

Information Sheet | Canberra Health Services

Diagnostic genomics

ACT Pathology provides a comprehensive clinical diagnostic service that includes conventional cytogenetic and molecular genomic techniques.

Conventional karyotyping

Conventional karyotyping plays a key role in both somatic (cancer) and constitutional (germline) investigations. Analysis of the structure and number of metaphase chromosomes is performed by skilled scientists, to identify acquired changes or germline variants.

Conventional karyotyping is required for the diagnosis and subsequent treatment of patients with haematological and other malignancies. e.g. AML, ALL, CML, lymphoma.

It is routinely used for couples (and individuals) experiencing fertility issues, including recurrent miscarriage. Diagnosis or exclusion of chromosomal variants informs family planning and treatment options.

Fluorescence In Situ Hybridisation (FISH)

Fluorescence in situ hybridisation is a molecular cytogenetic technique used to identify specific gene loci involved in chromosomal rearrangements in oncology samples such as;

- Acute Leukaemia
- Chronic Myeloid Leukaemia

Molecular PCR based tests

The diagnostic genomics laboratory performs several PCR based tests for inherited and somatic single gene mutations such as:

- Factor V Leiden and Prothrombin
- HLAB27
- BCR-ABL1 quantitative testing for the p210 transcript

Molecular karyotyping (Chromosomal microarray)

Chromosomal microarray is a molecular cytogenetics technique that uses single nucleotide polymorphisms (SNP) probe hybridisation to determine the copy number and zygosity of the SNP's. This provides a high resolution analysis of copy-number anomalies. This technique does not provide information on the chromosomal structure.

The diagnostic genomics laboratory performs SNP Microarray for recurrent miscarriage, and peri-natal postmortem.

Collection site for carrier screening and NIPT

ACT Pathology is a collection site for the following testing. Testing is provided by The Victorian Clinical Genetics Service (VCGS):

- carrier screening (individuals and couples)
- non-invasive prenatal testing

Specimen types

A variety of specimen types are used in diagnostic genomic testing, please contact the laboratory for specific specimen requirements.

For further information please contact

- Director 5124 7813
- Chief Scientist 5124 3449



Acknowledgement of Country

Canberra Health Services acknowledges the Ngunnawal people as traditional custodians of the ACT and recognises any other people or families with connection to the lands of the ACT and region. We acknowledge and respect their continuing culture and contribution to the life of this region.

ACT Pathology Publication PA12.06/ Edition:1

© Australian Capital Territory, Canberra 2024



Accessibility 📞 call (02) 5124 0000



Interpreter 📞 call 131 450

canberrahealthservices.act.gov.au/accessibility

