# Australian Genetics Services and Support Organisations

Centralised contacts for Genetics Services, familial cancer services, prenatal testing and support groups

<table>
<thead>
<tr>
<th>Location</th>
<th>Central Contact</th>
<th>Umbrella Support Organisation</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Australian Capital Territory</strong></td>
<td>The Canberra Hospital&lt;br&gt;PO Box 11, Woden, ACT, 2605&lt;br&gt;Ph: (02) 6244 2133&lt;br&gt;Fax: (02) 6244 4625</td>
<td>Self-Help Organisations United Together (SHOUT)&lt;br&gt;Pearce Community Centre – Collett Place, Pearce ACT, 2607&lt;br&gt;Ph: (02) 6290 1984&lt;br&gt;Email: <a href="mailto:admin@shout.org.au">admin@shout.org.au</a>&lt;br&gt;Fax: (02) 6286 4475&lt;br&gt;Website: <a href="http://www.shout.org.au">www.shout.org.au</a></td>
</tr>
<tr>
<td><strong>New South Wales</strong></td>
<td>The Centre for Genetics Education&lt;br&gt;PO Box 317, St Leonards, NSW 1590&lt;br&gt;Ph: (02) 9926 7324&lt;br&gt;Fax: (02) 9906 7529</td>
<td>Association of Genetic Support of Australasia (AGSA) Inc.&lt;br&gt;66 Albion Street, Surry Hills, NSW 2010&lt;br&gt;Ph: (02) 9211 1462&lt;br&gt;Email: <a href="mailto:info@agsa-geneticsupport.org.au">info@agsa-geneticsupport.org.au</a>&lt;br&gt;Fax: (02) 9211 8077&lt;br&gt;Website: <a href="http://www.agsa-geneticsupport.org.au">www.agsa-geneticsupport.org.au</a></td>
</tr>
<tr>
<td><strong>Northern Territory</strong></td>
<td>No service currently available&lt;br&gt;N/A</td>
<td>N/A</td>
</tr>
<tr>
<td><strong>Queensland</strong></td>
<td>Herston Hospital Complex&lt;br&gt;Genetic Health Queensland&lt;br&gt;Level 4, Building C28, Back Road Herston, QLD 4029&lt;br&gt;Ph: (07) 3636 1686&lt;br&gt;Fax: (07) 3636 1987</td>
<td>Self Help Queensland (SHQ) Inc.&lt;br&gt;121 Lister Street, Sunnybank, QLD 4109&lt;br&gt;Ph/Fax: (07) 3344 6919&lt;br&gt;Email: <a href="mailto:selfhelp@gil.com.au">selfhelp@gil.com.au</a>&lt;br&gt;Website: <a href="http://www.selfhelpqld.org.au">www.selfhelpqld.org.au</a></td>
</tr>
<tr>
<td><strong>South Australia</strong></td>
<td>Women and Children’s Hospital&lt;br&gt;South Australian Clinical Genetics Service&lt;br&gt;Youth and Women’s Health Service&lt;br&gt;72 King William Road, North Adelaide, SA 5006&lt;br&gt;Ph: (08) 8161 7375&lt;br&gt;Fax: (08) 8161 6088</td>
<td>Genetic Support Network Victoria (GSNV)&lt;br&gt;Royal Children’s Hospital, Flemington Road, Parkville, VIC 3052&lt;br&gt;Ph: (03) 8341 6315&lt;br&gt;Email: <a href="mailto:info@gsnv.org.au">info@gsnv.org.au</a>&lt;br&gt;Fax: (03) 8341 6390&lt;br&gt;Website: <a href="http://www.gsnv.org.au">www.gsnv.org.au</a></td>
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<tr>
<td><strong>Tasmania</strong></td>
<td>Royal Hobart Hospital&lt;br&gt;Tasmanian Clinical Genetics Service&lt;br&gt;GPO Box 1061L, Hobart, TAS 7001&lt;br&gt;Ph: (03) 6222 8296&lt;br&gt;Fax: (03) 6222 7961</td>
<td>Genetic Support Network Victoria (GSNV)&lt;br&gt;Royal Children’s Hospital, Flemington Road, Parkville, VIC 3052&lt;br&gt;Ph: (03) 8341 6315&lt;br&gt;Email: <a href="mailto:info@gsnv.org.au">info@gsnv.org.au</a>&lt;br&gt;Fax: (03) 8341 6390&lt;br&gt;Website: <a href="http://www.gsnv.org.au">www.gsnv.org.au</a></td>
</tr>
<tr>
<td><strong>Victoria</strong></td>
<td>Royal Children’s Hospital&lt;br&gt;Genetic Health Services Victoria&lt;br&gt;Flemington Road, Parkville, VIC 3052&lt;br&gt;Ph: (03) 8341 6201&lt;br&gt;Fax: (03) 8341 6390</td>
<td>Genetic Support Network Victoria (GSNV)&lt;br&gt;Royal Children’s Hospital, Flemington Road, Parkville, VIC 3052&lt;br&gt;Ph: (03) 8341 6315&lt;br&gt;Email: <a href="mailto:info@gsnv.org.au">info@gsnv.org.au</a>&lt;br&gt;Fax: (03) 8341 6390&lt;br&gt;Website: <a href="http://www.gsnv.org.au">www.gsnv.org.au</a></td>
</tr>
<tr>
<td><strong>Western Australia</strong></td>
<td>King Edward Memorial Hospital&lt;br&gt;Genetics Services of Western Australia,&lt;br&gt;374 Bagot Road, Subiaco, WA 6008&lt;br&gt;Ph: (08) 9340 1525&lt;br&gt;Fax: (08) 9340 1678</td>
<td>Genetic Support Council WA (GSCWA)&lt;br&gt;Level 1, Oasis Lotteries House, Unit 9, 37 Hampden Road, Nedlands, WA 6009&lt;br&gt;Ph: (08) 9389 6722&lt;br&gt;Email: <a href="mailto:info@geneticsupportcouncil.org.au">info@geneticsupportcouncil.org.au</a>&lt;br&gt;Website: <a href="http://geneticsupportcouncil.org.au">http://geneticsupportcouncil.org.au</a></td>
</tr>
</tbody>
</table>
Family history

- Where possible, take a three generational family history including 1° relatives (children, siblings, parents) and 2° relatives (aunts, uncles and grandparents) on both sides of the family
- Note:
  - Ethnic background (ancestry and culture)
  - Adoption
  - Age at diagnosis
  - Age and cause of death
  - Birth defects, stillbirths and miscarriages
- There are no insurance or privacy implications from taking a family history.

Genetic testing and screening in Australia

- The following DNA tests are available on the MBS:
  - HFE (haemochromatosis)
  - Fragile X syndrome
  - Factor V Leiden and some other inherited thrombophilias
- Inform the patient about the purpose and personal/family implications of a genetic test prior to obtaining consent
- Some genetic tests may be available at no cost to the patient through Genetics Services
- Other genetic tests may be available at a cost to the patient ranging from $200 to many thousands of dollars
- Patients who have had a predictive or presymptomatic genetic test have a duty to inform life insurers of the test result when applying for a new, or altering an existing, policy
- A GP has no duty to inform the relatives of a patient about a positive genetic test result. The patient should be encouraged and supported to share the information with their relatives.

Newborn screening

- Conditions in newborn screening include:
  - Cystic fibrosis
  - Phenylketonuria (PKU)
  - Galactosaemia (not available in Victoria)
  - Congenital hypothyroidism
  - Over 20 other rare metabolic conditions.

Testing and pregnancy

- Pre-pregnancy and pregnancy counselling relevant to genetics should include:
  - Family health history
  - Information on prenatal screening and diagnostic tests
  - Prenatal diagnosis indicated where specific genetic conditions exist in family
  - Folic acid before and during pregnancy:
    - 400µg most women
    - 5mg high risk women:
      - Family history or previous neural tube defect
      - Epileptic medication
  - Carrier tests relevant to the couple’s ethnic background:
    - Haemoglobinopathies
    - Cystic fibrosis
    - Ashkenazi Jewish specific conditions
  - Drug and medication use.

Figure 1. Maternal age-specific chance of having a live-born baby with Down syndrome

The maternal age (background risk) is modified by the gestational age, ultrasound (NT) and maternal hormone measurements to provide an overall (adjusted risk) of the fetus having Down syndrome. It is the adjusted risk that will usually determine whether to offer further diagnostic tests.
### Advantages and disadvantages of screening tests during pregnancy

<table>
<thead>
<tr>
<th>Screening test</th>
<th>Gestation (weeks)</th>
<th>% Down syndrome pregnancies detected</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
</table>
| Combined first trimester screening | 10-12 (blood test) 11.5-13 (ultrasound with nuchal translucency) | 85-90%                              | • Early screen and therefore early diagnosis  
• Highest detection rate  
• No added risk of miscarriage  
• Detection of some fetal abnormalities  
• Benefits relating to early scan:  
  > Accurate dating  
  > Diagnosis of multiple pregnancy  
  > Diagnosis of early pregnancy failure (miscarriage) | • Will detect some affected pregnancies that may spontaneously miscarry  
• Does not provide risk for neural tube defects but the ultrasound may detect anencephaly  
• Women may not access services so early in the pregnancy  
• Ultrasound requires accredited operator for accuracy  
• Out-of-pocket expenses vary |
| Second trimester maternal serum screening | 14-20 (15-17 ideal) | 70-75%  | • Available to women presenting in second trimester  
• No added risk of miscarriage  
• No out-of-pocket expenses for public patients if arranged through public hospital | • Later screening test  
• Inaccurate dates can result in inaccurate risk by calculations. A dating scan should be considered if dates are uncertain  
• Lower detection rate  
• No neural tube risk can be given if test done at 14 weeks  
• Out-of-pocket expenses vary |

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### Advantages and disadvantages of diagnostic tests during pregnancy

<table>
<thead>
<tr>
<th>Diagnostic Test</th>
<th>Gestation (weeks)</th>
<th>% of Down syndrome pregnancies detected</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
</table>
| Chorionic villus sampling (CVS) a | From 11 weeks b   | > 99%                                  | • Early detection  
• Definitive diagnosis  
• Results potentially available in time for termination of pregnancy (TOP) by curette | • Miscarriage risk (~ 1% above background in expert hands)  
• Detects chromosomally abnormal pregnancies that may otherwise spontaneously miscarry  
• 1% risk of equivocal results (placental mosaicism or maternal cell contamination of sample)  
• 0.1% failure to detect chromosome abnormality (abnormality is present in fetus but not in placenta, or maternal cell contamination of sample) |
| Amniocentesis a                   | From 15 weeks c   | 100%                                   | • Test with lowest miscarriage rate  
• Definitive diagnosis | • Miscarriage risk (~ 0.5% above background in expert hands)  
• Diagnosis in second trimester, when pregnancy is more established  
• Results available too late for TOP by curette – TOP may need to be performed by induction of labour or vaginal evacuation |

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*False positive rate set at 5% at these detection rates
*b Nuchal translucency measurement should be performed by a Fetal Medicine Foundation (FMF) and RANZCOG (Royal Australian and New Zealand College of Obstetrics and Gynaecology) accredited operator
*c May be limited access in some states: in Victoria, it is currently not funded for public patients; in Queensland the blood test is not available publicly
*d Assumes the use of the quadruple test (four analytes) and ultrasound dating

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*Out-of-pocket expenses vary for CVS, amniocentesis and scans according to state. Public patients attending a tertiary public hospital may not be charged if identified as ‘increased risk’
*b The timing of CVS is not uniform throughout Australia, e.g. in South Australia it is sometimes offered from 10 weeks gestation
*c Procedures after 20 weeks gestation may not provide results in a timeframe permitting second trimester termination of pregnancy. Refer to your State abortion laws, and policies of local perinatal units
### Cancer in the family

- Familial cancer services provide risk assessment, genetic counselling and, for high risk families, genetic testing for a causative mutation.
- Predictive genetic testing is only available where a causative mutation has been found in an affected relative.
- One or more 1° relatives with melanoma:
  - Annual skin examination and advice on self examination.
  - Genetic testing in research phase.
- Men with one or more 1° relatives diagnosed with prostate cancer under the age of 60 yrs:
  - Discuss risks and benefits of PSA and digital rectal examination screening.
  - Genetic testing in research phase.

**Family history criteria for triaging into moderately increased risk and potentially high risk for breast, ovarian and bowel cancer.**

<table>
<thead>
<tr>
<th>Breast Cancer</th>
<th>Ovarian Cancer</th>
<th>Bowel Cancer</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Moderately increased risk:</strong> Offer mammography every 2 yrs for women 50-69 yrs. Women aged 40-49 yrs are also eligible for free mammography. Mammographic screening is not recommended for women younger than 40 yrs.</td>
<td></td>
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</tr>
<tr>
<td>- One 1° relative diagnosed with breast cancer before the age of 50; or</td>
<td></td>
<td></td>
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<tr>
<td>- Two 1° relatives, on the same side of the family, diagnosed with breast cancer; or</td>
<td></td>
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</tr>
<tr>
<td>- Two 2° relatives, on the same side of the family, diagnosed with breast cancer, at least one before the age of 50.</td>
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<tr>
<td><strong>Potentially high risk:</strong> Refer to a familial cancer service or to Genetics Services.</td>
<td></td>
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<tr>
<td>- A woman at potentially high risk for ovarian cancer; or</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Two 1° or 2° relatives on the same side of the family diagnosed with breast or ovarian cancer, plus one or more of the following features occurs on the same side of the family:</td>
<td></td>
<td></td>
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<tr>
<td>- Additional relative(s) with breast or ovarian cancer</td>
<td></td>
<td></td>
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<tr>
<td>- Breast cancer diagnosed before the age of 40 yrs</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Bilateral breast cancer</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Breast and ovarian cancer in the same woman</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Breast cancer in a male relative</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Ashkenazi Jewish ancestry; or</td>
<td></td>
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</tr>
<tr>
<td>- One 1° or 2° relative diagnosed with breast cancer at age 45 or younger plus another 1° or 2° relative on the same side of the family with sarcoma at age 45 or younger; or</td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Member of a family in which a high risk breast cancer gene has been established.</td>
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<td></td>
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</tbody>
</table>

### Diabetes

- Screen for diabetes every 3 yrs for the following:
  - People over 45 yrs with a 1° relative with type 2 diabetes.
- Screen for diabetes every 12 months for the following:
  - People of Aboriginal and Torres Strait Islander origin >35 yrs.
  - People of Pacific Islander, Indian subcontinent or Chinese origin >35 yrs.
- Consider maturity onset diabetes of the young (MODY) if:
  - Early mild hyperglycaemia (<25 yrs).
  - Non-ketotic.
  - No association with obesity.
  - Autosomal dominant pattern of inheritance.

**Genetic testing for diabetes is not routinely available.**
Genetic testing is available through specialist cardiac or Genetics Services for:

- Familial hypercholesterolaemia (FH)
- Some forms of cardiomyopathy
- Primary arrhythmogenic disorders such as familial long QT syndrome
- Cardiac effects associated with connective tissue conditions (Marfan and Ehlers-Danlos syndromes)
- Congenital heart defects associated with specific syndromes

*Diagnostic criteria for familial hypercholesterolaemia are based on the modified UK criteria:

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Definite FH</td>
<td>(e or f) + a</td>
</tr>
<tr>
<td>Probable FH</td>
<td>(e or f) + b</td>
</tr>
<tr>
<td>Possible FH</td>
<td>(e or f) + (c or d)</td>
</tr>
</tbody>
</table>

• In a family history of sudden unexpected cardiac death, particularly in the young (<35yrs), consider referral of all 1° relatives to specialist cardiac and Genetics Services.

**Cardiovascular disease**

**Clotting and bleeding conditions**

**Hereditary thrombophilies**

- Screening for thrombophilia should be considered in individuals with:
  - DVT <50 yrs
  - Spontaneous thrombosis in absence of recognised risk factors
  - Recurrent thrombosis
  - Family history of thrombosis
  - Thrombosis in unusual sites eg CNS, abdominal veins, upper limb
  - Stillbirth or fetal death in utero

- A thrombophilia screen (factor V Leiden, prothrombin variants, antithrombin III deficiency, protein C deficiency, protein S deficiency and activated protein C resistance) is available on the MBS only if the patient has:
  - A personal history of proven venous thromboembolism or pulmonary embolism, or
  - A 1° relative who has a proven defect of any of the above

**Haemophiliases**

- Haemophilia A is caused by low or absent factor VIII and haemophilia B is caused by low or absent factor IX
- X-linked recessive pattern of inheritance
- Males are affected
- 10% of female carriers can have mild bleeding symptoms.

**Chromosomal conditions**

- Consider karyotype for:
  - Intellectual disability/developmental delay
- Consider parental karyotype for:
  - Personal/family history of recurrent miscarriage
  - Infertility
  - Family history of intellectual disability/developmental delay
- Where karyotypes are abnormal, refer to Genetics Services.

**Fragile X syndrome & other causes of developmental delay**

- Consider a karyotype and DNA test for fragile X syndrome (available on the MBS) in children and adults with one or more of the following features:
  - Developmental delay including intellectual disability
  - Autistic-like features
  - Behavioural and emotional problems
  - Dysmorphic features
  - Relative with a fragile X mutation
- Males are usually more severely affected than females
- Refer to Genetics Services for cascade testing of relatives
- There is no known single gene that causes autism; genetic testing is not currently available.

**Cystic fibrosis**

- Northern European ancestry: 1 in 25 are carriers
- 95% of new cases of CF will be detected by newborn screening
- Refer to a paediatrician/respiratory physician for sweat test where there is clinical suspicion of CF regardless of the newborn screening result
- All 1° relatives of an affected child should be referred to Genetics Services for cascade testing
- Only the common mutations for cystic fibrosis are tested in carrier screening
- Prenatal genetic diagnosis is available for CF.

**Neurological conditions**

- Diagnostic genetic testing is available through neurological/Genetics Services for:
  - Huntington disease (HD)
  - Early-onset familial Alzheimer disease (EoFAD)
- Criteria for EoFAD:
  - 2 or more affected relatives with onset <65 yrs in more than one generation with clinical or pathological diagnosis of Alzheimer disease
  - Relative with identified causative mutation
- Consider referral of asymptomatic at-risk relatives to Genetics Services for counselling and discussion of predictive genetic testing for HD and EoFAD.
Hereditary haemochromatosis

- Order iron studies (fasting transferrin saturation and serum ferritin) in the presence of one or more of the following:
  > Lethargy and weakness
  > Arthralgia
  > Loss of libido
  > Upper abdominal discomfort
  > Hepatomegaly
  > Grey/bronze skin pigmentation
  > Testicular atrophy
  > Joint swelling/tenderness
- Consider HFE gene testing or referral to a gastroenterologist if a patient has:
  > Fasting transferrin saturation > 45% or
  > Fasting serum ferritin >250µg/L (pre-menopausal women) or > 300 µg/L (post-menopausal women or men)
- Discuss cascade testing for 1˚ and 2˚ relatives when an individual is diagnosed with HH or found to be heterozygous for an HFE mutation
- The MBS covers HFE gene testing for patients with:
  > Raised ferritin or transferrin saturation levels on more than 1 occasion, or
  > 1˚ relative diagnosed with HH or with two HFE mutations.

Neurofibromatosis (NF)

- NF1 characterised by multiple café au lait spots, inguinal/axillary freckling and multiple neurofibromas
  > Wide range in severity of symptoms
- NF2 characterised by bilateral vestibular schwannomas; may include gradual hearing loss, balance problems and tinnitus
- Autosomal dominant pattern of inheritance; 50% of cases due to sporadic mutation
- Genetic testing NF1
  > Not necessary for diagnosis after birth
  > Prenatal genetic testing possible only when the family specific gene mutation is known
- Genetic testing NF2
  > Presymptomatic genetic testing is available to blood relatives of individuals in whom a mutation has been identified.

Psychiatric conditions

- There is a general population risk of 1-2% of developing a form of mental illness such as schizophrenia and bipolar disorder during a person’s lifetime
- A diagnosis of schizophrenia or bipolar disorder increases the risk for 1˚ relatives

<table>
<thead>
<tr>
<th>Affected relative</th>
<th>Schizophrenia (risk %)</th>
<th>Bipolar disorder (risk %)</th>
</tr>
</thead>
<tbody>
<tr>
<td>No close relative (general population)</td>
<td>1</td>
<td>2-3</td>
</tr>
<tr>
<td>Sibling</td>
<td>9</td>
<td>13</td>
</tr>
<tr>
<td>Parent</td>
<td>13</td>
<td>15</td>
</tr>
<tr>
<td>Sibling and 1 parent</td>
<td>15</td>
<td>20</td>
</tr>
<tr>
<td>Both parents</td>
<td>45</td>
<td>50</td>
</tr>
</tbody>
</table>
- Genetic testing for schizophrenia or bipolar disorder is not available.

Haemoglobinopathies

- Include α-thalassaemia, β-thalassaemia, sickle cell disease and other haemoglobin variant conditions
- Haemoglobinopathy carrier testing should be discussed as part of pre-pregnancy/prenatal care in the following individuals:
  > Family history of anaemia/thalassaemia/abnormal haemoglobin variant
  > From any of the following ethnic backgrounds: Southern European, Middle Eastern, African, Chinese, South-East Asian, Indian subcontinent, Pacific Islander, New Zealand Maori, South American and some northern Western Australian and Northern Territory Indigenous communities
  > Partners of any of the above at-risk patients
  > MCV <81fL or MCH <27pg
- If abnormal blood tests, refer to Genetics Services and/or haematology clinic for genetic counselling and testing. This is particularly urgent for pregnant couples.

Interpretation of haemoglobinopathy carrier testing results

<table>
<thead>
<tr>
<th>MCH (pg)</th>
<th>Ferritin</th>
<th>Haemoglobin electrophoresis</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>&gt; or = 27</td>
<td>Normal</td>
<td>HbF increased</td>
<td>Carrier for α-thalassaemia</td>
</tr>
<tr>
<td></td>
<td>Normal</td>
<td>HbA2 increased</td>
<td>Carrier for β-thalassaemia</td>
</tr>
<tr>
<td></td>
<td>Low</td>
<td>HbS present</td>
<td>Carrier for sickle cell disease</td>
</tr>
<tr>
<td>&lt; 27</td>
<td>Normal</td>
<td>HbA2 normal</td>
<td>Carrier for α-thalassaemia</td>
</tr>
<tr>
<td></td>
<td>HbH present</td>
<td></td>
<td>Carrier for α-thalassaemia</td>
</tr>
<tr>
<td></td>
<td>HbS present</td>
<td></td>
<td>Carrier for sickle cell disease</td>
</tr>
<tr>
<td></td>
<td>Normal</td>
<td>DNA testing indicated</td>
<td>Possible co-existent thalassaemia carrier state</td>
</tr>
<tr>
<td></td>
<td>Low</td>
<td>Iron deficiency</td>
<td>Thalassaemia may coexist</td>
</tr>
</tbody>
</table>

Contributors:

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National Health and Medical Research Council

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