Fact Sheet

Diagnostic Genomics

The Diagnostic Genomics Department, ACT Pathology, Canberra Hospital, provides a comprehensive clinical diagnostic service that includes conventional cytogenomic and molecular cytogenomic techniques.

Conventional Karyotyping

Conventional karyotyping plays a key role in the accurate characterisation of the diagnosis and subsequent treatment of patients with haematological and other malignancies. eg. AML, ALL, CML, lymphoma and requires metaphase chromosomes where the whole of each chromosome is examined:

Fluorescence In Situ Hybridisation (FISH)

Fluorescence in situ hybridisation is a molecular cytogenomic technique used to identify specific gene loci involved in chromosomal rearrangements in haemato-oncology and solid tumour samples such as;

- Acute Leukaemia
- Chronic Myeloid Leukaemia
- Lymphoma
- Lung Cancer
- Brain Tumours
- Sarcomas

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
HLAB27
Hereditary Haemochromatosis
BCR-ABL1 quantitative testing for the p210 transcript
KRAS, NRAS, BRAF and EGFR investigation in colorectal cancer, metastatic melanoma and lung adenocarcinoma.

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

A/Prof Adrienne Morey	Director	51248350
Fiona Webb	Chief Scientist	51243449

Phone 02 5124 2932 Fax 02 5124 2815
actpathology@act.gov.au
www.health.act.gov.au/services/act-pathology
Gilmore Crescent Garran ACT 2605 PO Box 11 Woden ACT 2606

ACT PATHOLOGY