

Produced by Specialist Haematology, Canterbury Health Laboratories

What is Haemoglobin?

Haemoglobin (Hb) is a protein found in red blood cells; it carries oxygen from the lungs to all parts of the body and gives blood its red colour. Hb is made up of haem groups, which contain iron and globin chains which are made of protein.

The combination of different globin chains with the haem groups produces the different types of haemoglobin present in people. The normal adult haemoglobin is Hb A.

Where a person has inherited Hb A from both parents, they will have the usual Hb combination, Hb AA.

What is Haemoglobin D?

Haemoglobin D (Hb D) is a variant Hb and occurs when the structure of the globin chain is altered.

Heterozygous haemoglobin D (haemoglobin D trait) occurs when a person inherits the normal haemoglobin from one parent and the haemoglobin D from the other (Hb AD).

When a person inherits the haemoglobin D from both parents, they are said to have homozygous haemoglobin D (Hb DD).

What is Thalassaemia?

Thalassaemia is a type of inherited anaemia (red blood cell deficiency).

People with thalassaemia don't produce enough globin chains. The two main types are called alpha (α) and beta (β) thalassaemia, depending on which globin chains are affected.

Thalassaemia occurs most frequently in people of

- Southern Asian
- Mediterranean, Middle Eastern
- Indian, African and Polynesian ancestry.

Alpha Thalassaemia

People with α thalassaemia don't produce enough α globin chains. To produce normal amounts of α globin, a total of **four** α globin genes are needed; two from each parent.

Depending on the number of functioning alpha globin genes, alpha thalassaemia can range from a very mild condition through to the most severe form which occurs when all four alpha globin genes are missing resulting in fetal or newborn death.

There are two types of α thalassaemia:

- α^+ thalassaemia
- α^0 thalassaemia

What is Haemoglobin D / Alpha Thalassaemia?

Haemoglobin D/alpha thalassaemia, can arise from either the inheritance of Hb D from one parent and alpha thalassaemia from the other or the inheritance of both the Hb D and the alpha thalassaemia from the same parent.

If a person is born with Hb D/alpha thalassaemia, they will have it all their life. It is **not** contagious or infectious.

Most people with heterozygous Hb D/alpha thalassaemia have a α^+ thalassaemia which is a mild form of α thalassaemia commonly seen in Indian and Middle Eastern people.

What is the Treatment?

Heterozygous Hb D/alpha thalassaemia is a relatively mild disorder. *It is **not** an illness and requires no regular medical follow-up or treatment.*

However, it may be mistaken for iron deficiency anaemia and iron tablets may be given unnecessarily. Iron tablets should only be taken if your doctor has done a special blood test to check that you lack iron.

Homozygous Hb D/alpha thalassaemia usually produces a moderate degree of anaemia.

Variant haemoglobins such as Hb D can interfere with other laboratory tests, for example, the measurement of haemoglobin A1c (a test used to assess diabetic control).

How Will This Affect My Children?

Every child inherits haemoglobin genes from each parent. The type of haemoglobin gene inherited from the parent determines the type of haemoglobin the child will have.

A person with Hb D/alpha thalassaemia will pass on either a Hb D gene or a Hb A gene **and** either an α -thalassaemia gene or a normal α -gene to each of their children.

When **both** parents carry a gene for a variant haemoglobin or thalassaemia, there is a chance that any child could have a severe form of thalassaemia or variant haemoglobin.

Why is Knowing About Haemoglobin D/alpha Thalassaemia so Important?

The inheritance of Hb D/alpha thalassaemia produces a mild disorder.

There are other types of variant haemoglobins or thalassaemia which if inherited together with Hb D can produce a more severe condition.

If you have Hb D/alpha thalassaemia it is important to have your partner tested for the presence of a variant haemoglobin or thalassaemia before you have children.

Is There a Test for Haemoglobin D and/or Thalassemia?

YES ! Blood tests can show whether an individual has haemoglobin D, thalassaemia or a combination of the two.

If you have Hb D/alpha thalassaemia it is important to have your partner tested for thalassaemia or a variant Hb before you have children. Other members of your family who are of reproductive age should also be tested.

The documentation of carrier status is important as:

1. It avoids the need for repeat testing in the future.
2. Thalassaemia trait either alone or in combination with a variant Hb, may be mistaken for iron deficiency anaemia and iron tablets may be given unnecessarily.
3. Haemoglobin D and α thalassaemia are inherited disorders which could be passed on to your child.

If two people who carry a variant haemoglobin or thalassaemia do become parents and there is a risk of a severe disorder in the fetus, special prenatal tests can be done to detect or rule this out.

Early diagnosis of a possible severe form of thalassaemia is important so that treatment can prevent as many complications as possible.

Can Haemoglobin D or Thalassemia Be Prevented?

Haemoglobin D and/or thalassaemia cannot be prevented at this time, but a programme of health education, testing for the trait and genetic counselling can provide families with full medical information to help them have healthy children.

People who think they may have Haemoglobin D and/or thalassaemia should go to their family doctor for advice and testing.

Your family doctor can contact a Haematologist at the Community Laboratory or the Haematology Department at Canterbury Health Laboratories on (03) 364 0300 if further clinical input is required.

The Specialist Haematology Laboratory at Canterbury Health Laboratories provides a comprehensive diagnostic service in conjunction with the family doctor.

Contact for Canterbury Health Laboratories

Laboratory Services: Specialist Haematology Laboratory, Canterbury Health Laboratories.

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On behalf of the Clinical Haematology Team at Christchurch Hospital