

Haemoglobin E (HbE)

What is Haemoglobin E?

Haemoglobin E (HbE) is a common inherited condition caused by the production of an abnormal haemoglobin protein. Haemoglobin is a protein in the blood that carries oxygen around our bodies.

HbE is passed from parent to child in genes. Genes carry information about human characteristics such as eye colour, hair colour and haemoglobin.

HbE is inherited.

HbE is not contagious.

HbE is not transmitted by germs.

Sometimes changes occur to genes, resulting in medical conditions. Such changes occur to beta (β) globin genes:

- A person normally inherits two β globin genes for the production of the beta globin protein in haemoglobin.
- A person may have the HbE alteration (mutation) in one of their two β globin genes. This person is called a **carrier of Haemoglobin E (HbE)** and is **healthy**. Carriers may be at risk of having a child affected with a severe blood condition.
- A person may have the HbE alteration (mutation) in both copies of their β globin genes. This person is said to be **homozygous** for HbE and has no major health problems apart from being slightly anaemic. They may be at risk of having a child affected with a severe blood condition.
- When a person is a carrier of the HbE alteration (mutation) and another type of β globin gene alteration (mutation) they may be affected with a severe blood condition that requires treatment. For example: when a person has inherited one copy of the HbE alteration (mutation) from one parent and one copy of a β thalassaemia gene alteration (mutation) from the other parent this results in a severe condition called HbE/ β thalassaemia (see Figure 2).

Treatment

HbE is not a severe condition on its own, however when combined with another β globin gene alteration it may result in a severe anaemia that requires life long treatment including blood transfusions to correct the anaemia.

The health of carriers of HbE and homozygous HbE

A carrier of HbE and individuals who are homozygous for HbE can expect to be **healthy**. It is important that their doctor knows their carrier status.

HbE and family planning

The HbE gene alteration is most common in people of South East Asian origin (Thailand, Burma, Cambodia, Vietnam, Laos and Indonesia) and Sri Lanka.

Couples planning a family, or early in pregnancy, should have a blood test to determine whether or not they are carriers, if the origin of either of their families is one of the areas listed above; or if they have a family history of any blood disorder or anaemia. This test is needed to determine if there is any risk of having a child affected by a genetic blood disorder.

Those at risk of having an affected child have options. A foetus can be diagnosed as early as the 12th week of pregnancy. Termination of pregnancy can then be considered, if appropriate. People can adopt or can consider assisted reproductive techniques (such as the use of donor eggs or donor sperm). Others may choose to take the chance of having an affected child. All of these options can be discussed with a Genetic Counsellor.

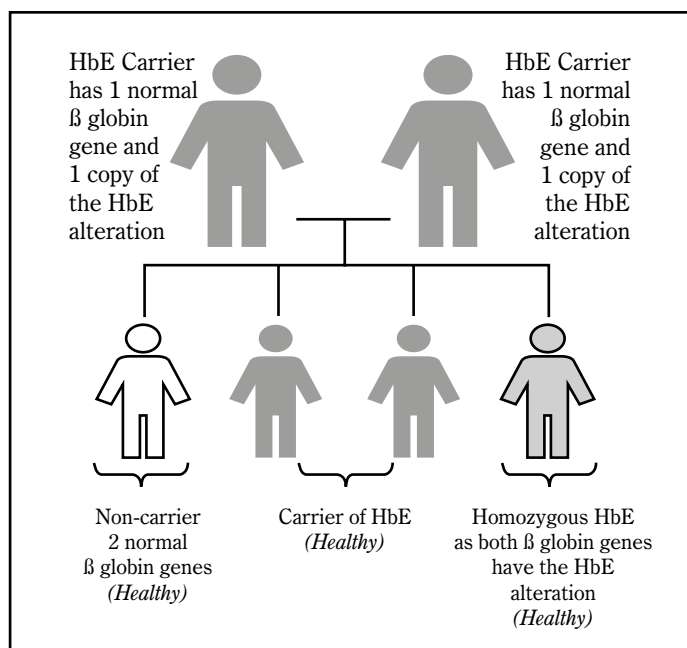
Testing can be arranged by your local doctor or by contacting the hospitals listed at the end of this pamphlet.

Important information for your family

If you are a carrier of HbE, other members of your family may also be carriers and at risk of having children with a severe blood condition. It is recommended that other family members **and** their partners be tested for their carrier status **prior** to having children of their own.

Outcomes of passing the HbE alteration to children

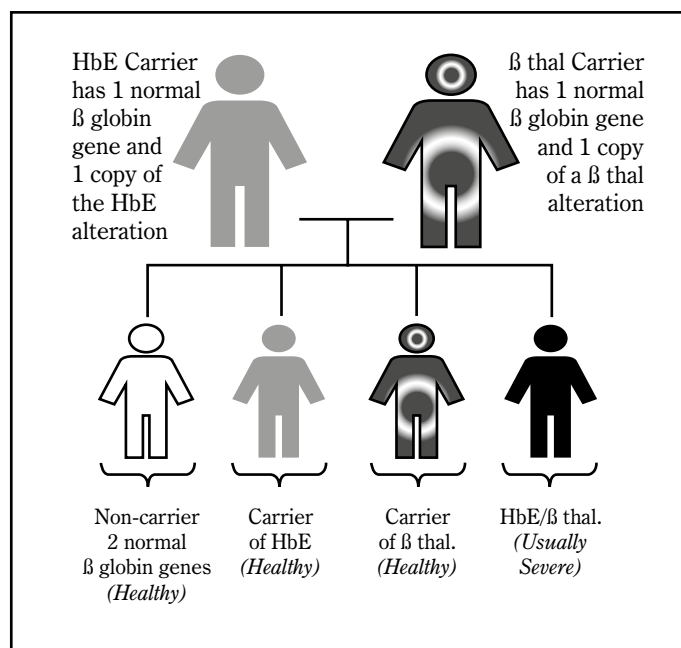
Figure 1:
Both parents are carriers of HbE.



With **each** pregnancy, this couple has a:

- 1 in 4 chance of having a child with 2 normal β globin genes.
- 2 in 4 chance of having a carrier of HbE.
- 1 in 4 chance of having a child who is homozygous HbE.

Figure 2:
One parent is a carrier of HbE and the other is a carrier β thalassaemia (β thal)



With **each** pregnancy, this couple has a:

- 1 in 4 chance of having a child with 2 normal β globin genes.
- 1 in 4 chance of having a carrier of HbE.
- 1 in 4 chance of having a carrier of β thalassaemia.
- 1 in 4 chance of having a child affected by HbE/ β thalassaemia.

Useful contacts

Thalassaemia Society of NSW

PO Box M120 CAMPERDOWN NSW 2050
Level 7 King George V Building
Missenden Road
CAMPERDOWN NSW 2050
Phone: +61 2 9550 4844
Mobile: 0400 116 393
www.thalns.org.au

Children's Hospital at Westmead

Haematology Department
Cnr Hawkesbury Rd and Hainsworth St,
Westmead NSW 2145
Ph: +61 2 9845 0000

Sydney Children's Hospital

Department of Haematology
High Street, Randwick NSW 2031
Ph: +61 2 9382 1111

The Prince of Wales Hospital

Department of Haematology
High Street, Randwick NSW 2031
Ph: +61 2 9382 4982

Royal Prince Alfred Hospital

Department of Haematology
Level 5, Missenden Road
Camperdown NSW 2050
Ph: +61 2 9515 7013

Westmead Hospital

Department of Haematology
Cnr Hawkesbury Road & Darcy Roads
Westmead NSW 2145
Ph: +61 2 9845 5555

Liverpool Hospital

Department of Haematology
Ground Floor
Cnr Elizabeth & Goulburn Street
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