

# Genetic testing and screening

Genetic testing is common. Almost every baby born in Australia has a blood sample taken from their heel within a few days of birth to look for the genetic condition phenylketonuria (where the body lacks a specific enzyme, which causes abnormal metabolism that may result in brain damage) and a range of other conditions (see fact sheet on '*Newborn screening*').

Testing is also done in many other situations:

- To make a diagnosis for someone who has a medical condition
- To look for altered genes in the relatives of people with a genetic condition
- To look for carriers of altered genes in people of certain ethnic groups
- To look at people with certain medical conditions such as breast cancer, to see if they have an altered gene, which indicates that they have a high likelihood of getting the condition
- To screen for, or diagnose, a genetic condition during pregnancy
- To work out if someone could develop a medical condition in the future
- To identify paternity and other important relationships in times of dispute or in coronial inquiries.

If you are considering having genetic testing, you should be aware that it has both advantages and disadvantages.

The advantages are that:

- You might clarify an uncertain situation
- You might find your fears were unfounded
- You might find that while you have a genetic condition or gene alteration you'd rather not have, at least you now know and can work out what to do
- What you learn might have implications for others in your family.

The disadvantages are that:

- You may learn something that, with hindsight, you'd rather not have learnt
- What you learn might have implications for others in your family
- What you learn might cause problems for you with issues like life insurance.

The limitations are:

- No test is perfect – sometimes results can give guidance only, sometimes the result will be unclear and, on rare occasions, the result can give false impressions
- A test can't predict what will happen to a person – it can only give an idea of what might happen.

If you're thinking about having a test for a genetic condition, then you need to talk to a doctor or genetic counsellor before the test. You need to find out:

- What it means for you and your family if the test is positive, or if it's negative, or if the result is unclear
- Whether or not it might be wise to take out life insurance before the test, so premiums are not affected by test results. Note that you have to declare your family history, regardless of whether or not you have had genetic testing
- The cost – some genetic tests are provided free through public hospitals, some are covered by Medicare and some have to be paid for.

### **Contacts and further information**

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- For other related fact sheets, you can contact the Gene Technology Information Service on **free call Australia-wide 1800 631 276** or email [gtis-australia@unimelb.edu.au](mailto:gtis-australia@unimelb.edu.au) or visit Biotechnology Australia's website at <http://www.biotechnology.gov.au>