Tasmanian
Clinical
Genetics
Service

Paediatric Genetics Clinic

What to expect at your appointment

This booklet provides an overview of our service and what you can expect at your appointment.

Why has my child been referred to the Genetics Clinic?

A paediatrician, or occasionally a different specialist or a GP, may refer a baby or child to the Genetics Clinic if the child

- has a physical or medical problem
- is delayed in reaching developmental milestones
- has learning difficulties
- has unusual physical features
- any combination of the above.

The referring doctor may suspect that there is a genetic cause underlying the problems, or may be seeking reassurance that there is *not* a genetic cause.

A chromosome test may have shown a variation and your doctor may refer you to the Genetics Clinic for advice on how to interpret the variation.

To answer the question, the Genetics clinic may use a combination of information including:

- the child's health history
- the family's health history
- a physical examination of the child, and sometimes parents or siblings
- chromosome testing of the parents of the child (ideally completed before the appointment)
- results of x-rays, other medical imaging, or blood tests.

In some cases the Genetic Clinic will be able to identify the genetic condition that is responsible for the problems. If a diagnosis can be made, this can be helpful because:

- It enables the doctors to predict the course of the condition, and be alert for any problems not yet observed in the child which are associated with that condition.
- There may be treatments or medications that are known to help for that condition.
- Many (but not all) genetic conditions are inherited – knowing the genetic cause can help clarify the likelihood of the condition occurring in subsequent pregnancies or in other family members.

In other cases, the Genetic Clinic will be able to provide reassurance that the child's problems are <u>not</u> genetic.

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Sometimes, despite a suspicion that the problems are genetic it will not be possible to make a diagnosis. Sometimes there is no blood test that can be done. Sometimes the results of a blood test are inconclusive or not useful in getting to the cause of the problem. In instances where the Genetics team cannot identify the underlying cause of a condition this can be frustrating for the parents.

Who will I meet at the Genetics Clinic?

The Tasmanian Clinical Genetics Service is staffed by Medical Geneticists and Genetic Counsellors. Depending on the issue in question, your appointment may be with a Medical Geneticist or a Genetic Counsellor, or both may be present.

Medical Geneticist
A Medical Geneticist is a medical doctor who has trained in a speciality, usually paediatrics, and has then gone on to further specialise in the diagnosis and management of conditions that are genetic or inherited. This doctor is sometimes referred to as a Consultant, or a Clinical Geneticist.

Genetics Fellow
A genetics fellow is a fully qualified medical doctor who has already trained in a specialty, such as paediatrics, and is in the process of training to be a Medical Geneticist.

Genetic Counsellor
A Genetic Counsellor is a person who has special training in both genetics and counselling. The Genetic Counsellor will arrange your appointment, and try to make sure things run smoothly while you are at the clinic. The Genetic Counsellor will also follow up on any concerns you might have following your clinic appointment. The Genetic Counsellor can also help you to understand the medical information you have been given, and the effects this information could have on you and your family. If a diagnosis of a genetic condition had already been made in your family, a Genetic Counsellor can provide advice about recurrence risk, arrange genetic testing for relatives, and organise pre-natal testing where needed.

Students
Medical students or genetic counselling students are required to observe a certain number of appointments as part of their training. This is a very valuable learning experience for them. You may be asked if you would be happy to have a student 'sit-in' on your appointment. If you are not comfortable with this it is OK to say 'no' and this will not affect the care you receive.

What will happen in the appointment?

Family history
Usually a genetic counsellor will telephone you to arrange the appointment and at that time he/she will ask you some questions about your family's health history. He/she will use this information to draw a genetic family tree. The genetic counsellor may ask about your relatives, including their names, dates of birth and death, causes of death and general health. If you have relatives who have already visited a Genetics Clinic in Tasmania or interstate, or if you have a relative who has been diagnosed with a genetic condition, or if you have relatives with unexplained physical, medical or developmental problems or learning difficulties, it would be helpful to let the genetic counsellor know, in case these are relevant to your child's situation.

Your child's history
At your appointment, the medical geneticist will ask about your pregnancy, the child's birth and early development, and will want to hear your perspective on any problems or issues the child may have.

Examination
The medical geneticist may wish to physically examine your child. Sometimes he/she will also wish to examine the parents or other family members, to compare family features.

Photos
Sometimes the medical geneticist may wish to take photos of your child. Usually this will be so that he can seek the opinion of his colleagues if he is unsure about a diagnosis. Occasionally it may be for research purposes. The geneticist will ask you to sign a consent form if he would like to take photos. It is OK to say 'no'.

Blood tests
The medical geneticist may request a blood test for your child. Depending on the age of the child and the hospital where you are seen, it may be possible for blood to be collected at the end of the appointment, or a separate appointment at the paediatric out-patient clinic may be required. If so this will be arranged for you. Depending on the test required, sometimes a saliva sample or cheek swab may be acceptable as an alternative to a blood test.

Other tests:
Sometimes the medical geneticist will want to organise other tests before making a diagnosis. Typically these include x-rays or other types of medical imaging. Sometimes the medical geneticist will refer your child to a different specialist such as an ophthalmologist (eye doctor) or a neurologist for an opinion on a particular issue. Usually these tests or referrals will be organised on the spot, but occasionally the medical geneticist will write back to your paediatrician with suggestions for further tests or referrals.

What should I bring?

Information about your family history
If there are details about the family health history that you cannot recall at the time the genetic counselor telephones you, it may be helpful if you are able to track these down prior to your appointment and bring this information with you. For older children it can be helpful for the medical geneticist to see photographs of your child as a baby or toddler. Sometimes a photograph of the whole family can be helpful if the mother/father or siblings will not be attending the appointment.

Your child's medical history
It is often helpful to keep the whole medical history together in one folder and bring it all along to every appointment. It may be helpful if you bring along the personal health record book (blue book) you received when your child was born.

Family member
You may want to bring your partner, a friend, or another family member. A family member can provide moral support, and can also help with information about family health history.

Interpreters
If English is not your first language, the hospital you are visiting can arrange an interpreter, so please contact the Genetic Counsellor to discuss this. Even if you speak some English, it can be helpful to have an interpreter when complicated medical matters are being discussed.

How long will it take?

Waiting time
Although everyone involved tries to run to time, sometimes appointments can take longer than expected for any number of reasons. A wait of more than 20 minutes would be very unusual.

The appointment
Each appointment usually takes approximately 1 hour.

What will it cost?

There is no charge for an appointment at the Genetics Clinic. Depending on your circumstances you may be required to bring with you, or forward a referral letter from your GP or specialist, and to bring your medicare card with you to the appointment.

The vast majority of genetic tests requested by the Genetics team are provided free of charge. In rare circumstances, such as when a specialised test is not available in Australia and is performed in a laboratory overseas, there may be charges that will be payable by the patient. If you will be required to pay for a test, this will be made very clear to you before you are asked to decide whether you want to go ahead with testing or not.

What will happen afterwards?

Written information
A letter will be written summarising your appointment. This will be sent to you, your referring doctor and usually your GP. If you think there is another person who should receive it please let us know.

Waiting for results
It can be an anxious time waiting for results. If blood tests or imaging have been organised, ask the medical geneticist how long it will be before you can expect to receive the results, and how you will be given the results (eg by telephone or in person).

Living with a diagnosis
To be told that your child has a genetic condition can be a life-altering experience. Sometimes a diagnosis provides answers to questions parents have had for many years. Sometimes, however, a diagnosis can come as a great shock. We will try to meet your needs for information at a pace that suits you so that you are not overwhelmed with information, nor left with unanswered questions. Advice and support from parents or patients living with the same condition can be invaluable at the time of a new diagnosis. For rarer genetic conditions, the nearest support group may be interstate or even overseas, however most support groups use the internet, email and newsletters to keep in touch with their members. Ask the Genetic Counsellor if he/she can give you the contact details of the most relevant group for you and your family.

Living without a diagnosis
As life-altering as receiving a specific diagnosis can be, to not receive a diagnosis but still believe something is wrong can be equally as difficult. The Genetic Clinic may suggest another appointment in a year or so to review the situation, as new tests are constantly being developed. Sometimes with time a child will develop new features that give clues to a diagnosis.

Privacy

Your appointment at the Genetics Clinic will remain confidential between you and the Medical Geneticist/Genetic Counsellor. We appreciate the special and sometimes sensitive nature of family health information. We keep your family health information securely stored in a Genetic File. Information in a Genetic File can only be accessed by staff of the Tasmanian Clinical Genetics Service.

A letter summarising your appointment is usually sent to you, with a copy to your referring doctor, and to any other doctors involved in your care that you nominate. If you do not want to receive a letter, or you want to nominate exactly which doctors are/are not to receive a letter, please discuss this with the Medical Geneticist/Genetic Counsellor at your appointment. Usually, a copy of the letter (not including the family tree and family health information) is filed in your child's confidential hospital record.

Details of your appointment will not be disclosed to friends or family members who enquire, or who attend the clinic following your appointment, without your express permission.

What if I am not satisfied?

Opportunities for Improvement
The Tasmanian Clinical Genetics Service is constantly striving to improve the service that it provides to its patients. If your experience was less than ideal, we would be grateful for your comments so that we can prevent similar experiences in the future. The Genetic Counsellors are responsible for receiving and resolving patient complaints or concerns regarding clinic appointments. Please don't hesitate to contact them on 6166 8296 with your feedback.

How do I contact the Tasmanian Clinical Genetics Service?

The office of the Tasmanian Clinical Genetics Service is in Hobart and enquiries should be directed to this office, even if your appointment is scheduled for Launceston or Latrobe.

Tasmanian Clinical Genetics Service
c/- Royal Hobart Hospital
PO Box 1061
HOBART TAS 7001

Telephone: 03 6166 8296
Fax: 03 6173 0328
Website: http://www.dhhs.tas.gov.au/services/
(type genetic counselling into the search field)

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