Genomics and precision medicine: The expanding role of general practitioners

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Background
Precision medicine tailors an individual's healthcare to their genetics, environment and behaviour. This has been driven by rapid advances in genomics, where an individual's entire genetic code can now be sequenced. General practitioners (GPs) are increasingly involved in genomics throughout the lifespan, including performing prenatal screening during pregnancy, requesting reproductive genetic carrier screening and arranging genetic referrals for cancer and other conditions. Patients are relying on their GPs to appropriately counsel, test, refer and manage genetic conditions, and provide genomic healthcare.

Objective
This article provides an overview of the GP's expanding role in genomic medicine, some tools to assist GPs to navigate this landscape, and barriers to genomic uptake.

Discussion
GPs are integral to supporting patients and families in the genomics and precision medicine era. Resources specifically aimed at, and codesigned with, GPs are required to enable broader and equitable access for the benefit of all Australians.

NOW MORE THAN EVER, general practitioners (GPs) have a key role in genomic medicine. With their existing expertise in risk screening and family- and patient-centered care, GPs are already arranging genetic testing of chromosomes and single genes and arranging referrals for genomic testing (incorporating an individual's entire genetic makeup). Genomic medicine is increasingly relevant for GPs caring for their patients across their lifespans, covering pregnancy-related screening, cancer, cardiology, neurology and many other areas of medicine.[1–4]

Recent rapid advances have led to cheaper, more accurate and faster genomic testing and screening options and the emergence of advanced therapies for conditions such as cancer, neuromuscular disease and genetic blindness.[5] This has paved the way for precision medicine – defined as a tailored healthcare approach incorporating relevant genetic, environmental and behavioural information into a person's care. This offers a major new frontier in medicine and much promise for disease prevention and cure.[6] For example, genomic-based targeted treatments for cancer; tailored medication prescribing guided by pharmacogenomics;[7] polygenic scores supporting stratification of the population by disease risk for common conditions such as heart disease and cancer;[8] and population-based genetic screening,[9] reducing unnecessary interventions and improving healthcare at scale.[5,10]

Precision medicine is listed as a future-focused healthcare priority by the Commonwealth in 'Australia's Primary Health Care 10 Year Plan 2022–2032'.[11] The PRECISE (Practitioner Readiness, Education, Capabilities with Implementation Science Evaluation) genomics project is currently embarking on improving genomic resources for Australian GPs to support their provision of genetic healthcare.

This paper will provide an overview of:

- GPs' expanding role in clinical genomics
- tools and resources that can support GPs to navigate genomics
- barriers to GP uptake of genomics.

GPs' expanding role in clinical genomics across the lifespan

GPs are pivotal in recognising patients who may benefit from genetic or genomic testing because there is suspicion of a genetic condition based on clinical features or strong family history of a disease. When genetic factors are

or may be involved, GPs should consider an approach based on family history, identifying 'red flags' for genetics and testing, including referrals to optimise outcomes for patients (Table 1, Boxes 1–3).[12]

Timely action may allow earlier diagnosis of condition(s) in pregnancy and at all life stages. It might also enable appropriate referral to non-GP specialists for medical management and improve access to National Disability Insurance Scheme (NDIS) funding and to current or future therapeutic interventions or clinical trials. Additionally, it might identify people who are carriers for the same conditions as their reproductive partners (co-parents or donors), facilitating a discussion about reproductive options, and guide testing for other family members who might in turn benefit from clinical screening, diagnosis and medical management.

Boxes 1–3 cover a fictional case study commonly encountered in general practice and highlight the expanding role of a GP. When a child presents with neurodevelopmental differences, chromosome microarray (CMA) and fragile X syndrome (FXS) testing are indicated as first-tier genetic investigations with attached Medicare Benefits Schedule (MBS) item numbers[13] that GPs and paediatricians can request. A diagnosis made prior to an appointment with a paediatrician can improve care for a child and their extended family (Box 2).[14,15]

After a diagnosis, the GP also has an important role in identifying whether referrals for further psychological or social support may be needed, directing families to resources to provide these supports (Table 1) and checking that screening appointments are being attended.

In the context of the case scenario example, GPs also have an important role to play in family planning and pregnancy care. Reproductive genetic carrier screening (RGCS)[16,17] can be discussed with all people who are planning a pregnancy or who are in the early stages of pregnancy (Box 3). MBS item numbers exist for RGCS for cystic fibrosis (CF), spinal muscular atrophy (SMA) and FXS. Non-invasive prenatal testing (NIPT)[18] is a well-established screening test in pregnancy available from 10 weeks of pregnancy (although it should be noted that NIPT is not Medicare funded).

Further to this paediatric scenario, there is an increasing number of indications for genetic referral and testing. This includes common conditions such as hereditary haemochromatosis and thrombophilia.[19] Patients requesting direct-to-consumer genetic testing and pharmacogenomics are also increasingly presenting to primary care.[20–22] Genomic diagnostics is also expanding into many medical subspecialties such as cardiology (eg familial hypercholesterolemia, cardiomyopathy and arrhythmias), nephrology (cystic kidney diseases), ophthalmology (inherited retinal dystrophies), oncology (especially familial cancer) and neurology (neurogenetics, dementia, mitochondrial disease).[5] These are beyond the scope of this preliminary paper and will be further expanded in future PRECISE publications.

Box 1. Case study: Jason and Jane

First appointment

Jane, aged 31 years, has come to you because of ongoing concerns about her son Jason.

Jason has just turned 3 years of age. His pregnancy, neonatal period and early development were unremarkable. He passed his newborn hearing test, and his newborn screening test was normal. His growth parameters are within normal limits.

Jason's parents are concerned about his development. His parents have noted differences in Jason's fine and gross motor skill development as well as his speech development – he walked at 22 months, finds using utensils and playing with small toys challenging and has only 10 recognisable words. His educators have voiced concerns about his ability to follow their instructions and complete age-appropriate activities at daycare, and they have raised the possibility of developmental differences with the parents.

Jason's parents are concerned about his development and are seeking further investigation. They are considering having more children and wonder whether it is likely that future children may also exhibit developmental differences.

You arrange a follow-up appointment to further discuss Jason's neurodevelopmental differences and consider referral to a general paediatrician.

Box 2. Case study continued: Jason and Jane

First follow-up appointment

At Jason's appointment you take a family history and find there are no other red flags. You refer Jason to a paediatrician for a comprehensive neurodevelopmental assessment.

Since Jane and her partner are considering having more children and are concerned about Jason's development and having another child with similar developmental differences, you arrange first-tier genetic testing: testing for fragile X syndrome (FXS) and a chromosome microarray (CMA) as recommended in The Royal Australian College of General Practitioners' 'Genomics in general practice' handbook[13] and your local HealthPathways guideline. You arrange another follow-up appointment for returning results.

Second follow-up appointment

The CMA result is back in a fortnight and shows that Jason has 22q11.2 deletion syndrome. You discuss this result with the local clinical genetics service, who advises trying to fast-track the referral to the paediatrician as well as providing some preliminary information for the family on 22q11.2 deletion.

The clinical genetics service advises you to give this diagnosis to the family as soon as possible.

What are the benefits to Jason and his family of timely action from their GP?

Tools and resources to help Australian GPs to navigate genomics

Despite the increasing role of genomics in almost all areas of medicine, there are currently relatively few Australian resources and tools specifically targeted to help GPs navigate genomics (Table 2). The Royal Australian College of General Practitioners (RACGP) has published a comprehensive point-of-care guide outlining the current

Table 1. PRECISE approach for GPs to genetics and genomics

Take a family history	• Three-generation family tree (first- and second-degree relatives) • Medical conditions in the family and age of diagnosis • Cause and age of death • Ancestry • Consanguinity • Note sperm/egg/embryo/mitochondrial donation or adoption[43]
Identify red flags for a genetic condition/referral	A mnemonic 'GENES' has been created to aid recall of red flags: • G – group of congenital anomalies • E – extreme or exceptional presentation of common conditions • N – neurodevelopmental differences or concerns • E – extreme or exceptional pathology • S – surprising laboratory values[44,45] Additional features to consider include: • Known genetic condition in the family • Multiple family members with the same medical features • An earlier onset of a condition than expected • One or more congenital features in an individual and/or multiple family members • Differences in growth • Significant neurological symptoms • Non-familial facial features in an individual and/or multiple family members • Sudden unexplained death, unexplained cardiac arrest, unexplained syncopal episodes • Multiple miscarriages in an individual and/or multiple family member
Identify and request genetic tests within scope	• Reproductive genetic carrier screening • Non-invasive prenatal testing • Consider requesting a chromosome microarray for neurodevelopmental or congenital differences • Consider testing for fragile X syndrome (which tests the number of CGG repeats within the *FMR1* gene) in neurodevelopmental delay • Karyotype
Communicate results	• Return results to the patient • If a genetic test result is complex and requires non-GP specialist input and discussion, refer the patient to clinical genetics
Identify when a person should be referred to non-GP specialists	• Refer to clinical genetics when: – a genomic test is indicated and is out of scope – one or more red flags are identified and input from clinical genetics is indicated • Refer to non-GP specialists when: – specialist input is required (eg paediatrician, neurologist, oncologist)
Follow management guidelines for an individual with a genetic diagnosis	• Some rare genetic conditions have management guidelines that may include input from GPs • Some common genetic conditions may be fully managed by GPs
Coordinate care	• General practice is well placed to coordinate any non-GP specialist referrals, screening tests and allied health or NDIS support.
Identify potential psychosocial support and arrange a referral as indicated	• Genetic counsellor via local genetics service finder: www.genetics.edu.au/SitePages/Genetic-Services.aspx • Psychologist • Psychiatrist • Social worker • Condition-specific patient and family support organisations (eg RARE portal[46] and RArEST mental health support[47] for Australians living with a rare disease) • Support organisations for genetics such as Genetic Alliance Australia[48]

GP, general practitioner; NDIS, National Disability Insurance Scheme.

and most common clinical applications of genomic medicine in general practice.[13] It covers specific conditions, information about available genetic and genomic testing, and when to refer to the local clinical genetics service, and it includes links to additional reading. The Centre for Genetics Education[23] provides more general genomics information, online learning and an Australia-wide genetic service for GPs (www.genetics.edu.au/SitePages/Genetic-Services.aspx; Tables 1 and 2).[24]

In the field of cancer genetics, EviQ[25] is an oncology-focused point-of-care guide. It assists GPs to assess an individual's personal and family history of cancer and provides advice about referring to a familial cancer service for various cancer types. For example, referral to a familial cancer service or local genetic counselling service should be considered for individuals with a strong family history of young-onset cancers or a known cancer predisposition genetic variant in the family. This is a rapidly evolving space, with a growing number of genomic-related investigations in cancer care, including private GP-referred laboratory testing options.[26]

PRECISE: Codesigning tools and resources to navigate clinical genomics in primary care

The Australian Medical Research Future Fund–supported PRECISE genomics project aims to address the need for further Australian genomic education by codesigning resources with GPs, consumers, primary care peak bodies and organisations. This will address many of the barriers to improving genomic literacy for GPs and consumers while meeting the need for up-to-date, accurate point-of-care information. The PRECISE team, based at the University of Sydney, will codesign education resources for rapid upskilling with GPs, including those from rural and remote centres. PRECISE includes industry and policy partners RACGP, the Australian College of Rural and Remote Medicine, HealthPathways, the Centre for Genetics Education, NSW Health, Genetic Alliance Australia, Australian Genomics and two Primary Health Networks (Western NSW and Western Sydney). Through focus groups with GPs and consumers, the PRECISE team has already identified key areas to target with genomic education and resources – especially in pregnancy (NIPT and reproductive carrier screening), genomic testing, referral guidelines and cancer genetics – and are building an online portal that will be hosted at Medcast (https://medcast.com.au/genomics). The PRECISE education team has also adapted the UK clinical topic guide on genomics for GPs to form the basis for a suggested approach to genomics in Table 1.[12]

Together with PRECISE, the Nepean Blue Mountains (NBM) HealthPathways is developing pathway content about reproductive genetic carrier screening and genetic and genomic testing in general practice. NBM HealthPathways is an integration enabler formally supported by the partnership between Wentworth Healthcare, provider of the NBM Primary Health Network, and the NBM Local Health District. HealthPathways teams across Australia will be able to adapt the NBM guidance for their own context and for use by their local GPs (Table 2).

Barriers to GP uptake of genomics

Although it is helpful to have tools and handbooks, they represent a relatively small number of resources targeting GPs in this growing area. There is increasing evidence that co-creating education resources (such as the PRECISE approach) helps improve workforce capacity and implementation,[27] especially in new areas such as genomics.[28] In addition, a recent scoping review[29] has shown that numerous barriers exist for GPs in genomics, including the need for education; low confidence; and lack of time, resourcing and remuneration for genetics. An ongoing issue is that new MBS item numbers cover genomic testing but do not cover the complexity and time required for these discussions.[30,31] Direct-to-consumer testing and the commercialisation of diagnostic pathology genetic testing provide additional challenges for GPs to navigate.[30–34]

Patient barriers include a lack of awareness of personal genetic risk and a lack of knowledge of genetic services.[35] In rural and remote areas, the barrier of geographical distance exists, and in other priority populations there is inequity based on socioeconomic status and cultural background, combined with inadequate local workforce capacity for genomics.[33–35]

Box 3. Case study resolution: Jason and Jane

At the follow-up appointment to deliver Jason's result of 22q11.2 deletion syndrome, Jane discloses that she is 6 weeks pregnant. She is understandably overwhelmed by the information she is receiving about Jason and how it might be relevant to her current pregnancy.

You reassure Jane that this information will be useful in advance of Jason's appointment with the developmental paediatrician. You also refer Jane urgently to the local clinical genetics service and high-risk obstetrics service, where she will be seen within the next week to discuss the chance of recurrence of 22q11.2 deletion syndrome in this pregnancy and her reproductive options, including whether they wish to have a prenatal diagnosis or not, and ultrasound screening. You also discuss the option of reproductive genetic carrier screening (RGCS) for Jane and her partner.

In this case, the family's general practitioner (GP) had a pivotal role in managing the family's concerns, coordinating care and facilitating a pathway to rapid genetic diagnosis. Their use of The Royal Australian College of General Practitioners' handbook and local HealthPathways helped facilitate this. It is helpful to reflect on the direct benefits for Jason and his family:

- Jason received a timely diagnosis of 22q11.2 deletion syndrome in advance of his paediatrician appointment. This will fast-track surveillance for the complications of his condition, ongoing management, a formal neurodevelopmental assessment and, importantly, access to National Disability Insurance Scheme support for developmental therapies.
- Jane received urgent prenatal genetic counselling, the offer of RGCS and non-invasive prenatal testing (NIPT) and a discussion about prenatal diagnosis options.

Cascade testing in Jason's parents was subsequently arranged by their local clinical genetic service. This showed that the deletion is de novo in Jason, and there are no health implications for his relatives. The chance that Jane's current pregnancy will also have the deletion is low, and Jane has had a reassuring NIPT result and ultrasound scans.

Across his lifespan, Jason's GPs will provide ongoing surveillance, support, advocacy and access to information or referrals regarding recurrence and family planning options if he chooses to have children.

Patients also report concerns about perceived discrimination (eg life and health insurance),[36] financial barriers, disinterest in genetics and negative attitudes of non-genetics healthcare professionals as potential barriers.[1,33–35,37]

Finally, ethical and societal considerations exist regarding the acceptability of genetic testing. Navigating the breadth and depth of these dilemmas can be challenging for GPs, who are time poor and need to prioritise clinical outcomes.[38] The routinisation of prenatal genetic testing might imply that people with genetic conditions are less worthy than others,[39] that certain genetic conditions should be prevented from occurring, and that people should elect to undergo screening/testing in order to prevent having a child with a genetic condition.[40] These concerns are at odds with the concept of reproductive autonomy whereby individuals should be able to access prenatal genetic testing to make an informed choice[41] that is aligned with their values and preferences.[40] Other dilemmas include what testing should be publicly funded, which genetic conditions should be included on RGCS panels and how results should be reported.[42]

Conclusion

Despite a growing number of genetics/genomics resources available, there is a need for more targeted and codesigned GP education with accessible point-of-care resources. As this field evolves with the introduction of pharmacogenomics, polygenic risk scores and more genetic therapies, there will be an ever-increasing need for GPs to be involved in their patients' genetic diagnostic and management journeys.

Key points

- Genomics and precision medicine hold the promise of earlier diagnosis, streamlined management and targeted therapies.
- Genomics is increasingly a part of mainstream primary care practice, especially in pregnancy, RGCS, paediatrics and cancer.
- The expanding scope of GPs to provide genomic care includes testing, counselling and timely referrals to genetics services.

Table 2. Australian genetic and genomic resources for GPs

Topic	Resource	Details	Link
Genetics and genomics for GPs	The RACGP's 'Genomics in general practice', 2nd edition (updated 2024)	Covers a broad range of common topics such as genetic testing (includes direct to consumer and pharmacogenomics) and written for Australian GPs.	www.racgp.org.au/getattachment/63568f23-e288-4a0e-a23a-39fbd046cc21/Genomics-in-general-practice.aspx
Rare diseases	The RACGP's *check* CPD activity – 'Rare diseases' (November 2023)	Includes four case studies centred on considerations when treating patients with rare disease including heritable aspects.	www.racgp.org.au/check/check-issues/2023/rare-diseases
Cancer genetics	EviQ	Provides updated information on relevant Australian cancer referral guidelines for GPs in cancer genetics.	www.eviq.org.au/cancer-genetics/referral-guidelines/1147-general-practitioner-referral-guidelines-for
Genetic and genomic tests and education; genetic services finder	Centre for Genetics Education	Genetic and genomic testing (Australia) covers topics such as basic genetics, genomic consent, genetic tests, genomic consi derations and results of testing.	www.genetics.edu.au/SitePages/General-Practitioner-Resources.aspx www.genetics.edu.au/SitePages/Genetic-Services.aspx
Genomic resources, guides, education codesigned with GPs and consumers	PRECISE (Practitioner Readiness, Education, Capabilities with Implementation Science Evaluation) genomics project	Medical Research Future Fund–funded program to codesign resources to help GPs navigate genomics. Hosted on Medcast online platform, this project will produce point-of-care resources, short- and longer-format e-learning, and webinars on pregnancy care, genomic testing, genetic referral guides and cancer genetics resources for GPs. These have been codesigned with GPs and consumers, informed by their needs and preferences.	https://medcast.com.au/genomics
Referral pathways for GPs to genetics	HealthPathways	Only accessible via local Primary Health Networks – has genetic and genomic pathways in development.	
Genetics webinar and e-learning resource	CheckUP Genomics for GPs	A series of webinars developed in Queensland in 2020 covering prenatal/paediatric genetics, genetic conditions in adults and cancer genomics.	https://checkup.org.au/what-we-do/online-training/genomics-for-gps

CPD, continuing professional development; GP, general practitioner; RACGP, The Royal Australian College of General Practitioners.

- Currently, GPs lack adequate time, resourcing, support and clear referral guidelines in genomics.
- There is a need for genomic education resources created for GPs and codesigned with GPs such as provided at the PRECISE portal (www.medcast.com.au/genomics).

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