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Understanding alpha thalassaemia

Haemoglobinopathies Service

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Introduction

Alpha thalassaemia can only be diagnosed by specific DNA testing. However, you may have been told you are suspected of being an alpha thalassaemia carrier due to the appearance of your red blood cells and your ethnic origin. DNA testing is not routinely required as alpha thalassaemia carriers are usually healthy and have no symptoms.

What is thalassaemia?

Thalassaemia is inherited blood disorder. This means it is passed to a child from one or both parents through their genes .Genes are the building blocks of the body and control most physical characteristics such as eye colour, hair type and nose shape.

When you have thalassaemia, your body makes less haemoglobin than normal. Haemoglobin is a substance in the red blood cells which carries oxygen to all parts of the body.

Thalassaemia can cause mild or severe anaemia . Anaemia occurs when your body does not make enough red blood cells or haemoglobin. The severity depends on the number of genes affected.

There are two main forms of thalassaemia : alpha and beta, and each type is affected by different genes. This leaflet is about alpha thalassaemia.

What causes alpha thalassaemia?

Normal haemoglobin is made up of four alpha globin genes and two beta globin genes. Alpha thalassaemia occurs when some or all of the four alpha globin genes are missing or damaged. There are two types of alpha thalassaemia:

Alpha Plus Thalassaemia
People whose parents, grandparents or ancestors originate from Africa, the Caribbean, India, Pakistan and Bangladesh have a higher incidence of alpha plus thalassaemia. It is a harmless condition. There are no clinically significant implications for a child when both parents carry alpha plus thalassaemia as this combination cannot cause a serious inherited anaemia in their children (see figure 1).

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Alpha Zero Thalassaemia

People whose parents, grandparents or ancestors originate from China, Hong Kong, Taiwan, Thailand, Cambodia, Laos, Vietnam, Burma, Malaysia, Singapore, Indonesia, Philippines, Cyprus, Greece, Sardinia, Turkey, Southern Italy or if family origins are unknown are considered high risk for being carriers of alpha zero thalassaemia.

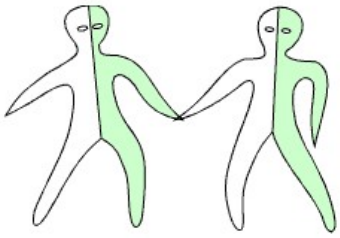
People whose ancestors are from the Middle East are considered medium risk for being carriers of alpha zero thalassaemia.

There are implications for a child when one parent carries **alpha zero thalassaemia** and the other carries **alpha plus thalassaemia**. This couple could have a child with **haemoglobin H disease** (see figure 2).

There are serious implications when both partners carry **alpha zero thalassaemia** as this couple could have a child with **alpha zero thalassaemia major** which is not compatible with life (figure 3).

Figure 1

There are no clinically significant implications for a child when **both** parents carry alpha plus thalassaemia as this combination cannot cause a serious inherited anaemia in their children.

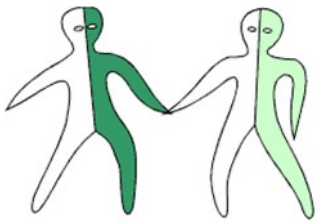
<table>
<tr><td>Parent who carries alpha plus thalassaemia</td><td colspan="2">  </td><td>Parent who carries alpha plus thalassaemia</td></tr>
<tr><td>  </td><td>  </td><td>  </td><td>  </td></tr>
<tr><td>Not a carrier</td><td>Carrier of alpha plus thalassaemia</td><td>Carrier of alpha plus thalassaemia</td><td>Child with "homozygous alpha plus thalassaemia"</td></tr>
</table>

In each pregnancy, there are three possibilities:

- 1 in 4 chance the child will not carry any alpha plus thalassaemia gene.
- 2 in 4 chance the child will carry alpha plus thalassaemia. This is harmless.
- 1 in 4 chance the child will inherit alpha plus thalassaemia. This is harmless.

Figure 2

This shows the implications for a child when one parent carries **alpha zero thalassaemia** and the other carries **alpha plus thalassaemia**. This couple could have a child with **haemoglobin H disease**.

Parent who carries alpha zero thalassaemia			Parent who carries alpha plus thalassaemia
			
Not a carrier	Carrier of alpha zero thalassaemia	Carrier of alpha plus thalassaemia	Child with "haemoglobin H disease"

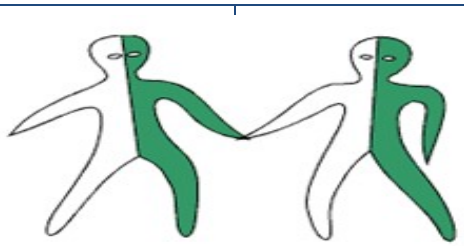
In each pregnancy, there are four possibilities:

- 1 in 4 chance the child will not carry any haemoglobin disorder.
- 1 in 4 chance the child will carry alpha plus thalassaemia. This is harmless.
- 1 in 4 chance the child will carry alpha zero thalassaemia. This is harmless.
- 1 in 4 chance the child will inherit alpha plus thalassaemia from one parent and alpha zero thalassaemia from the other. This child will have an inherited anaemia called **haemoglobin H disease**.

Haemoglobin H disease is a moderate form of anaemia. Most people with haemoglobin H disease lead a normal life. They go to school, work and have children just like other people. They occasionally need additional medical treatment, and should attend a haematology clinic every year for a check-up.

Figure 3

This shows the implications for a child when **both** parents carry **alpha zero thalassaemia** This couple could have a child with **alpha zero thalassaemia major.**

Parent who carries alpha zero thalassaemia			Parent who carries alpha zero thalassaemia
			
Not a carrier	Carrier of alpha zero thalassaemia	Carrier of alpha zero thalassaemia	Child with alpha zero thalassaemia major

In each pregnancy, there are three possibilities:

- 1 in 4 chance the child will not carry any haemoglobin disorder.
- 2 in 4 chance the child will carry alpha zero thalassaemia. This is harmless.
- 1 in 4 chance the child will inherit alpha zero thalassaemia from both parents. This baby would have **alpha zero thalassaemia major**. This is also called alpha thalassaemia hydrops fetalis or Haemoglobin Bart's hydrops fetalis.

There are serious implications when both parents carry alpha zero thalassaemia as this couple could have a child with no alpha genes. This severe condition is called **alpha thalassaemia major**. Alpha thalassaemia major is a condition causing a severe anaemia that affects the baby in the womb. The pregnancy may appear to progress normally up to about five months, sometimes for longer, however alpha thalassaemia major almost always leads to the baby not surviving during the pregnancy or after birth and may also lead to medical complications for the mother.

Useful contacts

Leicestershire Sickle Cell and Thalassaemia Service
Ground floor, Osborne Building
Leicester Royal Infirmary LE1 5WW
Telephone: 0116 258 6081

UK Thalassaemia Society
19 The Broadway
Southgate Circus
London N14 6PH
Telephone: 020 8882 0011
Email: info@ukts.org

Health information and support is available at www.nhs.uk or call 111 for non-emergency medical advice

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