

## Haemoglobinopathies (including thalassaemias)

Haemoglobin is an important protein in the blood. Haemoglobin is found in red blood cells and carries oxygen from the lungs to all the tissues.

Various genetic conditions affect haemoglobin, making it less able to carry oxygen, or causing damage to the red blood cell it is in.

The most common problems are thalassaemia (a genetic disorder inherited from both parents resulting in a reduction in the amount of haemoglobin produced by the body) and sickle cell anaemia (a genetic disease in which red blood cells may change shape under certain circumstances; this causes problems when the cells become stuck in blood vessels that carry oxygen and nutrients to individual cells). About 5 in every 100 people throughout the world are carriers for one of these conditions. Haemoglobinopathies are more common in some parts of the world than in others, particularly where malaria is common.

Haemoglobinopathies may be mild or they may be severe. They can cause a range of illnesses from slight tiredness, to severe anaemia (not enough red blood cells) needing regular blood transfusions, to episodes of severe pain.

You should consider having a blood test to see if you are a carrier for one of these conditions and possibly see a genetic counsellor if you:

- Have a relative with one of these conditions, or have a relative who is a carrier for one of these conditions
- Have unexplained anaemia or have people in the family with unexplained anaemia
- Have ancestry from where these conditions are common – southern Europe, the Middle East, South-East Asia, Africa, the Indian subcontinent, South America, the Caribbean and the Pacific Islands.

This is particularly important if you are pregnant or could become pregnant.

## Contacts and further information

- Your local genetic service, which you can contact through your nearest community health centre, public hospital or health department.
- Thalassaemia Society of Victoria at <http://www.tsv.org.au>
- National Organization for Rare Disorders at <http://www.rarediseases.org>
- Australasian Genetic Alliance at <http://www.australasiangeneticalliance.org.au>
- Better Health Channel at <http://www.betterhealth.vic.gov.au>
- MyDr at <http://www.mydr.com.au>
- The Centre for Genetics Education at <http://www.genetics.edu.au>
- HealthInsite at <http://www.healthinsite.com>
- MedicineNet at <http://www.medicinenet.com>
- 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>