

Alpha Thalassaemia

What is Alpha Thalassaemia?

In the blood stream oxygen is carried in the red cells. The chemical that carries the oxygen is called haemoglobin. The haemoglobin is made up of four pieces or subunits, and these come in two varieties alpha (α) and beta (β). The Thalassaemias are caused by a decrease in production of haemoglobin. A reduction in alpha chain production causes alpha thalassaemia

Why do people get it?

Alpha Thalassaemia is an inherited disorder.

We all receive a thirty thousand genes or genetic messages from our parents. The genes are the blueprints for how many fingers and toes we will have the colour of our hair etc. The genes come in matching pairs, one from each parent. The genes are carried like beads on strings called chromosomes and we all have 46 chromosomes. In males and females 44 of the chromosomes can be matched into 22 pairs. The last pair are known as the sex chromosomes and are different in males and females. Males receive a Y chromosome from their father and an X chromosome from their mother. Females receive an X chromosome from both parents.

We all probably carry two or three genes that are faulty but the other copy in most cases is enough to get by on.

We all receive four alpha haemoglobin genes, two from each parent. They are located on chromosome 16. They are arranged next to each other and are represented $\alpha\alpha/\alpha\alpha$. Alpha Thalassaemia is due to a deletion or mutation of one or more of the four alpha haemoglobin genes located on chromosome 16.

How can you tell who has it?

The alpha Thalassaemias can be generally categorized as: Silent Carrier, Alpha Thalassaemia Trait, Hemoglobin H disease, and Alpha Thalassaemia major.

The Silent Carrier status is characterized by three functional genes of alpha haemoglobin ($-\alpha/\alpha\alpha$). Outside the newborn period, it is not possible to make this diagnosis by conventional methods. The silent carrier will experience no health problems in his/her lifetime.

Alpha thalassaemia trait is characterized by two functional genes. The two genes can either occur on the same chromosome (cis-type) ($--/\alpha\alpha$) or on each of the pair (trans-type) ($-\alpha/-\alpha$). Cis-type α -Thalassaemia trait tends to be found in individuals of Asian descent, while the trans-type tends to run in individuals of African descent. The individual with a Thalassaemia trait will experience no significant health problems except a possible slight anemia, which cannot be treated with iron. During pregnancy, the anemia can be mistaken for anemia of pregnancy. Cis-type can be co-inherited with another cis-type to result in alpha Thalassaemia major.

Haemoglobin H disease is characterised by one functional gene (α-α-β-β). Haemoglobin H disease should be considered in the case of a neonate in whom all of the red blood cells are very pale (hypochromic).

The most severe form of alpha thalassaemia is Alpha Thalassaemia Major or hydrops fetalis, characterised by a deletion of all four genes (α-α-β-β).

How common is it?

The Thalassaemias occur predominately in tropical and subtropical areas of Europe, Africa and Asia. Thalassaemia is uncommon in native Northern Europeans.

What treatment is available?

There is no treatment needed for silent carriers or trait. There are a variety of treatments for haemoglobin H. The treatment of alpha thalassaemia major is extremely difficult

What about future children?

Brothers and sisters of affected individuals are at increased risk.

If both parents are carriers of the cis arrangement there is:

- a one in four chance that the baby will receive no copies of the normal genes and will develop Thalassaemia major
- a one in four chance that the baby will receive four alpha haemoglobin genes and not carry the condition
- a two in four chance that the baby will not have the condition but will be trait.

Are other relatives at risk?

The risk of other relatives being affected is small but if they are worried they should seek advice.

Can it be diagnosed before birth?

As we all receive our genes at conception it is possible to look for gene changes and work out which of our parents' chromosomes we have received.

Further Information

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