RACGP

Curriculum and syllabus for Australian general practice (7th edition)

Genomic medicine

RACGP Curriculum and syllabus for Australian general practice (7th edition): Genomic medicine.

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We acknowledge the Traditional Custodians of the lands and seas on which we work and live, and pay our respects to Elders, past, present and future.

Contents

Genomic medicine 4
- Context 4
  - References 4
- Learning outcomes 5
- Key areas of learning 6
- Opportunities for learning 7
  - Words of wisdom 7
  - Learning strategies 8
  - Case example 10
- Learning resources 10
- Assessment 11

Genomic medicine

Context

Genomic medicine includes both genetics and genomics. Genetic refers to the study of single genes. Genomics refers to a person's complete genetic makeup (or genome).[1] Genomic medicine is using the genetic or genomic information as part of the care of a patient.

Genetics has been part of a GPs practice for many years, with examples including newborn blood spot testing, cystic fibrosis gene testing, reproductive genetic carrier screening, and haemochromatosis gene testing. The research into genomics is always expanding our knowledge, and the indications for genomic testing and genetics referral in primary care are increasing. Direct-to-consumer genetic testing has also expanded the variety of questions a GP may be asked in a consult. Movement in some countries (including Australia) to offer genome sequencing for all newborns in the next decade may lead to changes practice.[2] Cancer is an area of expanding knowledge where both risk related to genes and ability to identify treatment options and responses has changed practice.

A diagnosis of a genetic condition impacts the patient, and potentially reproductive planning, but may also identify other relatives at risk of the condition who require assessment, information, and support to understand the implications. Complex legal and ethical issues can also arise. Knowledge of their patients' circumstances provides GPs with a sound foundation for this care.

Specialist general practitioners need to be able understand the potential and reality of genomic medicine as it evolves. GPs already integrate genomic testing into care and will need ongoing access to relevant resources as it further impacts practice.[3] However, as science continues to advance, the GP's core role will remain; communicating complex information to patients, in a way they can understand, and supporting their decision making.

References

1. The Royal Australian College of General Practitioners. Genomics in General Practice – A note on the title of the resource. 2nd edn. East Melbourne: RACGP; 2023. Available at: https://www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics-in-general-practice/genomics-in-general-practice/a-note-on-the-title-of-the-resource [Accessed 10 Oct 2025]
2. Davies M, NHS plans to DNA test all babies to assess disease risk. BBC news 21 June 2025. Available at: https://www.bbc.com/news/articles/c1ljg7v0vmpo. [Accessed 10 Oct 2025]
3. Tehrani MB, Hayward J, Gusen T et al. Genomics and precision medicine: The expanding role of general practitioners. *Aust J Gen Pract* 2026;55(1-2):72-78.

Learning outcomes

The GP is able to:

- understand and communicate the risk of a genetic condition to a patient or family, based on history and demographic factors
- clearly explain the results and potential implications of common tests and genetic screening to patients
- support patients considering genetic testing to make informed choices based on their preferences and values
- identify presentations, conditions, or family history where genetic testing should be considered
- demonstrate a safe and clinically appropriate response to unfamiliar genetic test results
- discuss with patients the potential relevance of their results and ethical considerations including family member information and reproductive decision making when appropriate
- understand the role of genetic testing in current and emerging treatments
- adapt and integrate genomic medicine into further areas of practice as evidence and clinical practice evolves
- be able to communicate with patients when relevance and implications of a genetic test result is uncertain.

Key areas of learning

Understand the principles of genetic inheritance:

- describe patterns of inheritance
- from a family history be able to identify potentially higher risk of a genetic condition
- clearly explain when a patient's concern about the risk of a specific condition is unlikely given the family history
- use evidence-based resources and calculators to inform clinical decision making.

Explain genetic concepts in a way the patient can understand.

Explain and support patients to understand:

- the relevance or risks that a result may mean for them
- when the meaning of a result is clear
- penetrance and what it may mean for them
- when results are not clear-cut, such as variants of uncertain significance
- their values and preferences when making their decisions.

Identify indication, discuss with patients, gain informed consent, and order results for genetic tests initiated in general practice, such as:

- reproductive carrier screening
- non-invasive prenatal screening
- chromosome microarray and karyotype
- fragile X testing
- cystic fibrosis
- familial hypercholesterolemia
- thalassaemia
- hereditary haemochromatosis.

Identify potential genetic 'red flags' and investigate appropriately, such as:

- recurrent miscarriages
- intellectual disability
- high probability of familial hypercholesterolaemia
- high risk family history for genetic contribution to cancer risk.

Appropriately refer to non-GP specialists and genetic counsellors.

Support patients with information and discussion to clarify their values and preferences when they are considering having genetic testing arranged by a non-GP specialist.

Support patients who have been notified of a potential family genetic issue to decide on their next steps, including ordering investigations or referral as appropriate.

Adapt and integrate new knowledge in genomic medicine into clinical practice.

Acknowledge the limits of the current understanding of some genetic testing results, with regards to clinical utility.

Understand medico-legal and insurance issues around genetic testing.

Opportunities for learning

Words of wisdom

- Think about how you can explain clearly and simplify genetic concepts to patients. Have a few versions of wording and potentially diagrams, handouts, and websites you can utilise to aid understanding.

- Be able to promptly access evidence-based resources to assist you in consultations, working with the patient to guide decision making and care.

- Genomic medicine will influence clinical practice, but every new piece of information in the media may not yet (or ever) lead to a clinically useful test or action. Have an approach to assess what you hear or are asked about, being open to possibilities but also considerate of current evidence.

- Regularly update family histories and, when there are new significant diagnoses in a family member, reassess the risk for the patient.

- Utilise wording and communication that makes it clear to patients that they have choices about whether they want to have genetic tests such as reproductive carrier screening or non-invasive prenatal screening. They should be routinely offered but, depending on the patient's preferences and values, they may make an informed choice not to have testing.

Learning strategies

On your own

Explore the breast cancer risk assessment tool iPrevent.

- *Which factors does iPrevent consider may have genetic basis, and which may not?*
- *When and how would you use this tool in practice?*
- *When might you discuss or refer a case to the local familial cancer clinic or genetics service?*

A patient has just been diagnosed with Huntington's chorea. They are estranged from their young adult children – they do not want them to know about the diagnosis.

- *Consider and explore your ethical and legal responsibilities.*
- *Would it differ if the diagnosis was breast cancer?*

With a supervisor

A 30-year-old woman comes requesting MTHFR gene testing. Their older sister has a MTHFR gene variant (p.Cys677Thr) identified when ordered direct-to-consumer gene testing. Their father has had a heart attack recently aged 72 years.

- *How would you approach this consultation?*
- *What resources would you utilise to assist?*
- *Would you order the test? Explain your thinking*

Discuss how you approach reproductive carrier screening with patients.

- *When do you raise it?*
- *How do you explain the options including no screening, triple screening and expanded carrier testing?*
- *What are the common questions patients have?*
- *How do you ensure results are followed up and documented?*
- *Are there differences or similarities in your approaches?*

In a small group

Rotate through your group role-playing consultations where you discuss the following results:

- *Reproductive carrier screening showing maternal fragile X result in the premutation length (55-199 CGG repeats).*
- *Reproductive carrier screening where mother carrier of cystic fibrosis gene and now father has the same result.*
- *A 33-year-old woman presenting as her mother has been identified to have the BRCA 1 gene.*
- *A 50-year-old man presenting with a result from direct-to-consumer genetic test showing he has one copy of the APO-Ɛ4 gene. His grandmother has just died from Alzheimer's disease.*

Discuss your approach to requests to order or interpret genetic tests.

- *What influences your decision making on ordering? How do you explain that?*
- *How to help patients decide which tests are appropriate, and within scope of primary care, or require referral to non-GP specialists?*

With a friend or family member

Explain how non-invasive prenatal testing works:

- *Why might it not report a result? What are your options from there?*
- *Why might it give a falsely reassuring low-risk result?*
- *What are the options following a higher risk result?*
- *What feedback do they have on the communication? Is their understanding correct post the explanation?*

Take a family history of cancer (which can be hypothetical) and then assess risks and discuss what if any next steps are recommended.

- *Did the person understand your explanation of their risk?*
- *Did they find anything confusing?*

Case example

Uma attends for a pre-pregnancy consultation and decides to have three-gene reproductive carrier screening that is eligible for an MBS rebate and is reported as a low-risk result. She then becomes pregnant and has non-invasive genetic testing. She then attends with her 18-month-old son Amir who has global developmental delay. She has been engaged with early intervention services who have said to her that you will, today, probably refer her son to a paediatrician and may also order genetic tests. She asks how he could have a problem that was only evident now given she had 'all the tests.'

In a case-based discussion, questions relating to each role could be:

Communicator

- How would you respond to her question?
- Consider how the earlier results were potentially communicated. How could effective communication have avoided her asking this question?
- Would your response differ if she had not had any carrier screening?
- What strategies could you use in this consultation to aid communication and check understanding?

Clinician

- What further information do you want from Uma?
- What genetic testing may the intervention team have been referring to? When would you consider ordering genetic testing for Amir? What tests could be ordered in primary care?
- What are elements of uncertainty and complexity in this scenario? How would you manage them?

Health advocate and leader

- What role should the GP play in the care of Amir? Who else may be in the team?
- How may the technology in the practice and eHealth systems have helped or made more difficult the care of Uma and then Amir?
- If, when reviewing the medical records, you were to find that Uma had never received her carrier screening results, what actions would you take? When would that be an adverse event in care?

Professional

- If the reproductive carrier screening result had showed Uma had a premutation for fragile X and they were not offered testing for the father, are there any legal issues that may arise?
- What issues may arise if Uma is no longer with Amir's father?

Scientist, educator and researcher

If you were developing an education session for practice nurses on reproductive carrier screening, what may be three learning objectives?

What are some potential areas where practice education may assist in similar scenarios?

Learning resources

The curriculum website includes learning resources within each unit.

Assessment

Content in this unit will be assessed in a variety of ways.

Workplace based assessments

Mini-CEX	RCA	MSF	CBD	Reflective exercise	Audit	Mid and end term assessment

Fellowship examinations

AKT	KFP	CCE

KEY

- Expected to be assessed here always
- Often assessed here but not in every example / case / assessment
- May at times be here or contribute to the assessment

ABBREVIATIONS

Mini-CEX – Mini-clinical evaluation exercise
RCA – Random case analysis
MSF – Multi source feedback
CBD – Case-based discussion
AKT – Applied knowledge test
KFP – Key feature problem
CCE – Clinical competency exam