TESTING IN AUSTRALIA

Recent immigration to Australia means screening programs, particularly antenatal testing, are important. In Melbourne approximately 10% of women in their first pregnancy required DNA studies to adequately characterise their carrier state and to provide sufficient information to estimate the risk that their children would have severe disease.

The identification of carriers of clinically significant haemoglobinopathies is a two-stage process. Initially evidence for the carrier state is sought by carrying out a full blood examination and Hb electrophoresis.

Iron deficiency can be excluded as a complicating factor by iron studies in individuals who show a haematological abnormality consistent with this diagnosis.

In this relatively simple way evidence for all but single gene deletion α-thalassaemia (and most nondeletional point mutations) will usually be obtained.

Where appropriate, further studies including DNA analysis can then be carried out for final clarification of the carrier state. In this way it is usually possible to identify all but a few mutations and to provide informative counselling for individuals and couples.

Ref: https://www.nps.org.au/australian-prescriber/articles/screening-for-thalassaemia

READINGS & WEBSITES

Thalassaemia International Federation (TIF):
http://thalassaemia.org.cy/

TIF Publications including - Prevention and diagnosis of haemoglobinopathies: A short guide for health professionals:
http://thalassaemia.org.cy/publications/tif-publications/

Screening for Thalassaemia - NPS MedicineWise:
https://www.nps.org.au/australian-prescriber/articles/screening-for-thalassaemia

Genetics in General Practice - RACGP:
https://www.racgp.org.au/afp/2014/july/genetics-in-general-practice/

GPs - What you need to know about sickle cell and thalassaemia screening (UK Govt.):
https://phescreening.blog.gov.uk/2016/10/26/gpswhat-you-need-to-know-about-sickle-cell-and-thalassaemia-screening/

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CARRIER SCREENING FOR HAEMOGLOBINOPATHIES & KNOW MY TRAIT AWARENESS CAMPAIGN

+ **Information for General Practitioners**

KnowMyTrait

knowmytrait.org

THALASSAEMIA & SICKLE CELL ANEMIA
& THE GP

WHAT IS IT?

The thalassaemias are the commonest single gene disorders in the world's population and are a common cause of hereditary anaemia and allied complexities like iron-overload.

The **Major forms or disorders** require lifelong blood transfusion and daily medication treatment. In Australia, patients are usually within the care of a hospital or specialist treatment centre.

CARRIER SCREENING

The **Minor form or trait is a carrier state**. This carrier population often have little or no symptoms and are unaware. Some carriers may have mild anaemia and knowledge of their carrier trait is an important for medical treatment of iron deficiency.

The minor form should be suspected in any individual who has reduced red blood cell indices.

The trait can be passed on by one parent. It is important for people to be aware of their carrier status. When both parents carry the genetic trait there is a 25% risk of the child inheriting the major disorder or disease.

WE ASK

We ask you, as a family GP, to support awareness and encourage blood testing for the genetic trait where you feel appropriate. The test is covered by Medicare.

ANCESTRAL RISK

People have increased risk if they have an ancestry/ethnic background from:

Mediterranean / Southern European, African (including Americas and Caribbean), Middle Eastern, Chinese, Indian subcontinent, Central and South East Asian, Pacific Islander, New Zealand Maori, South American and some northern Western Australian and Northern Territory Indigenous communities.

INVESTIGATIONS / REFERRAL

Mean corpuscular volume, mean corpuscular haemoglobin, ferritin, haemoglobin electrophoresis and iron levels.

Where appropriate, seek advice from haematology or genetic services about DNA testing (esp. for alpha-thalassaemia carriers).

Couples should be tested prior to pregnancy or in first trimester.

Ref:
https://www.racgp.org.au/afp/2014/july/geneticsin-general-practice/

INFORMATION & SUPPORT

NSW & ACT
Thalassaemia Society of NSW
http://thalnsw.org.au

VICTORIA & REST OF AUSTRALIA
Thalassaemia & Sickle Cell Society of Australia
www.tascsa.net.au

THE PICTURE IN NSW

There are between 230 - 250 transfusion dependent patients within NSW/ACT. It is likely, there are between 60,000 - 100,000 carriers of the genetic trait for the various haemoglobin disorders in NSW/ACT. **Most of them would be unaware.**

Western Sydney is likely to be the main geographical location for carriers. Liverpool Hospital currently has the highest number of transfusion-dependent patients.

Patients in Liverpool Hospital come from Middle Eastern and South-East Asian countries, while Prince of Wales Hospital and Royal Prince Alfred Hospital's patient profile includes significant numbers of patients with Mediterranean ethnicity and some African ethnicity.

TREATMENT / REFERRALS

POWH and RPAH are specialist centres with haematologists and clinical nurses having specialist knowledge for thalassemia and sickle cell anaemia. Liverpool Hospital while not a primary specialist centre has specialist knowledge and holds a monthly multidisciplinary meeting for haematologists and paediatricians.

The treatment of children is provided at Westmead Children's Hospital *(highest patient case load)*, Sydney Children's Hospital and John Hunter Hospital Newcastle.

Outreach for adults and children is also conducted at hospitals in Canberra, Campbelltown and Bankstown.