Timely and accurate pathology results are critical to the functioning of our entire medical system.

Pathology informs the clinical decisions of medical practitioners across the healthcare spectrum.

Given its critical role, the risks of not adequately supporting a strong national pathology system are:

- Incorrect diagnoses;
- Delayed diagnoses;
- Incorrect assessment of risk of getting a disease;
- Incorrect treatment;
- Possible early patient death; and
- Erroneous cessation of a pregnancy.

These issues may impact upon the physical, emotional and financial well-being of individual patients, their families and the community at large.

As the peak body representing the profession, the RCPA believes the underlying principles of a world class pathology service are:

- A commitment to patient safety and quality
- A highly trained and sufficiently resourced workforce
- Efficient services that ensure timely and accurate results
- Equity of access and choice of provider
- Timely adoption of appropriate new tests reflecting international best practice
- A commitment to ongoing education, research and teaching

"Genetics impacts on all areas of medicine such as paediatrics, oncology and obstetrics as well as pathology in general with microbiology, haematology and immunopathology."

Dr Melody Caramins

Pathology disciplines

70% of all diagnoses are made using a pathology test. All chronic conditions require monitoring via pathology testing. Pathologists work across a range of different specialities in addition to genetics. These include:

Anatomical pathology, which looks at tissue analysis of disease;

Chemical pathology, which deals with the entire range of disease, and encompasses detecting changes in a number of substances in blood and body fluids (such as electrolytes, enzymes and proteins);

Forensic pathology, which seeks to investigate and define the cause of unexpected death;

Haematology, which deals with diseases that affect the blood such as anaemia, leukaemia, lymphoma, clotting or bleeding disorders as well as management of blood transfusions;

Immunopathology, which deals with the diagnosis and management of conditions in which the immune system does not function properly;

Microbiology, which deals with diseases caused by infectious agents such as bacteria, viruses, fungi and parasites; and

General pathology, which covers the profession as a whole.

For brochures and videos about each discipline, go to the RCPA website at **www.rcpa.edu.au.**

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The Royal College of Pathologists of Australasia
Durham Hall, 207 Albion Street, Surry Hills NSW 2010
Tel 61 2 8356 5858 **Fax** 61 2 8356 5828
Email rcpa@rcpa.edu.au **Web** www.rcpa.edu.au

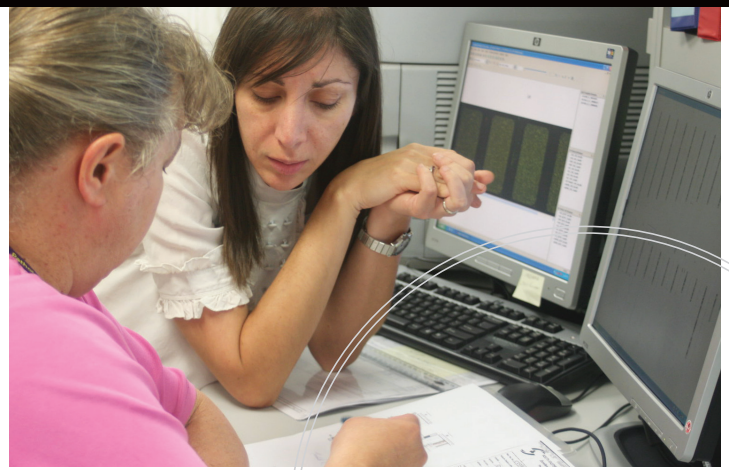
Medicine is Pathology

PATHOLOGISTS ARE INDISPENSABLE TO QUALITY PATIENT CARE

The Genetic Pathologist

RCPA
The Royal College of Pathologists of Australasia

PATHOLOGISTS ARE INDISPENSABLE TO OUR MEDICAL SYSTEM – BUT WHO ARE THEY, AND WHY ARE THEY SO CRUCIAL?



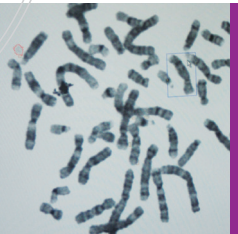
Genetics is one of the most complex areas of medicine. Genetic pathologists search for abnormalities in our DNA to help them diagnose and research disease.

Much of their work focuses on the key areas of prenatal diagnosis, predictive testing and the confirmation (or exclusion) of a diagnosis. In these areas, genetic pathologists liaise closely with clinicians to ensure the best possible outcome for patients.

Many genetic pathologists are also intimately involved with research and the introduction of new tests into laboratories. Currently, more than 400 genetic tests are available in Australia, and the number is continuing to increase.

Genetic pathologists perform more than seven thousand tests per million population per year nationally, and this number is growing rapidly.

The interpretation of genetic results is a very labour intensive process.



Enriching Patient Care

Genetic pathologists are involved in myriad aspects of patient care:

Prenatal testing: genetic pathologists examine chromosomes and DNA in foetal cells looking for possible abnormalities associated with conditions such as cystic fibrosis, Tay Sachs disease or brain malformations. They also test for extra or missing chromosomes in foetuses, in conditions such as Down Syndrome.

Carrier testing: to identify whether an individual carries a genetic abnormality that can be passed on to future children.

Predictive testing: essentially a 'risk assessment' for pre-symptomatic individuals about their genetic susceptibility to a particular disease, such as BRCA testing for breast cancer. This allows individuals to take preventative measures to stop the disease occurring or enables close monitoring and treatment at an early stage.

Confirmation of diagnosis: the confirmation or exclusion of a diagnosis will provide important indications for the patient's prognosis and indicators for the most appropriate therapy.

The Genetics Revolution: 'Tailored Medicine'

Genetic pathologists are intimately involved in advising clinicians on the most effective and appropriate treatment.

Increasingly, new therapies are tailored for patients with a particular genetic make up, or with a disease with a certain genetic profile. Genetic testing can identify patients who will benefit from a specific treatment. This is referred to as **'pharmacogenetics'.**

"It's the equivalent of trying to find a spelling mistake in an encyclopaedia."

Dr Melody Caramins

The implications of pharmacogenetics for the country's health system are enormous, including more targetted treatment, decreased side effects and improved survival rates.

Patient – Doctor – Laboratory Interface

Genetic pathologists provide an important interface between the laboratory and patients or patients' referring doctors.

A genetic pathologist has the knowledge and expertise to balance what the doctor is looking for and what the science is capable of. Specifically, they advise clinicians on the most appropriate approach to investigate a particular patient or family, and the best scientifically available test to be used to accurately and reliably make their diagnosis.

National Framework

With the 'explosion' of genetic knowledge in medicine, there is an increasing gap between the genetic testing that could be provided and the resources that are currently available.

Rapid advances and changes in the availability of genetic tests mean it is imperative that Australia and New Zealand implement appropriate frameworks to enable their populations to benefit from the 'genetics revolution'.

Counselling

The results of genetic tests may have wider implications, not just for the patient, but also for their future reproductive options and for other members of the family, so adequate counselling is always required.