



Fact File

The Royal College of Pathologists of Australasia

**Australian Pathologist Workforce 2018
Genetic Pathology**

AUSTRALIAN PATHOLOGIST WORKFORCE – GENETIC PATHOLOGY

Overview

The two main branches of Genetic Pathology are:

Medical Genomics which includes:

- Diagnostic detection and interpretation of genomic/ epigenetic variants in symptomatic patients (children, adults, foetuses);
- Pedigree analysis and diagnostic assessment of segregation in kindreds of disease-causing mutations or genomic regions;
- Diagnostic detection and interpretation of mosaic genomic variants e.g. in cancer, pregnancy, and inherited diseases (e.g. tumour material; constitutional mosaicism; foetal DNA in maternal blood, circulating tumour DNA etc. and quantitative assessment of mosaic genomic variants;
- Predictive/pre symptomatic assays in unaffected relatives (or a foetus) to determine the risk of inheritance of a familial disorder;
- Population-based screening for genomic abnormalities (antenatal and newborn screening programs);
- Application of probability, statistics, bioinformatic databases, and other aspects of computer science relevant to the practice of genetic pathology.

Biochemical Genetics which includes:

- Population-based screening for inborn errors of metabolism by enzyme, protein and metabolite assays, and diagnostic screening for inborn errors of metabolism in symptomatic patients by analysis of metabolites such as organic acids and amino acids;
- Diagnostic assays for specific disorders by analysis of specific analytes in body fluids, enzymatic studies, or DNA studies of specific genes;
- Predictive assays in unaffected relatives (or a foetus) to determine the risk of developing the disorder known to be present in the family;
- Pedigree analysis and diagnostic assessment of segregation in kindreds of disease-causing mutations or genomic regions
- Monitoring the biochemical status of patients for long term care or to guide acute care in metabolic crises.

Workforce profile and trends

There are only 18 Genetic Pathologists, with five females (27.8%) and 13 males (72.2%). There are only five Genetic Pathologists less than 45 years of age (two females, three males). There are no pathologists in this discipline less than 35 years.

The modal age ranges are 40 to 44 years, 55 to 59 years and 60 to 64 years (each with four pathologists).

Trends in trainee numbers

Genetic Pathology trainees increased from five to six trainees over the period 2011 to 2016. This was a low growth of 20% over the period.

Workforce demand and supply

There is an increasing awareness of genomics and molecular methodologies with increasing incorporation of genomics/molecular testing into routine practice. The value-add role was scored as high demand growth while cancer incidence and prevalence, precision medicine and genetic testing were scored as medium. There is increasing multidisciplinary management (value adding) in interpreting results because clinicians are not genomic/ molecular literate. There is an increasing incidence of cancer with the ageing population and the Genetic Pathologist has a role in pre-diagnosis i.e. determining at cancer risk genetic markers (e.g. BRACA), diagnosis and treatment design. In terms of precision medicine, cancer will be at the forefront of these developments with the need to consider the type of tumour, switches and delivery of targeted therapeutics. The use of liquid base cancer detection will also increase demand for genetic pathology services.

There is increasing evidence of cost effectiveness of genetic testing in healthcare with increased testing (demand and volume) and increasing complexity but technology may offset workforce demand to some extent. On the other hand the complexity of the interpretation of genomic results for genetic pathologists may offset this. Genetics services benefit from consolidation of laboratories as it fits into a group of specialties where this is the preferred model because of high expertise, low volume of samples and throughput in one lab is preferable.

The High Scenario Growth in Demand figure based on increase in MBS services listed was 9.1% and the Low Scenario Growth in Demand figure based on workforce growth was 2.4%

Results of workforce analysis

No quantitative modelling was undertaken for this workforce due to the small workforce size. The results of qualitative information gathering and quantitative data on the major growth in MBS service items indicate the need for growth in this discipline. This role is a particularly important one for pathologists to ensure quality standards and monitoring of standards are applied, as well as education regarding genetic literacy.

Therefore, there should be an additional number of trainees funded for this discipline.