

Some of the reasons you may be referred for genetic consultation

- Newborn Screening Program*
- Congenital anomalies
- Facial differences (also called 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
- Craniofacial malformations
- Metabolic disorders
- Other genetic conditions.

* 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.

For more information contact

Genetic Services of Western Australia Genetic Paediatric Services

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Website: <http://kemh.health.wa.gov.au/services/genetics/index.htm>



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

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Government of Western Australia
Department of Health

Genetic Paediatric Services



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Genetic Paediatric Services

Our Service

Genetic Paediatric Services is part of Genetic Services of Western Australia (GSWA), a statewide service based in Perth. Our regular clinics are currently held at Princess Margaret Hospital and King Edward Memorial Hospital.

Children are referred to our service for many reasons, most commonly when a parent or doctor has concerns about a child's growth, development or health. Our role is to provide assessment of children to determine if an underlying cause can be identified. We can also provide information, support and resources.

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

About us

- **Genetic counsellors** are health professionals with training in genetics and counselling. We provide information and support to individuals and families who have concerns about a genetic condition in their family.
- **Geneticists** are specialist medical doctors with training in clinical genetics and genetic counselling. We offer genetic counselling, diagnostic services and arrange genetic testing.



Before your genetic appointment

In order to accurately assess your family, it is important to gather as much information as possible about the family health history.

- Once we receive a referral we will send you forms to help you collect your family history details.
- We may also send you consent forms that allow us to access records confirming other diagnoses in the family.
- We may contact you for further information prior to your appointment. Copies of medical reports, doctor's letters and/or other documentation may be useful.

All information provided before, during or after your appointment is treated confidentially.

What to expect at your appointment

- an opportunity to raise your questions and concerns
- a review of your family health history
- a physical examination may be required
- sometimes it is necessary to gather more information and/or arrange further tests.

What should you bring to the genetic appointment?

- your child
- child health record (yellow or purple book)
- family photos
- recent reports and investigations.

You are welcome to bring a support person or other family members.

Outreach Services

Metropolitan outreach genetic clinics are held in Joondalup and Rockingham.

Regional outreach genetic clinics are held in:

- Albany
- Bunbury
- Geraldton
- Kalgoorlie
- Port Hedland.

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.