



Genetic Paediatric Services

Some of the reasons you may be referred for genetic consultation:

- Newborn Screening Program*
- Congenital anomalies
- Dysmorphic features
- Developmental delay
- Unexplained abnormality of growth
- Skeletal dysplasia
- Osteogenesis imperfecta
- Ehlers-Danlos & Marfan syndrome
- Neurofibromatosis
- Tuberous sclerosis
- Neuromuscular disorders
- Muscular dystrophy
- Myotonic dystrophy
- Spinal muscular atrophy
- Limb deficiency
- Ectodermal dysplasia
- Ichthyoses
- Retinoblastoma
- Retinal dystrophies
- Albinism
- Cleft lip and palate
- Craniofacial malformations
- Metabolic disorders
- Other genetic conditions

*Newborn Screening Program

The parents of newborn babies identified as carriers of Cystic Fibrosis through the Guthrie test (heel prick) are referred to Genetic Services for counselling and further assessment.

Contact details

Administration:

King Edward Memorial Hospital
374 Bagot Road, SUBIACO WA 6008
Tel: (08) 9340 1625
Fax: (08) 9340 1685

Clinic:

Princess Margaret Hospital
Level 5, Hay Street Building, SUBIACO WA 6008

Staff:

Clinical Geneticists Dr Jack Goldblatt, Director
 Dr Gareth Baynam
 Dr Cathy Kiraly
 Dr Fiona McKenzie
 Dr Sharron Townshend

Genetic Counsellors Stephanie Broley
 Karen Harrop
 Amy Pearn
 Marian Rawlins
 Michelle Ward

Secretary Anne Stevens



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What are Genetic Services?

The Genetic Paediatric Clinic is part of Genetic Services of Western Australia (GSWA). It is a state-wide service based at King Edward Memorial Hospital/Princess Margaret Hospital in Perth. It provides diagnosis, assessment and genetic counselling to parents/families of children with single or multiple congenital anomalies, dysmorphic features, cytogenetic disorders, disorders of special senses or organ systems, developmental delay or unexplained abnormalities of growth. The service also provides diagnosis, assessment and counselling for patients with disorders involving bones, joints and other connective tissues, neurogenetic disorders, genetic disorders involving the skin and/or teeth and genetic eye disorders.

These services are provided through a multidisciplinary team that includes clinical geneticists and genetic counsellors, working closely with DNA and cytogenetic laboratories and other associated disciplines.

What are hereditary conditions?

Hereditary (genetic) conditions are problems in growth, development and health which result from changes (mutations) in the genetic information, which may be inherited in families.

The genetic information is passed from parents to their children in their genes. However, it is important to note that not all genetic conditions are “inherited”, ie sometimes a genetic condition occurs for the first time in an individual.

What is a Clinical Geneticist?

The clinical geneticist is a medical practitioner with a genetics specialty, also certified by the Human Genetics Society of Australasia and the College of Physicians of Australia, to provide clinical genetics services.

What is a Genetic Counsellor?

Genetic Counsellors are usually graduate health professionals with specialist training in genetics and counselling. They undergo a training process certified by the Human Genetics Society of Australasia to provide genetic counselling in conjunction with a clinical geneticist.

What happens at a genetic appointment?

The genetic counsellor and/or geneticist will discuss with you any of your concerns and answer your questions. Your family’s medical details and family tree will be discussed and confirmed with you. Your child may be examined and their height, weight and head circumference measurements recorded.

Sometimes it is necessary to gather more information and/or arrange tests.

Clinics

Clinics are held by our staff at:

- Princess Margaret Hospital for Children
- Joondalup Child Development Centre
- Rockingham/Kwinana District Hospital
- Country Outreach Clinics at:
 - Bunbury
 - Albany
 - Port Hedland
 - Kalgoorlie
 - Geraldton

What should you bring to the genetic appointment?

- Your child
- Child health record (yellow or purple book)
- Family photos
- Recent reports and investigations
- A support person if required

Review appointments

Sometimes the Geneticist will arrange a review appointment to answer further questions or to discuss results of investigations. You will be notified of such an appointment.

Please contact one of our genetic counsellors if you wish to discuss/arrange a review appointment.