male	11 (24)
No. of years in practice	
• < 10	18 (42)
• 10-19	3 (18)
• ≥20	25 (40)

\*Not all respondents answered all questions.

the provider. "If something goes wrong, someone is going to look for who is responsible and where you did fall through the gap." (FP, FG3).

**Primary care providers relying on personal experience.** Many PCPs described a context wherein their personal experiences sometimes informed personalized medicine clinical issues. Past personal experience or the experiences of family and friends also influenced PCPs' attitudes toward and perceptions of personalized medicine. When relating personal stories, providers voiced that personalized medicine was helpful in diagnosing disorders but believed it made little difference to the patient's outcome.

So, coming from personal life, my mom and my aunt both had breast cancer. Do I want to go and get tested, you know? Know what? I will go for the screening when it's due and that's what I will do, but do I want to know if genetically I would have that gene, that I'm more at risk, how will that affect the way I'm thinking, am I going to go for prophylactic [surgery]? No. So, I kind of ... maybe I'm using my personal feeling, but I may apply it to my patients because, really, why do you want to know, like why, why? (FP, FG4)

**Primary care providers' perception of inevitable involvement in personalized medicine.** Despite infrequent experiences, PCPs speculated that personalized medicine might become increasingly entrenched in primary care. Although concerns exist about insufficient knowledge and a potential "forthcoming tidal wave" of patients seeking genetic testing, many see inevitable involvement in personalized medicine.

Primary care providers' relationship with their patients was cited as an important reason for involvement in personalized medicine, with patients often seeking their advice because of this trusting relationship and because PCPs' knowledge of patients' medical and personal histories was valued by patients in decision making. "If we don't do it, who will? ... [A]nd who's going to know their history better than us?" (FP, FG4)

**Unfamiliar relationship with genetics specialists and clinics.** Primary care providers described frequently being unsure when a patient should be referred and, consequently, most referred liberally to genetics clinics. They reported it as erring on the side of caution but sometimes described "referring willy nilly" owing to lack of knowledge of referral guidelines. Participants recounted primarily positive experiences, with genetics clinics seen as "reliable go-to places" that provide outstanding care. Primary care providers appreciated genetics consultation letters and described them as an excellent source of education. However, they observed

that relationships and communication patterns were different from those with other specialists. Few personal connections exist and many never connected directly with genetics specialists. One participant described the referral process as, "You feel like you're referring to the abyss." (FP, FG2)

"I speak to oncologists on the phone often, but I've never spoken to somebody from genetics before. I wouldn't actually know where to start ... I don't know anybody in that field." (FP, FG4)

Another difference noted was that genetics clinics sometimes declined referrals, generally because the family history did not meet guidelines for genetic testing. When this occurred, PCPs reported no guidance on what should happen next and were left wondering about patient management. Primary care providers described feeling somewhat criticized because they had flagged the patient as concerning enough to require referral. "What do clinics need?" was a common question highlighting PCPs' desire for detailed referral guidelines.

**Primary care providers' role in personalized medicine.** Initially, participants seemed uncertain what role they might have in personalized medicine, but as discussion proceeded, they identified that their role included taking family histories, conducting risk assessments, and referring to genetics clinics for genetic counseling or testing. They reported that taking a good family history was an important role for PCPs. "I think what has been missing in family medicine, as we've seen the growth and importance of genetics, is the ability to take a good history." (FP, FG1)

Primary care providers also described their role as being a resource to patients, answering questions and responding to requests for genetic testing. Currently, this was described as a more reactive role with patients driving the request for testing. Some PCPs questioned, however, whether they should more proactively identify patients who might benefit from genetic tests.

Counseling patients on the risks and benefits of genetic testing was also seen as a responsibility, with some PCPs seeing themselves as gatekeepers, particularly in relation to DTC-GT. "So as a physician, I would want to know exactly what I had to counsel my patients before even accessing the test and what the implications of that would be." (FP, FG3)

A minority of PCPs did not want a role in genetic testing at all, describing lack of knowledge as an "out."

I'm not sure that our role as a family doctor is becoming some kind of a specialist in genetic medicine. I think we come back to basic concepts as far as family medicine is concerned, which is taking a good history and physical ... I'm not in a position to start trying to interpret the results. (FP, FG1)

**New models of care and resources.** When asked about system changes that would facilitate personalized medicine integration, participants suggested having a “buddy” in a local genetics centre or an in-house nurse or FP with genetics training as an internal expert.

I think a buddy would be great .... It's always helpful if you have a go-to person who can help you out. So over the years we all develop people that we can reliably get good opinions from and as you get to know them personally ... you get a little bit better service I think, and sometimes a phone call will solve your issue and it won't necessarily need to go through the whole process. (FP, FG3)

Recommendations were also provided for personalized medicine resources. Primary care providers described needing tools that were easily accessible, up to date, from a reliable source, and reflecting local resources, both online and available at the point of care. **Table 2** provides further details on PCPs' needs in personalized medicine. “Well I guess for me it would be point of care, it'd be right there ... right in front of the patient ... either online or downloadable ... so that there's a decision-making algorithm.” (FP, FG3)

Additional quotes to support and illustrate findings are found in **Table 3**.

**Table 2.** Identified information and resource needs in personalized medicine

REQUIREMENTS	IDENTIFIED NEEDS
Information	Participating PCPs requested information on the following: <ul style="list-style-type: none"> <li>• mutations and genetic causes of cancer</li> <li>• guidelines for screening and referral</li> <li>• referral process with genetics clinics</li> <li>• available genetic tests</li> <li>• preventive treatment</li> <li>• benefits of testing</li> </ul>
Resources	Reliable, updated, non-biased information source Point-of-care tools (eg, EMR algorithms and decision support tools) Up-to-date Web-based resource (an app) Education sessions by FPs with expertise Patient handouts Resource within the clinic (eg, a nurse, nurse practitioner, or FP with expertise in genetics)

EMR—electronic medical record, PCPs—primary care providers.

medicine experience and knowledge, and relied on personal experiences to inform their practice in some circumstances. Primary care providers were aware of the usefulness of genetic testing with respect to identifying individuals or families with high cancer risk; however, this was primarily limited to breast cancer. They had limited knowledge of genetic testing for colorectal cancer or applicability of personalized medicine to areas outside of cancer, suggesting some benefits might be overlooked.

Our findings supported those of another Canadian study in which 90% of FPs were aware of genetic testing for breast or ovarian cancer, but only 53% were aware of genetic testing for colorectal cancer.<sup>14</sup> In that study, 67% had ever referred to a hereditary cancer clinic with a mean of 3 patients in the past year. These findings are similar to a survey of US PCPs in which respondents reported ordering genetic testing for disease susceptibility 1 to 2 times per year; however, 32% had never ordered a genetic test.<sup>15</sup> In this study 73% had heard of pharmacogenetic testing but 80% had never ordered a test. In another US survey of PCPs, 19% had ordered a genetic test in the previous 6 months, with cancer risk testing being most frequent.<sup>6</sup>

### Lack of knowledge

Primary care providers' lack of knowledge of personalized medicine has been reported internationally,<sup>5,16-19</sup> with many proposing that improvement in knowledge and skills is the first step toward integrating genetics into primary care.<sup>7</sup> Knowledge gaps reported by others<sup>20</sup> are similar to those identified by our participants. In a survey of Canadian physicians (FPs, cardiologists, and oncologists) on personalized medicine, only 21% agreed they were sufficiently informed about personalized medicine and 29% said they were able to interpret results of genetic tests.<sup>18</sup> A similar survey of US PCPs about personalized medicine found that while 55% reported feeling confident in interpreting genetic test results, only 22% thought their training in genetics was sufficient to work with patients who had genetic testing.<sup>19</sup>

A new concerning finding is that some PCPs rely on their personal experiences to guide patients. This is not surprising given the minimal training in genetics in medical school and residency,<sup>21</sup> but it might result in incorrect or biased information for decision making.

Providers described a profound lack of knowledge of DTC-GT and felt challenged to support patients in this area. This is concerning as patients might ask their PCPs about testing. A US study found that 19% of PCPs reported having patients ask questions about DTC-GT or bring in test results.<sup>22,23</sup> They found that only 39% were aware of DTC-GT, 85% believed they were unprepared to answer patient questions, and 74% wanted to learn more.<sup>22</sup> In one study, 63% of respondents who had purchased personalized DTC-GT planned to share their

## DISCUSSION

Primary care providers described limited personalized

**Table 3. Examples of participant verbatim quotations**

THEMES	PARTICIPANT QUOTATIONS
Infrequent experience with personalized medicine in primary care	<p>"In practice, the only genetics I've come across would be the <i>BRCA</i> genes or in prenatal screening the cystic fibrosis as well comes up. So outside of that I've come across more the, the marketing advertisements for these companies that refer to it as well, whether I've seen advertisement on TV or hear it on a podcast or something and CBC <i>Marketplace</i> did a good comparison, a review of the different ones" (FP, FG5)</p> <p>"I think we're seeing more and more people may come up and if we are not prepared, you know, that would be a disconnect in there. Because sometimes our patients are more knowledgeable about those things. And I think my best understanding is from the ads I see on TV, you know, in terms of 'Send me your spit and we'll tell you exactly how your life is going to shape up in the future.' So, I'm sure we're going to see more and more people, that they coming and talking about some genes or mutations that I am not even aware of it. So, that's where I see the gap for me, because I have no training on any interpretation of any of those" (FP, FG1)</p> <p>"My experience and concern with genomics actually is to do more with kind of the field and where it's heading, and sometimes I think that the whole genomics project is gonna to be very upfront on diagnosing or on identifying traits but very short on identifying interventions and so that's going to create millions of profiles of people at risk for things with very little to offer them and I think the intervention's gonna be, you know, 1 or 2 decades down the road. So what are the ethics of alerting all these people to shortfalls of their genetics and yet you can't do anything about it and where does that information go and, you know, how do people handle it personally, right?" (FP, FG3)</p>
PCPs' lack of knowledge	<p>"I would say that one of my concerns would be the volume of information that you're talking about. I mean, you're talking about 200 genetic tests and I maybe know about, you know, <i>BRCA</i>, and I know about a few other things and I know about the stuff we do prenatally and, boy, I'm probably missing an awful lot of stuff there" (FP, FG1)</p> <p>"In that particular instance or instances they came to me, the material [DTC-GT] was already done. I had nothing to do with organizing it or discussing it or counseling prior to that and it was not anything that they were particularly concerned about at that point ... I had no level of expertise that could help them identify ... they were literally long lists of numbers with no qualifying information on the side" (FP, FG3)</p> <p>"And I think the scary ... scary might be too ... but the scarier part of these testing and being able to do a full genetics panel is we're not comfortable with these tests and we don't know, necessarily know what to do with them, which makes it harder" (FP, FG1)</p>
PCPs relying on personal experience	<p>"I have to declare a little bit of a bias. As you were talking, I was thinking, 'Oh, OK.' I quite by accident found out in medical school that I am a heterozygote for <math>\alpha</math>-1 antitrypsin deficiency ... so then you have to go talk to a geneticist to know what does this mean and then you realize there's nothing that can be done about it. All that I did was at 25 years of age, find out I'm a heterozygote for something that might cause me lung disease and liver disease in the future, right, and I've lived for the last 30 years wishing I didn't know about it" (FP, FG3)</p> <p>"Again referring back to this case, it wasn't a doctor-patient relationship, it was a physician-physician or a friend relationship, and after many agonizing discussions, really it came back to, as we use the expression in [town name], 'ice the puck back' to the specialist, and even at that level, there was such a huge discrepancy in what was being advised—chemotherapy, surgery, preemptive surgery, whatever—that had that person not been a friend but had been a patient, I would absolutely have said, 'I'm sorry, I don't have that level of expertise,' because even the super-duper specialists in Toronto at that time were not able to ... I mean they obviously helped, there were lots and lots of discussions, but the definitive answer was not ... there wasn't a definitive answer given" (FP, FG3)</p>
PCPs' perception of inevitable involvement in personalized medicine	<p>"But I think that it is inevitable that we will get pulled into this. If my patient comes in and they've sent the test off and the result is sitting there in front of me, I'm going to have to deal with it however I figure out how to deal with it. I don't think I'll have a choice" (FP, FG1)</p> <p>"You know, from a point of view of how we want to manage things versus what comes through the door, we don't really have control over that, in the sense that if the patient decides that they're—without necessarily having that conversation with their family doctor about the pros and cons, which [participant name] alluded to—if they're not having that conversation and they're just coming in to your office with it, I mean, you don't really have that choice, in that respect, right? It's arrived at your door and, and you therefore have to run with it ... I mean, you know, if they have access to that, and they obtain it and they bring it to you because they trust your opinion, we have to have some sort of formulation in terms of how we're going to deal with it" (FP, FG1)</p> <p>"The other thing I wanted to say, I know it's a changing field, but so often our patients will get recommendations from an oncologist or genetic counselor, but they trust us and so they'll come back to ask us to help them with decision making and so I'm not on top of what the current prophylactic regimen and being kept up to date on that would be good, you know" (FP, FG2)</p> <p>"Yeah, they're the ones educating us because they come back and they say ... they sometimes will say, 'What do you think?' And I actually don't know but it sounds like the letter [genetics consultation letter] that I got, this is probably the best idea. You put it right back onto them, but a lot of your patients if you've been with them for 20 some years, they have a trust in you, even if they're aware that you actually don't have that knowledge they still want your opinion, right?" (FP, FG4)</p>

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THEMES	PARTICIPANT QUOTATIONS
Unfamiliar relationship with genetics specialists and clinics	<p>"I just got this result back, what would you suggest doing?' And that's kind of the relationship we tend to develop with our specialists at the hospital anyways, since you usually have somebody you can call up and say, 'I have no idea what to do next,' but there's no geneticist at [workplace name], so I've never ... I don't recall really calling the clinics very often because I do find that the feedback I get from them is pretty good, but if I was stuck it would be awesome to be able to call someone" (FP, FG1)</p> <p>"Yeah, I had a patient with severe osteoporosis since her early 40s and she came and she's also a professor and she said, 'Oh there is this testing they're doing for hereditary testing for the patient.' She gave me the information. That's when I said, 'patients never come with any information right, so I do the referrals, send the referral, and the letter I got 2 weeks later ... 'This testing is not available in Canada,' and then ... and then at the end of the letter, which I think is like a reprimand to me because that's what I told the patient, but that's what this specialist put in the letter, and it wouldn't really have mattered even if there's genetic testing for osteoporosis, it will not change the management of this patient .... I thought that was harsh ... we're managing her osteoporosis, but what are we going to do about the information whether you have the gene or not, so not only I was told it's not available, but how would that change the management?" (FP, FG4)</p>
PCPs' role in personalized medicine	<p>"I think one of the first things would probably be proper identification of those patients that are appropriately sent to genetics or should be sent towards genetics, and so when we're reviewing family histories, having a good understanding as to who should be appropriately refer[red], or who's appropriately referred and who doesn't require that, that potential step of genetic referral. And yeah, so I think, I think we play a fairly handy role from that perspective and similarly, although the discussion about the increased or modified screening might take place at the genetics clinic, it usually warrants some further discussion with a patient's primary care provider and then frequently we are the ones who are then implementing and monitoring that, ensuring that" (FP, FG5)</p>
New models of care and resources	<p>"If I had a way—and [participant name] alluded to it earlier—of taking a really refined family history and, yeah maybe there's a callback that has to happen afterwards or a follow-up appointment or more information gathering or whatever, that if I knew how to do that and then I had some very clear guidelines of what should trigger my genetics referral, then maybe I wouldn't end up sending off somebody that I didn't really think they needed genetics testing but I wanted confirmation right now that I'm not missing something, which I'm probably doing right now, because I'm not 100% sure all the time. So ..." (FP, FG1)</p> <p>"If I have a site I can go quickly read, so if someone asks me, then I can always say, OK, well, I'll look it up and get back to you." (FP7)</p> <p>"Or you can read it together there, yeah, it's more just having a place to know that they're, the information's there and available and reliable." (FP2)</p> <p>"And up to date as well .... Because it's obviously evolving quickly, so if you know of some place that is actually actively managing their website so you can say, OK, if I go here, I'm going to find out what's new from 2 months ago, not from 2 years ago .... Yeah, I think that would be good." (FP5)</p> <p>"And what's actually available here ...." (FP2)</p> <p>"Locally" (FP4) (FG4)</p> <p>"I'm kind of app friendly for that kind of thing, if there was something either online or downloadable where it was a simple, you know—menu-driven kind of informational thing so that there's a decision-making algorithm. I don't mean like a comprehensive database, a decision algorithm, just you know, along those lines anyways" (FP, FG3)</p>

DTC-GT—direct-to-consumer genetic testing, FG—focus group, PCP—primary care provider.

results with a PCP; however, at 6-month follow-up only 27% reported having done so.<sup>24</sup> Among those who discussed results with their PCPs, 35% were very satisfied and 18% not at all satisfied, with some citing lack of PCP interest.<sup>24</sup> In our study, PCPs were unsure about their desired level of involvement in DTC-GT, but expressed concern that it might be a growing trend and that the trusting physician-patient relationship might mean inevitable involvement.

### Relationship with clinical geneticists and genetics counselors

Primary care providers valued genetic consultations as providing excellent patient services and informative consultation letters. However, they described almost no connection with genetic services; not knowing who to contact with a question; frustration with not knowing when to refer; and "inappropriate" referrals being

rejected—experiences different from those with other specialists. Studies have shown that where genetic services have not been systematically adopted, there have been barriers to their use.<sup>16</sup> Understanding the organization and role of clinical genetic services has been reported as an important educational need.<sup>25</sup> It is possible that closer relationships between PCPs and genetics specialists might mitigate the feelings of "referring into the abyss," feeling rebuked when referrals are declined, and reliance on personal experience. Not every hospital has a genetics department, thus PCPs refer elsewhere, meaning links that naturally exist with other specialists owing to proximity are missing in genetics.

### New models of practice

The inevitability of PCPs having a role in personalized medicine, combined with lack of knowledge and concern over the magnitude of information and discovery,




point to an urgent need for both effective knowledge dissemination strategies and systemic practice changes. Similar to others, our findings suggest the need for continually updated, easily accessible point-of-care tools from a reliable source integrated into the EMR with decision support and adequate reimbursement.<sup>3,7,16,26</sup> Integration of an individual's genomic data into the EMR could potentially contribute to diagnosis, treatment, and disease prevention strategies for the patient and family; however, there are many challenges in integrating these data into the EMR and providing clinical support.<sup>27</sup> High-quality educational materials are likely insufficient on their own to increase personalized medicine competency.<sup>26</sup> Websites have been developed to address some of these educational needs (eg, [www.geneticseducation.ca](http://www.geneticseducation.ca), <http://g-2-c-2.org>, <https://www.jax.org/education-and-learning/clinical-and-continuing-education>). New genetic service delivery models need to be implemented and evaluated. These might include having a "buddy" or connection to the local genetics clinic with ready e-mail or telephone access, developing local expertise such as a nurse practitioner or FP with extra training in genomic medicine, or an electronic consultation genetic service.<sup>28,29</sup>

## Limitations

Our sample consisted of team practices with EMRs in Ontario and Alberta. Further studies are needed in different practice models (eg, solo practices). Our FGs contained more physicians than other health care professionals, which might have affected discussion.

## Conclusion

Our findings provide insight into the extent to which PCPs are prepared to discuss personalized medicine within primary care, the resources needed, and potential new models of care to facilitate its successful integration. Models of care that foster awareness of genetic services and a more meaningful relationship with clinical geneticists and genetics counselors should be pursued. Knowledge gaps have been highlighted in other areas of personalized medicine but our study indicates this includes DTC-GT, a relatively new area provoking concern among PCPs. Continuing education strategies should be inclusive of this new area in genomic medicine. Primary care providers should also be mindful of using personal experiences to guide care. 

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### Contributors

All authors contributed to the design of the study. **Drs Carroll, Manca, Sopcak, O'Brien, Heisey, Sawka, Urquhart, and Grunfeld**, and **Ms Makuwaza, Ms Permaul, Ms Schneider, and Ms Versaemel** contributed to the analysis of the study. All authors contributed to writing and editing of the manuscript and have agreed to the version for submission.

### Competing interests

None declared

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