Genomics Capability Framework for non-genetics health professionals

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June 2025

Australian Genomics

Background

Genomics contributes to early diagnosis, better targeted treatments and disease prevention (National Health Genomics Policy Framework, 2018–2021), with genomic testing increasingly integrated into clinical pathways (O'Shea et al., 2022). The Australian Genomics Workforce Education program (2020–2025) thus focused on widespread, quality education to develop a workforce prepared to use genomics in healthcare.

We have coordinated the development of a **genomics capability framework for non-genetics health professionals** to outline the knowledge, skills and attributes (attitudes and/or behaviours) that underpin safe and effective genomic medicine practice. The capability framework focuses primarily on heritable genetic conditions, rather than on cancer or precision oncology. However, the framework may be applicable to, or be adapted for, familial cancer settings.

We expect that the framework will primarily be used by educators and educational institutions to align programs and resources to support workforce development in genomics. However, it may also provide a foundation for stakeholders to recognise and develop the varied genomic capabilities required across the many professions and specialties involved in genomic healthcare delivery (Long et al., 2023).

Who is it for?	How can it be used?
Health professionals	As a self-assessment tool to inform professional development
Managers, employers and professional bodies	To identify workforce needs and development opportunities. To plan education and training
Educators and educational institutions	To guide the development of learning, education and training opportunities

Approach to development

During the development of this framework, stakeholders expressed a preference for considering capabilities rather than competencies. Capability as a concept has been used in many business and educational contexts (Fraser & Greenhalgh, 2001), and capability frameworks can provide a common language to describe the required knowledge, skills and attributes (attitudes and/or behaviours) that underpin safe and effective healthcare practices (Clinical Excellence Commission, 2021; NSW Public Service Commission, 2020). Where competencies are designed for practice in predictable situations and stable environments, capabilities recognise changing and complex contexts of practice (Fraser & Greenhalgh, 2001).

The methodology is shown in Appendix 1. The draft framework was developed through:

- A landscape review of genetic and genomic competencies, healthcare capabilities, competency and capability frameworks for non-genetic health professionals
- A landscape review of knowledge guides and genetic and genomic learning outcomes from workforce development programs for Australasian non-genetic health professionals
- Interviews with key stakeholders in genomic medicine, including genetic specialists, non-genetic clinicians, representatives of medical and allied-health Colleges, and patient representatives, which also identified aligned position statements and guidance to review (Appendix 2)
- Contributions from members of research groups and alliances with expertise in workforce education and training, including Australian Genomics, Melbourne Genomics, and the Murdoch Children's Research Institute Genomics in Society group;
- A modified Delphi process with invited experts in education, research, and healthcare to reach consensus on the final content, structure and wording of the framework.

Key decisions: Capabilities and professions

Acknowledging that models of genomic testing are currently shifting and may continue to change in future, the capabilities are not designed to relate directly to a specific profession or level of experience (Pichini & Bishop, 2002). Capability may vary within a profession depending on interests, experience, and role. Professions that can undertake counselling and consent for genomic testing will also vary depending on the condition/s being tested for, the test itself and related reimbursements (Burns et al., 2019; Hallquist et al., 2021).

It is inevitable that roles of different health professionals will continue to evolve; however, some capabilities may always align with specific professions, such as ordering testing or conducting specific medical procedures, assessments or examinations. Potential roles for some key professions are listed in Appendix 3.

We expect that variations between jurisdictions and services with respect to resourcing and models of care will determine which capabilities are adapted and/or adopted into local systems.

Acknowledgements

We acknowledge the contributions of clinicians, patient advocates, researchers, policymakers and educators to development of this capability framework. We also thank all those who were interviewed. We recognise the members of a Delphi expert group, including those members who wish to be acknowledged as follows:

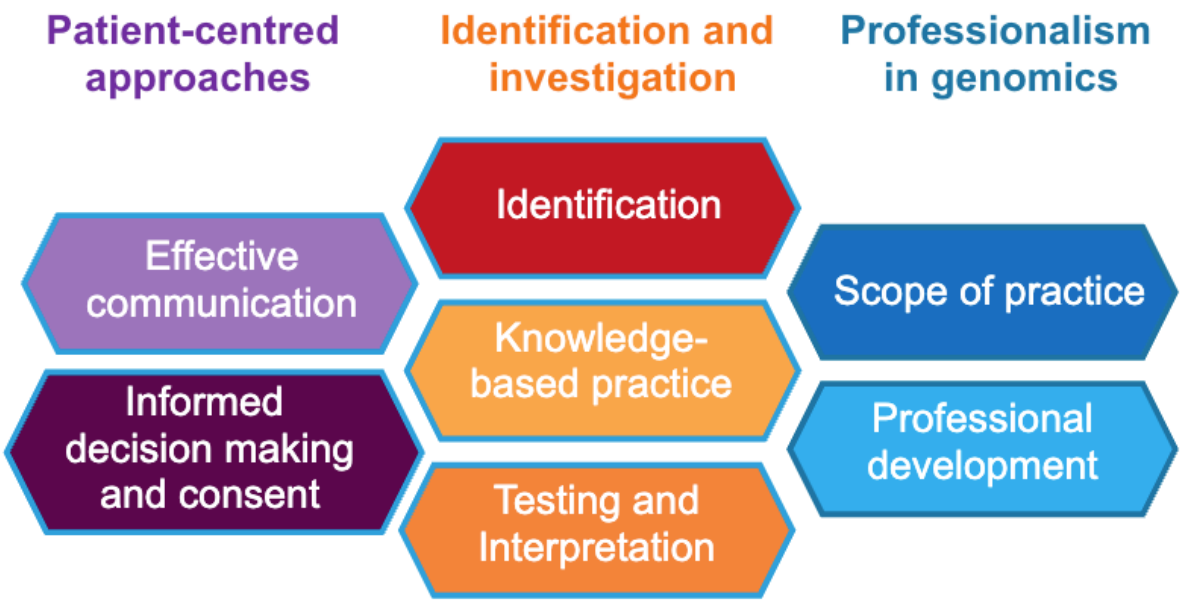
- Aideen McInerney-Leo, The Frazer Institute, Dermatology Research Centre, University of Queensland
- Andrew J. Mallett, KidGen Collaborative, Townsville Hospital and Health Service, James Cook University, The University of Queensland
- Cassandra McDonald, Centre for Genetics Education/Health Education and Training
- Chirag Patel, Genetic Health Queensland, Royal Brisbane & Women's Hospital, Brisbane
- Dhanushi Abeygunawardena, School of Biotechnology and Biomolecular Sciences, UNSW Sydney
- Elizabeth Emma Palmer, UNSW Sydney and Sydney Children's Hospitals Network
- Gabrielle Reid, Genomics Coordinator, Master of Genetic Counselling and Master of Genomics and Health, The University of Melbourne; Certified Genetic Counsellor, Inherited Cancers Australia.
- Helen Wright BSc MSc PhD, RN
- John Christodoulou, Murdoch Children's Research Institute and University of Melbourne
- Johanna Barclay, Australian Alliance for Indigenous Genomics (ALIGN)
- Kirsten Boggs, Australian Genomics
- Kortnye Smith, Medical Oncologist, Peter MacCallum Cancer Centre and Eastern Health
- Monica Ferrie, Genetic Support Network Victoria (GSNV)

**Please note that acknowledgement for contribution does not constitute endorsement by expert members' affiliations and/or organisations.

Citation

Cite this framework as: Terrill, B., Nisselle, A., & Gaff, C. for the Australian Genomics Workforce Education Working Group. (2025) *Genomics Capability Framework for non-genetics health professionals.* Australian Genomics, Melbourne. Available at www.australiangenomics.org.au

GENOMICS CAPABILITY FRAMEWORK



TOP LEVEL CAPABILITIES

Patient-centred approaches

Maintaining person-centred approaches in genomic healthcare that deliver high quality care to patients and their families

Effective communication

Health professionals can:

- Apply core clinical skills to communicating about genetic and genomic information
- Demonstrate the importance of sensitivity to personal, social and cultural perspectives when communicating about genetic/genomic information
- Tailor genetic/genomic information and services to a patient's or family's culture, knowledge, language ability and developmental stage

Informed decision making and consent

Health professionals can:

- Respect patient autonomy when making decisions about genetic/genomic information
- Consider potential practical, emotional and/or psychosocial benefits and risks of genetic/genomic information for patients and their families
- Clearly convey the purpose and process of genetic/genomic tests and/or procedures that may be offered
- Ensure that processes of obtaining, recording and communicating consent are appropriate for the test

Identification and investigation

Identifying patients who might benefit from genetics services and facilitating access to genetic/genomic information for patient care and management

Identification

Health professionals can:

- Collect and document health and family history relevant to a patient's clinical and psychosocial status
- Interpret health and family history to assess whether genetic/genomic testing may benefit a patient, within scope of practice

Knowledge-based practice

Health professionals can:

- Demonstrate knowledge of core genetic/genomic concepts, including inheritance and mechanisms of disease
- Recognise the role and application of genetic/genomic information in the prognosis, diagnosis, treatment and prevention of disease
- Apply knowledge of recommended care pathways that incorporate genetics services and genetic/genomic information
- Describe ethical, legal and social issues related to testing, recording, sharing and storing genetic/genomic information

Testing and interpretation

Health professionals can:

- Apply knowledge of the utility and limitations of genetic/genomic testing and information to underpin care, support for patients and their families

If within their scope of practice and experience, health professionals can also:

- Use genetic/genomic testing to guide patient management
- Communicate the results of genetic/genomic testing, their implications and limitations

Professionalism in genomics

Acting based on awareness and understanding of their role in recommended patient care pathways for genetics services and information

Scope of practice

Health professionals can

- Support, provide, or seek assistance to facilitate, genetic/genomic healthcare within scope of practice
- Contribute to medical management before and after genetic/genomic testing
- Recognise the role of research in clinical genomics

Professional development

Health professionals can:

- Maintain genetic/genomic knowledge and skills relevant to their specialty, context or role

DETAILED CAPABILITIES

Patient-centred approaches

Maintaining person-centred approaches in genomic healthcare that deliver high quality care to patients and their families

Effective communication

Health professionals can:

Apply core clinical skills to communication about genetic and genomic information

- Communicate with patients and their family or carers clearly, respectfully, empathetically and collaboratively
- Assess patient capacity, understanding and expectations of genetic information and tests offered
- Check effectiveness of communication with patients, families and professionals involved in patient care
- Consider factors that may influence a patient's or family's response to genetic/genomic information, including the timing of the communication and life events
- Manage periods of uncertainty (e.g., while waiting weeks or months for a test, or if an uncertain variant is returned) through effective communication and evidence-based support

Demonstrate the importance of sensitivity to personal, social and cultural perspectives when communicating about genetic/genomic information

- Appreciate the diverse ethical, social, cultural and religious issues that may arise in genetics/genomics communication with patients and their families from diverse backgrounds e.g., family dynamics and relationships, reproductive choices or stigma
- Acknowledge that patients and their families vary in their emotional response to, and capacity to process, complex information about genetics/genomics, which may require additional time and flexibility
- Appreciate the potential for discrimination and/or misuse of genetic information, including family history
- Develop, implement, evaluate and reflect on strategies to promote a culturally safe practice environment

Tailor genetic/genomic information and services to a patient's or family's culture, knowledge, language ability and developmental stage

- Consider that a person's social and cultural perspectives, expectations, capacity and life stage may affect their access to, and uptake of, genetic/genomic information and services
- Make reasonable adjustments to promote equitable access to genetic/genomic healthcare, acknowledging barriers for individuals, families and communities
- Elicit preferences for timing and method of communication to integrate into patient care
- Use a range of methods and resources to facilitate appropriate and culturally safe communication about genomic information and testing
- Respond to the changing information and support needs of patients and their families throughout genetic/genomic healthcare

Informed decision making and consent

Health professionals can:

Respect patient autonomy when making decisions about genetic/genomic information

- Advocate for patients to make informed decisions and act voluntarily
- Demonstrate respect for patients' personal and cultural values, practices and health beliefs in decision making
- Ensure shared decision making between a patient and practitioner throughout the medical planning process
- Acknowledge that patient choices and actions may differ with each situation, influenced by medical history, current care, personal values and life events
- Respect and support patients' right to decline genetic/genomic testing and explain implications and limitations of decisions and/or alternatives for patient care

Consider potential practical, emotional and/or psychosocial benefits and risks of genetic/genomic information for patients and their families

- Describe the possible familial implications of genetic/genomic test information and the importance of family communication
- Outline potential financial, clinical, and psychological implications for patients and their families
- Deliver genetic/genomic information, recommendations and care without bias or judgement, within an appropriate process of facilitating informed consent
- Recognise the potentially profound impact of genetic/genomic testing and test results for patients and their families
- Acknowledge potential power relations by creating respectful and collaborative environments that empower patients to make informed decisions

Clearly convey the purpose and process of genetic/genomic tests and/or procedures that may be offered

- Describe the rationale for genetic/genomic testing and the benefits, risks and limitations, and any alternatives
- Explain key concepts in genetics (such as variation, inheritance and probability) and the influence of genetic factors in health and disease
- Describe the different types of results or outcomes of a genetic/genomic test (identifying a variant, not identifying a variant, or an unclear or uncertain result), including the potential for unexpected or incidental findings
- Explain clinical actions that may or may not be recommended based on test results, and the possible implications for patients and their families
- Outline how biological samples and data may be stored and accessed and a patient's rights to provide or withdraw consent for future use, in their own care or in research
- Summarise uncertainties related to genetic/genomic information, including the potential for inconclusive results
- Clarify that reanalysis or retesting may be possible in future, when new evidence is available

Ensure that processes of obtaining, recording and communicating consent are appropriate for the test

- Act according to the national and local regulations, processes and governance for obtaining consent for genetic/genomic testing, and changes to consent
- Recognise that decision making and testing processes during pregnancy or acute care settings are time-critical
- Ensure that decisions and options are effectively communicated to all involved in patient care
- Recognise privacy requirements in storing and communicating family and genetic/genomic information

Identification and investigation

Identifying patients who might benefit from genetics services and facilitating access to genetic/genomic information for patient care and management

Identification

Health professionals can:

Collect and document health and family history relevant to a patient's clinical and psychosocial status

- Recognise the importance of documenting clinical and family history in assessing risk of having or developing genetic conditions
- Elicit and acknowledge patient experiences of a condition and perceptions of health and disease, recognising the role of taking a family history in building a relationship and rapport
- Respect privacy of a patient and their family members in assembling and documenting family history, and navigate any potentially competing interests
- Recognise that social and cultural contexts can shape how patients understand, share and respond to questions about family and health
- Explain social and legal risks and protections related to family history, including family implications e.g., identifying increased disease risks for family members and/or unexpected family relationships
- Document a clear and comprehensive multigenerational family health history relevant to a patient's clinical status

Interpret health and family history to assess whether genetic/genomic testing may benefit a patient, within scope of practice

- Interpret a pedigree and clinical investigations, as appropriate, to identify patient and/or family presentations that suggest a genetic condition or confirm a suspected inheritance pattern
- Recognise key indicators, variability of presentation and implications of genetic conditions within their specialty or context
- Recognise indicators of targetable genetic variants based on their frequency/importance in specific tumour types or genetic conditions, as appropriate
- Determine whether, or when, clinical genetics evaluation or genetic counselling may be appropriate

Knowledge-based practice

Health professionals can:

Demonstrate knowledge of core genetic/genomic concepts, including inheritance and mechanisms of disease

- Recall the structure and function of the human genome, including the function of human genes and proteins within cells
- Explain how genetic variation in coding and non-coding regions of the genome can affect gene function, regulation and expression
- Describe how genetic variants can influence disease presentation, onset and severity within their specialty or context
- Recognise that genetic and environmental factors play important roles in common diseases
- Differentiate between inheritance patterns, multifactorial inheritance and polygenic risk
- Describe the indications for testing and referral of genetic conditions encountered in specialty or context
- Describe different implications of somatic and germline analysis of genetic/genomic information
- Recognise targetable genetic variants and their frequency/importance in specific tumour types and/or conditions

Recognise the role and application of genetic/genomic information in the prognosis, diagnosis, treatment and prevention of disease

- Describe key clinical settings of genetic/genomic testing, e.g., screening, preimplantation genetic diagnosis, prenatal, diagnostic, predictive/presymptomatic or personalised medicine
- Recognise the indications for, and utility and limitations of, different types of genetic/genomic tests, relevant to specialty or context
- Recognise that genetic/genomic test results require interpretation in context of a patient's clinical status
- Describe relevance of genetic variants in drug metabolism and personalised/precision medicine
- Demonstrate awareness of the limitations of genetic/genomic information and healthcare for patients of non-European ancestry, including in Aboriginal and Torres Strait Islander peoples and Māori, e.g., lack of diversity in genomic datasets and ongoing impacts of colonisation
- Recognise utility and limitations of emerging genomic therapeutic strategies and pathways

Apply knowledge of recommended care pathways that incorporate genetics services and genetic/genomic information

- Describe the role of genetics services and laboratories in providing specialist care for patients
- Explain the requirements for, and processes of obtaining, informed, written consent for genetic/genomic testing
- Describe the role of multidisciplinary teams (MDTs) and/or consultation with experts across fields and organ systems to deliver genetic diagnoses, counselling and patient care
- Describe professional and legal obligations in safeguarding individual and family genetic/genomic data and/or communicating and disclosing genetic/genomic information for ongoing care of patients or others
- Obtain credible, current information about genetics/genomics for self, patients, families and colleagues

Describe ethical, legal and social issues related to testing, recording, sharing and storing genetic/genomic information

- Recognise the potential for family history or genetic/genomic information to reveal unexpected family relationships
- Consider the personal and/or social impact genetic/genomic information may have for a patient and their family, e.g., effect on familial relationships, ability to obtain life insurance or support
- Demonstrate cultural understanding and respect for cultural practices, perspectives and health beliefs that may influence engagement with, and response to, genetic/genomic information or healthcare
- Acknowledge contemporary ethical issues in genomics, such as testing minors for adult-onset conditions
- Reflect on any perspectives on genetic/genomic issues that may influence their personal views of patients and/or families, and their decisions

Testing and interpretation

Health professionals can:

Apply knowledge of the utility and limitations of genetic/genomic testing and information to underpin care and support for patients and their families

- Take appropriate and timely action to seek assistance from, and refer patients to, genetics services, other specialties or peer support resources, as appropriate
- Facilitate access to genetic/genomic information and/or testing as appropriate, within scope of practice, and support a patient in ongoing care
- Be aware of patient support options beyond the healthcare system to inform patients and their families about options for practical guidance, support and/or shared experience
- Act according to established protocols for facilitating informed consent and exchange of genetic/genomic information

If within their scope of practice and experience, health professionals can also:

Use genetic/genomic testing to guide patient management

- Order and interpret genetic/genomic tests and apply genetic/genomic information to guide treatment or management options, within scope of practice
- Take appropriate and timely action, adhering to local guidelines for genetic/genomic testing, including funding, sample requirements, and local test pathways including multidisciplinary teams (MDTs)
- Recognise the potential utility of testing multiple family members to clarify the genetic basis of a health condition
- Maintain dialogue and coordinate with multidisciplinary teams (MDTs) and/or genetic testing laboratories, to ensure that appropriate tests are ordered and conducted
- Interpret genetic/genomic test results in the context of a patient, their medical and family history
- Recognise the utility of reanalysis and review of genomic results based on new information
- Use reputable resources and accepted guidelines to develop a management plan with a patient that acknowledges their genetic risk factors or conditions
- Recognise the influence of genetic/genomic results on the quality and safe use of medicines

Communicate the results of genetic/genomic testing, their implications and limitations

- Describe findings and implications of genetic/genomic test results, including additional or alternative explanations
- Consider a patient's and their family's personal and cultural perspectives when delivering genetic/genomic test results
- Clearly describe current or future clinical management options, including benefits and limitations, to support patient autonomy in decision making
- Provide patient information beyond a diagnosis or test result, such as future family planning and/or screening
- Describe the wider familial impact of a genetic/genomic test result, including clinical management and testing available
- Draw on a range of counselling approaches to support patients to optimise health behaviours

Professionalism in genomics

Acting based on awareness and understanding of their role in recommended patient care pathways for genetics services and information

Scope of practice

Health professionals can:

Support, provide, or seek assistance to facilitate, genetic/genomic healthcare within scope of practice

- Define scope of practice within relevant legislation, regulations, policies and guidelines, based on an understanding of professional roles and according to responsibility, knowledge and experience
- Seek assistance, consult with genetic specialists, and refer appropriately, when a procedure or genetic/genomic test is outside role, scope or level of competence
- Collaborate effectively with other healthcare providers to promote quality and equity in patient care
- Communicate effectively with healthcare professionals, recognising the need for confidential management of personal and familial genetic information

Contribute to medical management before and after genetic/genomic testing

- Recognise ongoing responsibilities to provide patient care, information or support beyond testing, and act as appropriate
- Recognise the importance of continuity of genetic/genomic healthcare through timely referral/provision and follow up
- Proactively coordinate care within a multidisciplinary team (MDT), as appropriate
- Identify and connect patients, with and without diagnoses, with relevant healthcare professionals, mental health and wellbeing services, and/or patient support organisations

Recognise the role of research in clinical genomics

- Contribute to quality improvement and relevant practice-based research as appropriate
- Recognise that genomic research can develop new approaches to prevent, diagnose and treat disease
- Facilitate access, as appropriate, to clinical studies and trials based on genetic/genomic test results
- Acknowledge the voluntary nature of clinical studies and trials
- Recognise the differences between clinical care and research in genetics/genomics, ensuring that any consent processes are considered separately
- Explain risks and benefits of participating in research and sharing data

Professional development

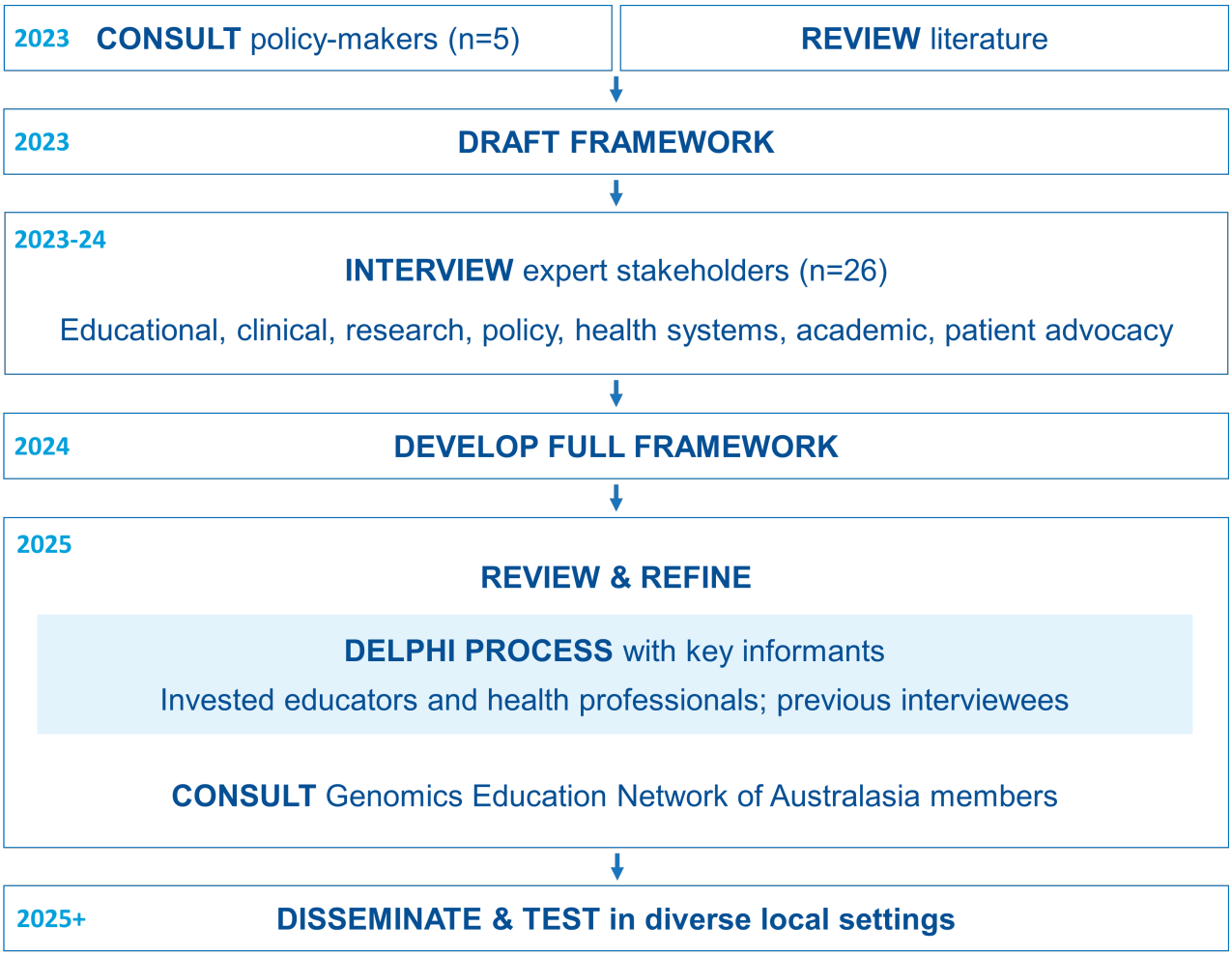
All health professionals can:

Maintain genetic/genomic knowledge and skills relevant to their specialty, context or role

- Reflect on their own practice, appraising strengths and limitations of their genetics/genomics expertise and capability, based on an understanding of professional role
- Identify areas of knowledge or capability where professional development may be beneficial
- Participate in learning, research and/or quality improvement opportunities to fill knowledge gaps and develop and maintain capability in genetics/genomics
- Recognise that genomics is a rapidly evolving field and knowledge of genomic advances, technologies and/or methods relevant to specialty, context or role should be updated regularly

Appendix 1: Methodology

The framework was developed across five stages, including multiple consultations with key stakeholders and informants.



Appendix 2: Position statements, guidelines and resources suggested by stakeholders and/or key informants

Informants advised aligning the capability framework with the following position statements, curricula and guidelines. These were all reviewed and informed the framework.

- Australian College of Nursing (2020). *Nurses, Genomics and Clinical Practice.* www.acn.edu.au/wp-content/uploads/position-statement-nurse-genomics-clinical-practice.pdf
- Australian Commission on Safety and Quality in Health Care (2022). *Requirements for Medical Testing for Human Genetic Variation* (3rd ed). Sydney: ACSQHC. www.safetyandquality.gov.au/sites/default/files/2023-03/requirements_for_medical_testing_for_human_genetic_variation_third_edition_2022.pdf
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- Royal Australian College of General Practitioners (2022). *Curriculum and Syllabus for Australian General Practice.* www.racgp.org.au/education/education-providers/curriculum/curriculum-and-syllabus/home
- Rare Disease Awareness, Education, Support and Training (2024). *National Recommendations for Rare Disease Health Care.* https://rarevoices.org.au/national-recommendations/

Informants also advised the following documents would be useful, or referred to these in their responses.

- Australian Genomics (2024). *Workforce Education Tools and Resources* www.australiangenomics.org.au/categories/workforce-education/ and www.genomicsinfo.org.au/
- Genetic Health Queensland (2024). *Resources for Health Professionals: Genetic Testing & Genomics.* https://metronorth.health.qld.gov.au/rbwh/genetic-health-queensland/information-for-practitioners/resources
- NSW Health Centre for Genetics Education (2022). *Education for Paediatricians: Genomic Testing.* www.genetics.edu.au/SitePages/Paediatrics-Educational-Videos.aspx
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- GeneEQUAL (2025). *Toolkit for Health Professionals.* www.genetics.edu.au/SitePages/Intellectual-disability-patient-communication.aspx

Note, these lists are not intended to be comprehensive.

Appendix 3: Potential roles for key professions

Capabilities in this framework do not relate to a specific profession or level of experience; however, some informants suggested including a table of items that may be performed by specific professions. For example,

General practitioners	Take family and medical history Refer to specialist or genetics services, and follow up Facilitate consent for, and order, defined genetic/genomic tests or screening Return genetic/genomic test results with limited scope Support and/or provide pre-test and ongoing post-test care
Midwives, nurses and nurse specialists Nurse practitioners Pharmacists / allied health	Take family and medical history Advocate for informed decision making and continuity of care Return genetic/genomic test results with limited scope Support and/or provide pre-test and ongoing post-test care
Physicians	Take family and medical history Refer to other specialist or genetics services, and follow up Facilitate consent for, and order, defined genetic/genomic tests or screening relevant to specialty Return genetic/genomic test results with limited scope Support and/or provide pre-test and ongoing post-test care and treatment
Genetic counsellors	Take family and medical history Facilitate consent for broad, complex and/or uncertain genetic/genomic tests Support and/or provide pre-test and ongoing post-test care Return results of genetic/genomic tests, including broad, complex and/or uncertain results
Clinical geneticists	Take family and medical history Facilitate consent for, and order, broad, complex and/or uncertain genetic/genomic tests Support and/or provide pre-test and ongoing post-test care Return results of genetic/genomic tests, including broad, complex and/or uncertain results

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