



A Guide for Health Professionals

Genetic Services of WA provides a comprehensive state-wide diagnostic and confidential counselling service in clinical genetics.



Current DNA tests offered by our laboratories for diagnostic/carrier/predictive and antenatal testing include more than 100 conditions such as:

- Neurobehavioural including:
 - Muscular Dystrophy
 - Fragile X
 - Mitochondrial disorders
 - Prader-Willi/Angelman syndromes
 - Spinal Muscular Atrophy
 - Spino-Cerebellar ataxia
- Familial Cancer including:
 - Breast and Ovarian Cancer (BRCA1 & 2)
 - Familial Adenomatous Polyposis (FAP)
 - Hereditary Non-Polypotic Colonic Cancer (HNPCC, Lynch syndrome)
 - Neurofibromatosis
- Haematological/Immunological including:
 - Thalassaemia
 - Haemophilia A & B
- Metabolic including:
 - Alpha 1-antitrypsin deficiency
 - Galactosaemia
 - Congenital Adrenal Hyperplasia
- Other including:
 - Cystic Fibrosis
 - Oculocutaneous Albinism
 - Incontinentia Pigmenti
 - Deafness
 - Ectodermal dysplasia
 - Noonan syndrome
 - Skeletal dysplasias (including Achondroplasia)
 - Craniosynostosis
 - Tuberous Sclerosis

Cytogenetics and Molecular Cytogenetics

- Routine karyotype on blood, skin, fetal tissue including amniocentesis, CVS or cordocentesis.

FISH Studies for Microdeletions:

- DiGeorge region on 22q11.2
- Miller-Dieker syndrome on 17p13.3
- Prader-Willi syndrome on chromosome 15
- Sex determining region of Y chromosome
- Smith-Magenis syndrome on 17p11.2
- William syndrome on 7q11.3
- Wolf-Hirshhorn syndrome on 4p16.3.

For further DNA testing available locally and nationally, check the Human Genetics Society of Australasia website: www.hgsa.com.au

Inquiries regarding testing should be directed to GSWA, phone (08) 9340 1525.

Genetic Services of Western Australia

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Clinical Geneticists

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Familial Cancer Program

Tel: (08) 9340 1603 Fax: (08) 9340 1725

Genetic Paediatric Services

Tel: (08) 9340 1625 Fax: (08) 9340 1685



This document can be made available in alternative formats on request for a person with a disability.

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Genetic Services of WA (GSWA) is a multi-disciplinary service which offers the full range of diagnostic, educational and training programs relevant to the modern practice of medical genetics. Services are provided by clinical geneticists and counsellors working closely with DNA and cytogenetic laboratories and associated disciplines, such as diagnostic imaging and obstetrics. The protocols and procedures accord with the Human Genetic Society of Australasia Guidelines for Genetic Service.

Indications for Referral to Genetic Services

- A family history of a specific genetic disorder, particularly when considering pregnancy.
- An individual with features suggestive of a specific genetic disorder.
- Unexplained congenital abnormalities and childhood disabilities, e.g. intellectual, hearing, visual, skeletal.
- Familial clustering of a specific cancer, e.g. breast, bowel – particularly with two or more affected first degree relatives with a young age of onset.
- Fetal anomalies or abnormal pregnancy screening tests, e.g. ultrasound detected fetal structural anomalies or increased risk first or second trimester screening test results.
- Maternal age 37 years or greater at estimated date of delivery.
- Planning or confirmed pregnancy between close relatives e.g. first cousins.
- Exposure to known teratogens during pregnancy.
- Multiple miscarriages where a parental balanced chromosomal translocation has been identified.
- Unexplained male infertility with congenital absence of the vas deferens or less than 500,000 spermatozoa/ml.

Referring a Client

Clients are referred to GSWA by:

- GP or specialist
- hospital inpatient/outpatient ward or department
- self-referral.

How to arrange a referral:

- referral letter or consult request sent to GSWA.

This should include:

- patient's full name (including maiden name if relevant)
- patient's current address and contact details
- patient's date of birth
- reason for referral
- relevant personal or family history
- other medical information that may be relevant
- the referring doctor's name and address
- copies of relevant test results.

Please ensure that the client is informed of the referral to GSWA. Once the referral has been received, a genetic counsellor will contact the client to obtain further details and arrange an appointment.

Genetic Clinics

Clinics are either:

- general genetic clinics in public teaching hospitals or country regions
- disease specific or combined specialist clinics often integrated with other medical services.

Metro clinics	Focus	Times
KEMH	General genetic - antenatal - adult	Wed 9-12 Fri 9-12
PMH	General genetic - children	Mon 9-12 Thu 2-5
PMH	Genetic dermatological disease	Quarterly
PMH	Genetic eye disease	Monthly
Familial Cancer Program	Familial cancers	Wed 9-12 Fri 9-12
Outreach Clinics	Focus	Times
Joondalup	General genetic	Monthly
Rockingham	General genetic	Monthly
Albany Bunbury Geraldton Kalgoorlie Port Hedland	General genetic - adult - children	Approx. 3 monthly

Screening and Diagnostic Laboratory Services

Cytogenetic, DNA and Biochemistry services are available to the public. The DNA and cytogenetic laboratories have the infrastructure and expertise to provide a range of screening and diagnostic tests on specimens from any tissue, including fetal cells from antenatal procedures. The list of disorders for which testing is offered is dynamic and can be modified according to local needs and demands.