



Cytogenetics Service

A Complete Range of Cytogenetic Services for Pre Natal Diagnosis and Post Natal Analysis

The cytogenetics laboratory at Healthscope Pathology provides a comprehensive constitutional cytogenetics service.

Sue Dale, the head of the laboratory, has more than 30 years experience and is a Fellow of the Human Genetics Society of Australasia (Cytogenetics).

The laboratory is staffed by highly skilled, experienced cytogenetic scientists. Our team has in-depth knowledge of cytogenetic and fluorescence *in situ* hybridisation (FISH) analyses and takes pride in providing the highest quality of service to our customers.

We currently perform cytogenetic testing on amniocentesis samples, chorionic villi samples (CVS), products of conception, peripheral blood and tissue biopsy.

CHROMOSOME STUDIES

Chromosome studies are an important laboratory diagnostic procedure in certain patients with intellectual or developmental delay of unknown etiology, unexplained minor or major malformations, abnormalities of growth or sexual development and in individuals with apparent infertility or recurrent pregnancy loss.

Chromosome analysis on prenatal specimens is warranted in pregnancies with a higher Down Syndrome risk (due to age or high risk serum screening), when foetal abnormalities have been found on ultrasound, or when there is a known parental chromosome rearrangement.

Results of chromosome analysis on prenatal specimens are available within 10-14 days while results from peripheral blood can take 4-6 weeks.

focused on you

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FLUORESCENCE *IN SITU* HYBRIDISATION (FISH)

FISH is used as an adjunct to conventional cytogenetic analysis and provides a rapid screen for the most common numerical chromosome abnormalities observed in the newborn: trisomy 13, trisomy 18, trisomy 21 and aneuploidy for X or Y. Because these studies are performed on uncultured cells, results are available within 1-2 working days.

PRIORITY FOR URGENT CASES

We appreciate that waiting for the results of chromosome analysis is a very anxious time, so priority is routinely given to cases in the following patient categories:

- Abnormal ultrasound or maternal serum screening results.
- Late gestational age (>18 weeks).
- Pregnant patient with family history of chromosomal abnormality.
- Parents of a chromosomally abnormal foetus.
- Newborn with phenotypic abnormality.

OUR SERVICE

A 7 day courier service is available to pick up specimens collected at medical clinics.

- Clear, easy to interpret reports are produced.
- Once completed, reports will be faxed to doctors' rooms and a hard copy of the report will be sent in the mail.
- Abnormal reports are telephoned directly to the referring doctor.
- Specimen collection is available at any of Healthscope Pathology's many licensed collection centres. Collection of prenatal specimens is usually done by an experienced ultrasonographer.

TEST NAME	SPECIMEN REQUIREMENTS
Peripheral Blood Analysis	Lithium Heparin
Prenatal Analysis	Amniotic Fluid, Chorionic Villi
Solid Tissue Analysis	Products of conception or Foetal tissue
FISH	Peripheral Blood (Lithium Heparin), Amniotic fluid, Chorionic Villi, Solid tissue

Specimens need to be collected aseptically and blood specimens should not be placed on ice.

CONTACTS

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If you have any questions or require further information, please contact our Customer Service Centre.

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