

Position Statement

Subject: **Role of the genetic pathologist**
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The Royal College of Pathologists of Australasia has developed this position statement to provide information to health care providers, pathology trainees, regulatory and administrative bodies, governments, and the broader community on the skills and role of genetic pathologists. In particular, this document outlines how genetic pathologists contribute to the delivery of effective and efficient healthcare that is of high quality.

Pathology is the branch of medicine that focuses on identifying the cause and processes of disease by analysing a sample from a patient. There are numerous professional groups involved in the delivery of pathology. Pathologists are medical specialists who, together with medical scientists and technicians, deliver pathology services. This document details the role of genetic pathologists in particular, as it is a relatively new discipline of pathology that is not widely understood.

What is genetic pathology?

Genetic pathology does not represent an exclusive domain that excludes other stakeholders; genetic pathology represents growing recognition of the complexity and sophistication available in genetic testing. An analogy could be that it is no longer accepted medical practice for a general surgeon to perform routine neurosurgery. Nonetheless, a general surgeon will utilise knowledge and expertise that has some application in neurosurgery. Similarly, genetics is a component of every aspect of pathology, but there is an important place for a specialist in genetic testing and their role in patient care.

Pathology is divided into disciplines relating to particular organ systems, biological processes, methods of analysis, or the clinical context under consideration. For example, haematology is a subspecialty of pathology focusing on the underlying science and investigation of blood disorders. There is overlap between all disciplines of pathology, yet there is sufficient distinction and complexity to justify these discrete disciplines.

Genetic pathology could be defined as the subspecialty focusing on analysis of human genes for mutations which cause disease. However, such a definition fails to encompass biochemical genetics, the analysis of proteins (encoded by genes) and metabolites (produced by them); biochemical genetics has been an integral component of genetic practice for decades. Furthermore, our understanding of the genetic and biochemical basis of disease is increasingly recognised as a component of every discipline of pathology, and hence it is challenging to limit the scope of genetic pathology. Similarly, the application of genetic knowledge in clinical practice is increasingly required of general practitioners, medical specialists (of all types), and other health care professionals, in addition to the expertise demonstrated by specialist clinical geneticists.

There is no accepted international definition of the term “genetic testing”. As noted elsewhere, “there is no clear boundary between genetic and non-genetic conditions and tests”. For the purpose of this document, we define genetic pathology as being the analysis of human genetic material, or of substances derived from it, for identifying the cause and process of disease.

Our understanding of genes, variations in genes, and their impact in human health and disease is relatively recent. The medical testing of genes represents a tiny proportion of the 50 million pathology tests that are provided annually in Australia. Nonetheless, the volume of genetic testing is growing rapidly (23% per year), and the diversity of genetic testing is also increasing rapidly (5% increase per year in the types of genetic test available).

How is a genetic pathologist trained?

The training of a genetic pathologist starts with training as a medical practitioner, including two years of broad clinical practice as a junior medical officer, ensuring a broad understanding of disease processes, their impact on individuals, and their management within a healthcare system. A genetic pathologist then completes a training program of at least five years to develop specific expertise in the scientific, technical, and clinical aspects of the genetic basis of disease, and of the management, safety, and quality aspects of delivering genetic testing for healthcare.

As a result of training in medical care and the science of genetic testing, a genetic pathologist has the broad foundation of experience to provide clinical guidance, medical accountability, and administrative responsibilities necessary to running a medically effective laboratory operation.

Other health care providers, including pathologists in other disciplines and medical scientists, often have expertise in genetic knowledge (as it relates to their particular practice) or genetic testing (for a specific disorder in their discipline). This is both appropriate and necessary for the delivery of quality healthcare. Genetic pathologists have both the breadth and depth of training in medical care and the science of genetic testing to provide a high level of expertise across the scope of this discipline.

What are the responsibilities of a genetic pathologist?

Genetic pathology is a broad service which provides information underpinning much of healthcare delivery. An understanding of the genetic basis of disease is now a component of every discipline of health care. The reach of genetic pathology is growing such that errors and inefficiencies at any point in the testing process can have magnified consequences across many health disciplines.

To reduce the risk of such adverse outcomes, the genetic pathologist has three overlapping areas of responsibility:

The genetic pathologist as a medical consultant

- A genetic pathologist works at the interface of genetic science and medical practice, and this knowledge of both domains means they can apply this knowledge in clinical reasoning for patient benefit.

- A genetic pathologist is a communicator who understands the needs of the requester and the patient, and is able to provide clear interpretations of genetic test results to inform and guide decision-making by the requester and patient.
- The consultative role of a genetic pathologist is particularly important with the implementation of genomic testing, involving analysis of 100s to 1000s of genes simultaneously. Such testing should only be undertaken in the context of an explicit medical consultation involving the requesting clinician, the patient, and the genetic pathologist at a testing laboratory. This consultation needs to include issues of consent and the reporting of incidental findings.
- The consultation may include technical aspects of test performance, recognising that a test may have high validity in one clinical context, but diminished validity in another. With the rapid developments in genomic testing now being implemented, this ability to relate contemporary genetic testing to specific clinical settings is an essential role as many clinicians will not be familiar with the strengths and weaknesses of the newer investigations.
- A genetic pathologist is a registered medical specialist and thereby is accountable both professionally and legally for expert judgements made regarding the medical significance of clinical laboratory data. This accountability is reflected in the requirement for documented continuing professional development, membership of a national medical specialty body (the College) and annual medical registration.
- A genetic pathologist advises requesting clinicians of the comparative attributes and performance of genetic tests in a specific clinical setting. This may involve determining whether the requested test is fit for purpose for a particular patient and the limits that may be placed on the interpretation of the test result for a particular patient and setting.
- A genetic pathologist uses an understanding of genetic science, technical aspects of genetic testing, and the clinical implications of genetic knowledge to guide the development and maintenance of accurate genetic tests. This requires consultation with other laboratory personnel, pathologists, clinicians, and stakeholders to ensure that an efficient test is provided in an appropriate fashion.

The genetic pathologist as an educator and researcher

- The genetic pathologist must ensure that there are sufficient personnel with adequate documented training and experience to meet the current needs of the laboratory. This is particularly pertinent at a time of rapid change in our understanding and the technical capabilities in delivering genetic testing. The genetic pathologist communicates with other staff in the genetic laboratory and with other laboratory professionals to facilitate the development and maintenance of accurate useful pathology tests. This is particularly important with the increasing genetic analysis in all disciplines of pathology.
- The genetic pathologist contributes to formal and informal activities such as multidisciplinary meetings, peer review sessions, and educational activities to enhance the genetic understanding of colleagues.
- The genetic pathologist is responsible for establishing and maintaining communication with the broader healthcare community of funders, providers, and regulators to guide policy and resourcing of genetic testing.
- The genetic pathologist is responsible for developing communication with the broader community to foster understanding of the strengths and limitations of this type of testing. This is of particular importance with the rise of “direct to consumer” genetic testing that is provided without the involvement of a healthcare practitioner.
- The genetic pathologist is responsible for training the next generation of medical and science students, trainee pathologists and scientists and other medical specialists and health professionals in the rapidly advancing field of genetics and genomics.

- A genetic pathologist is also involved in conducting and managing genetic research. It is essential that the rapidly expanding knowledge in genetics and genomics is translated into meaningful outcomes for patients. The College recognises the importance of such research in furthering the utility and availability of genetic testing in healthcare and the central role of genetics pathologists in this research.

The genetic pathologist as an administrator

As head of a genetic laboratory, a genetic pathologist is legally, morally, and ethically responsible for the overall operation and administration of the laboratory, particularly in relation to safety and the quality of the genetic testing provided.