General practitioners' views on genomics, practice and education

A qualitative interview study

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Background and objective
Genomics is moving rapidly into mainstream medicine through clinical genomic testing and consumer-initiated online DNA testing. The aim of this study was to identify Australian general practitioners' (GPs') views on genomics, impact on practice and educational needs to inform continuing education.

Methods
Semi-structured interviews were conducted, with constant comparative inductive analysis and governance from a national taskforce.

Results
Twenty-eight GPs (43% female) were interviewed; 71% worked in a metropolitan workplace. Most initially reported little experience with genetic/genomic tests but, when prompted, recognised encountering genomics, mainly non-invasive prenatal and single-gene tests. Many GPs referred patients for cancer screening to genetic services or specialists. GPs reported needing continuing education and resources, with preferences underpinned by relevance to practice.

Discussion
GPs are integrating genomic testing into care, mainly through prenatal screening, and anticipate further impact. They want diverse and context-dependent education but are unaware of some available resources, such as The Royal Australian College of General Practitioners' *Genomics in general practice* guideline.

GENOMICS IS MOVING RAPIDLY into mainstream medicine,[1] and general practitioners (GPs) increasingly face decision making regarding clinical genomic testing and responding to the demands of consumer-driven (personal and/or online) DNA testing. Genomic testing, including partial or whole-genome sequencing that simultaneously looks for variants in up to 20,000 genes, brings new demands.

There are significant challenges in incorporating genomics into medicine, including into general practice. GPs' lack of knowledge and skills have been reported as major barriers to integrating genetics into their practice in Australia,[2,3] the Netherlands,[4,5] the USA,[6–8] Canada,[9,10] Europe and the UK.[11] Challenges identified by GPs in the literature that particularly relate to genetic (rather than genomic) testing include: a lack of confidence to carry out basic medical genetic tasks,[6,11,12] longer consultation times,[3,8,13] keeping up to date,[14] lack of evidence for tests,[2,8] and knowing who and how to refer to genetic services.[2,7,9,10,15] Information about the education needs of GPs in genomics is limited. In Australia, studies to identify GPs' educational needs in genetics were mainly conducted prior to 2006. In a recent study on providing personal genomic risk information in Australia, GPs were open to the delivery of genomics in primary care, but most felt they were unprepared and had gaps in their knowledge.[16]

The Australian Genomics Health Alliance (Australian Genomics) was established in 2016 with National Health and Medical Research Council funding. Australian Genomics conducts research to inform the integration of genomic medicine into mainstream healthcare and includes a focus on workforce and education research through multiple projects aiming to understand current clinical practice, training, needs and preferences of health professionals.[17]

The aim of this study was to inform continuing education of GPs in Australia by identifying GPs' views on genomics, the impact of genomics on their practice, and their educational needs.

Methods

A national taskforce (n = 15) was convened to oversee all aspects of the study, including design, data collection, analysis and reporting. The taskforce consisted of practising GPs (n = 5), medical professional college representatives (n = 2) and genomic education providers and researchers (n = 8).

The study had University of Melbourne Human Research Ethics Committee approval (1646785.7).

Recruitment of GPs was purposive, with the aim to include participants from all states in Australia, initially via convenience sampling (direct invitation from taskforce members), then via electronic news of the Primary Health Networks, professional colleges and GP education organisations. Interested GPs were mailed participant information and consent forms. Recruitment ceased when

data saturation was reached (ie when no new findings were identified).

This study was guided by a constructivist paradigm, which acknowledges multiple perspectives of a phenomenon based on the context of an individual's own experience.[18] It also takes into account the subjectivity of the participants and the researcher.[19] Therefore, a qualitative approach was deemed appropriate for this study.

Semi-structured interviews were conducted by an experienced educator of GPs (MBC), face to face or by telephone, in 2018 (refer to Appendix 1 for the interview schedule, available online only). Interviewed GPs were reimbursed for their time. For the purposes of this study, the researchers used The Royal Australian College of General Practitioners' (RACGP's) definition of 'genetic' as referring to the study of single genes, and 'genomic' referring to the study of multiple genes and genomic regions.[20]

Interviews were audio-recorded, transcribed verbatim and imported into NVivo qualitative data analysis software (QSR International Pty Ltd. Version 11, 2015). Interview data were analysed using inductive content analysis,[21] which allows for analysis of manifest (explicit) and latent (implicit) meaning.[22] Initially, three researchers each read 2–4 different transcripts for familiarity, then all transcripts were independently coded by MBC and CH, using constant comparison to help identify patterns across transcripts,[23] and codes were grouped to create categories. Categories were discussed by all researchers (who have expertise in qualitative research and/or genomic education) and reviewed and refined iteratively with other members of the taskforce until consensus was reached. This process ensured rigour of analysis as well as coder reflexivity through guidance and discussion.

Results

There were 28 GPs interviewed (Table 1). Females accounted for 43% of interviewees; 71% of interviewees worked in a metropolitan workplace, and 29% worked in a rural or remote location. Clinical experience ranged from general practice registrar to more than 30 years. All Australian states and territories were represented.

No GPs reported any formal training or education in genetics/genomics except for 'a few' lectures in medical school. Only three had attended continuing professional development (CPD) activities on genetics/genomics, and almost all asked for the distinction between genetics and genomics to be explained at the beginning of the interview (as defined in the interview schedule).

There were four different categories of findings, outlined as follows.

Uncertainties around genetic/genomics in general practice

Initially, most GPs reported little experience with genetic or genomic testing. However, when prompted during the interview about different genetic or genomic tests, almost half disclosed they had ordered DNA-based tests, and all had viewed test results. This suggests that they may not always realise the genetic nature of tests.

The test that most GPs had directly ordered or requested was non-invasive prenatal testing or screening (NIPT/NIPS):

> *We've certainly done [NIPT product name] testing at patients' requests which is partially of course a financial issue, people have to pay for that.* [GP10, rural]

Other tests GPs had ordered were mainly Medicare Benefits Schedule–funded single-gene tests such as tests for hereditary haemochromatosis, coeliac disease and the methylenetetrahydrofolate reductase (*MTHFR*) gene test. When asked about preconception carrier screening (also known as reproductive genetic carrier screening), a few GPs discussed NIPT instead, indicating confusion with carrier screening tests. More than three-quarters of GPs reported referring patients to genetic services or specialists. The most common test results viewed by GPs were for hereditary breast or colorectal cancer:

> *They're [BRCA] about the only ones I've ordered myself. If I'm going to order something myself I usually talk to a geneticist or refer them because I don't want to order something that could have difficult ramifications. I don't feel that I'm equipped to counsel the patients adequately when the result comes back.* [GP15, metropolitan]

> *There [are] special criteria [for BRCA] that we adhere to when we deal with family risk, or criteria of patients who, based on that, we refer them to the family cancer clinics.* [GP21, rural]

Some GPs discussed that they had helped patients interpret results of consumer-driven genomic, including pharmacogenomic, tests, although most had never viewed an online DNA test result of a patient and were unsure how to manage these:

> *Someone who has done their own DNA test, which I think is from overseas, and they are given information about their risk of developing certain diseases based*

Table 1. General practitioner (GP) participant characteristics in the study (n = 28)

Characteristic	Number (%) of GPs
Gender	
Female	12 (43%)
Male	16 (57%)
Location of practice*	
Metro	20 (71%)
Rural	5 (18%)
Remote	3 (11%)
Years of practice	
0–9 years	14 (50%)
10–19 years	5 (18%)
20–29 years	5 (18%)
≥30 years	4 (14%)

**Based on Rural, Remote and Metropolitan Area classification*[35]

> *on the genetic information and they come to me to know whether this is legit and I'm not really sure what to tell them.* [GP19, metropolitan]

Lack of evidence and reliability of online DNA testing was a concern for some GPs, who stated they lacked confidence interpreting results to support their patients. In particular, lack of evidence for *MTHFR* testing was concerning for some GPs:

> *Someone who's come in with a piece of paper saying they've had the MTHFR gene positive. 'What does that mean?' And I was … I don't think I really had any great answer to them about the implications of that, apart from that there [were] very limited data as to what that meant in terms of a well person.* [GP24, metropolitan]

A few GPs who self-identified as having an interest in integrative medicine (nutritional, environmental and lifestyle medicine) had completed specific training in this area and reported feeling confident managing patient enquiries on *MTHFR* testing and discussing nutrition:

> *I'm familiar with that [MTHFR] and part of that is because of the integrative medicine training that I've had. So I would refer some patients to [have an MTHFR gene test] … I would normally go by a biochemical pathway, like a physiological and biochemical pathway of treating that. I am aware of the medical evidence for it as well so I try to put it all together.* [GP4, metropolitan]

Increasing the scope of practice to incorporate genomics into general practice

GPs' current roles with genetics/genomics were diverse and context dependent. Most felt their role was to assist or counsel patients to help them understand these types of tests and results, and to refer or seek advice from genetic specialists as required:

> *[My current role is] counselling and education, as far as I see it. [When] people want advice on detailed genetic testing, which is maybe warranted, then I'll generally … If it's an area I'm not familiar with, for example, SMA [spinal muscular atrophy] or MPS [mucopolysaccharidosis] or something like that, I'll get advice from the genetics team.* [GP24, metropolitan]

Generally, GPs predicted genomics would play a bigger part of their future practice, especially for risk prediction and to inform treatment and management of their patients; a small number were uncertain:

> *We will increasingly be using genomics in terms of screening, of risk stratification, of diagnosis, of dictating the best management of practices by way of medications or other therapies. I just think it's just going to influence everything we do, reasonably swiftly as it becomes more embedded into practice.* [GP18, metropolitan]

When questioned about the impact of reproductive genetic carrier screening on their future practice, some thought there would be increased requests for testing:

> *Definitely in the field of the preconception carrier screening I can see that taking off like quite quickly really to be almost much more universal than it currently is.* [GP3, metropolitan]

Relevance to practice underpins education topics preferred by GPs for genomic competency

When asked their opinions about what would assist them to feel confident in genomic medicine, many GPs mentioned a need for more education and training. GPs also identified additional supports beyond education. When asked what would help them with genomics in their practice now, GPs said they needed resources such as clear, up-to-date guidelines on genomic testing; decision supports; RACGP resources; patient handouts; and opportunities to discuss issues with a genetic specialist:

> *Lots more education, and time and practice. Discussions and perhaps better availability to specialists in the area.* [GP16, metropolitan]

> *If, at the time that someone came in asking for, say, BRCA gene testing, and then I could go and have a look at an online resource, say, what tests do I need to order, and then the interpretation of the results – that would be useful.* [GP25, metropolitan]

In general, GPs were interested in learning about genomics with relevance to their practice, such as conditions they would encounter or be more likely see in their day-to-day work:

> *Some clear recent guidelines that I can refer to, would be very good. Maybe [an] online database of tests … so that one could type in a test and to get interpretation.* [GP8, metropolitan]

Many GPs had difficulty identifying topics they would want included in future education activities and so were prompted by the interviewer with possible topics to consider. Some GPs were mainly interested in breast cancer genetics, followed by bowel, prostate and ovarian cancer genetic education. GPs who were aware of existing cancer guidelines were interested in keeping up to date as information emerged. Reproductive genetic carrier screening was also a topic of interest for half the GPs:

> *Preconception carrier [screening] would be probably one of the most important [topics]. I think that's the biggest one, by far.* [GP24, metropolitan]

When pharmacogenomics was suggested as a possible genomic education topic with GPs, most agreed it was relevant, although only a couple of GPs reported having seen pharmacogenetic/ pharmacogenomic test results in their practice.

> *I don't know a whole lot about pharmacogenetics … Even just the basic approach would be great.* [GP17, rural]

Other topics of interest included: prenatal testing, common diseases with a significant genetic component, availability and reliability of online DNA testing and explanation of results, and understanding

genomic tests and interpreting results to explain to patients.

When prompted with a list of educational approaches, GPs showed diverse preferences: case studies were most preferred, followed by face-to-face events (including seminars or workshops), online learning, journals, accredited CPD events, webinars and podcasts. Online learning was liked by many GPs for the convenience and capacity to be self-paced, and it was used by a few to gain background information on concepts or topics. Many GPs saw self-directed reading as an important way to acquire knowledge, such as reading hard copy or online articles in Australian GP medical journals. Shorter summaries of one to two pages were preferred by a few GPs:

> *If it's brief information, sure like a couple of pages I'm always happy to read things. But when it ends up being a 30-page document the chance of reading it's pretty low.* [GP13, remote]

Keeping up to date with genomics in general practice

While GPs acknowledged that education and supports would improve their genomic knowledge and skills, some also identified challenges in managing the volume and complexity of genomic information through their generalist practice:

> *The main challenge is keeping up to date with everything … [I'm] so busy already with keeping up with patient load it's hard to find the time to keep reading and learning constantly about everything that's changing and that is new … and patients come in knowing about something that you didn't know about.* [GP25, metropolitan]

Furthermore, approximately half the GPs commented that longer consultation times, including explaining limitations of the test and implications to the patient, could be challenging. The availability and cost of genomic tests to patients was also potentially problematic:

> *There's the medical and scientific side of things, but then there's the emotional and genetic and education side of things … And the other thing is, if it's genetic information then it's not just the patient's information, it has implications for the patient's family a lot of the time. So that will take up further time.* [GP16, metropolitan]

Discussion

This study provides current information about Australian GPs' views on genomics, practice and continuing education. Overall, GPs believed that genomic medicine will have an impact on healthcare and were optimistic about its potential benefits and it playing an increasing part in their future practice. However, many were unclear about the difference between genetic and genomic testing and reported lacking confidence in this area, with little formal genetics training received – similar to past studies.[2,24] Interestingly, many had already encountered genomic testing, not always realising the nature of these tests. The role of the GP was diverse and context driven, with longer consultations challenging. GPs felt education, training and resources would increase their confidence in managing genomics in their practice.

Understanding the relevance of genomic testing to practice was regarded as a barrier to integration by the GPs interviewed in this study, and it is also mentioned as a key challenge in other studies.[2–5,14,24,25] However, in the present study, it was noted that many GPs had already integrated some aspects of genomic testing into their practice through prenatal screening (eg NIPT), with support from genetic services. Some had managed requests for *MTHFR* gene tests, and a few had viewed patient-initiated online DNA test results. A few GPs confused preconception/reproductive carrier screening with prenatal screening, particularly NIPT/NIPS. Building a skilled workforce that is literate in genomics, which may address some of these issues, is a key strategy of the Australian National Health Genomics Policy Framework,[26] requiring investment in primary care. The Framework aims to harness the health benefits of genomic knowledge and technology into the Australian health system in an efficient, effective, ethical and equitable way to improve individual and population health.

The researchers tried to elicit what GPs specifically thought their education needs were, but most responses focused on the potential impact of the genomic era, reporting concerns about greater complexity and volume of genomic information; these concerns were also reported in a Canadian study.[27] Being generalists, trying to integrate complex genomic information into their practice likely contributes to GPs' ongoing concern about lack of knowledge. In a recent US study, GPs reported lacking confidence in having a central role in genetics because they needed to focus more on keeping up their skills to manage diverse conditions seen in primary care.[28]

GPs in the present study reported they preferred education topics in genomics relevant to their practice but were not always clear what these topics were. At present, there are no genomic competencies for GPs; therefore, a focus for educational activities should be on clinically relevant topics, such as those identified in this study.

GPs differed in their preferences for educational approaches; however, most liked case studies delivered through face-to-face events and online learning. In addition, GPs consistently requested support to help manage genomics in clinical practice, including online resources, clear up-to-date guidelines on genomic testing, links to genetic specialists where feasible and 'just in time' resources accessible at point of care, similar to preferences identified in previous studies.[11,29–31]

Paneque et al suggest that the ability to find relevant genomic information when needed might be a more appropriate goal in GP education.[30] Online genomic education resources developed specifically for GPs have been available overseas for some time;[11] Australian cancer genetics GP referral guidelines can be accessed on the NSW Cancer Institute's eviQ[32] and, in 2018, the RACGP produced the *Genomics in general practice* online resource in consultation with GPs, specifically to provide 'just in time' information.[20] This RACGP resource meets many of the needs mentioned by the GPs interviewed

in the present study, including direct-to-consumer/personal genomic testing, but very few were aware of its existence.

Since the completion of this study, further efforts to prepare GPs for managing genomics in general practice include a focus edition of *Australian Journal of General Practice* on genomics in March 2019, comprising key topics on preconception/antenatal carrier screening, cardiovascular genomics, pharmacogenomics and insurance issues related to genetic testing.[33–36] Some HealthPathways, which are web-based information portals, are developing genetics pathways information for GPs. Postgraduate study in genomics for health professionals has commenced at both the University of Melbourne and the University of Sydney, and GP training was conducted in a number of states, including workshops in Queensland. These programs provide great opportunities for GPs to upskill in genomics; however, study participants asked for 'just in time' genomics resources, which are currently not accessible within their medical software. Enabling GPs to see the relevance of education in genomics will be crucial to its impact.

Strengths and limitations

A strength of this study is that interviews were conducted with GPs across all states of Australia, in a range of work settings, with guidance from a taskforce that included practising GPs and representatives from relevant medical colleges. Although this approach provided rich themes to inform education strategies, gathering information on all relevant genomic education topics would require input from a larger number of GPs nationally.

Implications for general practice

The results highlight that GPs are concerned about their lack of knowledge and skills to manage the integration of genomics into their practice but that they are already ordering prenatal genomic tests. The rapidly changing nature of genomics and its consumer-driven testing, such as online testing, differentiates it from previously clinically indicated single-gene testing, which may not have been as relevant for many GPs. Driven by consumers, genomics is likely to be part of general practice even if GPs feel ill-equipped. The development of case-based professional education (online or face-to-face) will assist and will raise awareness of resources regularly updated by experts, increasing their use by GPs.

It would be beneficial for GPs to familiarise themselves with existing resources to support their practice, in particular the RACGP's *Genomics in general practice* resource[20] as first-line information and the eviQ cancer genetics GP referral guidelines[32] for cancer genetics assessment. Given that many GPs are not aware of the existence of these resources, raising awareness is a priority. Investment in their promotion by a range of organisations including professional GP organisations, Primary Health Networks and genomics education groups and the inclusion of relevant information in information portals and medical software will increase the reach of these programs and help meet GPs' current point-of-care needs in genomics.

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