What is Thalassaemia?

Thalassaemia is a group of blood disorders affecting haemoglobin production. Haemoglobin is a protein in red blood cells that carries oxygen around our bodies. Haemoglobin is made up of haem groups containing iron and globin chains. The main globin chains are call alpha (α) and beta (β). People with thalassaemia don't produce enough globin chains and therefore not enough haemoglobin, resulting in anaemia.

Thalassaemia is an inherited condition meaning it is passed from parent to child in genes. Genes carry information about human characteristics such as hair colour. Thalassaemia is **NOT** contagious and **NOT** transmitted by germs.

What is HbH disease?

Haemoglobin H disease (HbH) is a form of alpha thalassaemia, in which moderately to severe anaemia develops due to reduced formation of alpha globin chains. In this condition, as with the other forms of thalassaemia, there is an imbalance of globin chains needed to form haemoglobin. Normally, there are four genes to produce alpha globin chains. When three out of four of these genes become inactive, there are too few alpha globin chains to combine with beta chains and give rise to normal haemoglobin (haemoglobin A). The excess beta globin chains then combine with each other to form haemoglobin H, which is the origin of the name "haemoglobin H disease."

Treatment for HbH disease

Patients with Haemoglobin H disease may require blood transfusions to help manage their anaemia. These transfusion requirements may be exacerbated by pregnancy and other periods of stress.

HbH disease and Family Planning

The genes for α thalassaemia are common in some ethnic groups including people from Asia, Africa, the Middle East and Mediterranean.

For couples planning a pregnancy where HbH disease has been identified in one partner, then the other partner should be tested for thalassaemia as soon as possible.

Testing can be arranged by your local doctor.

By testing we can determine whether there is a risk of having a child with thalassaemia.

Where there is a risk of having an affected child, several options are available, and they should be referred by their clinician for further discussion with a Genetic Counsellor.

Important information for your family

If you have HbH disease, other members of your family should be tested as they may also be α thalassaemia carriers and at risk of having children with a severe thalassaemia. It is recommended that other family members and their partners are screened before having children of their own.

Chances of having a child affected with α thalassaemia:

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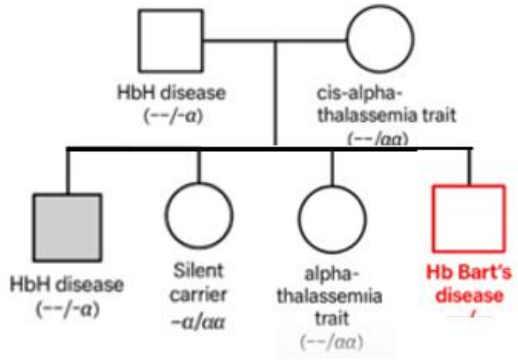
<td> 
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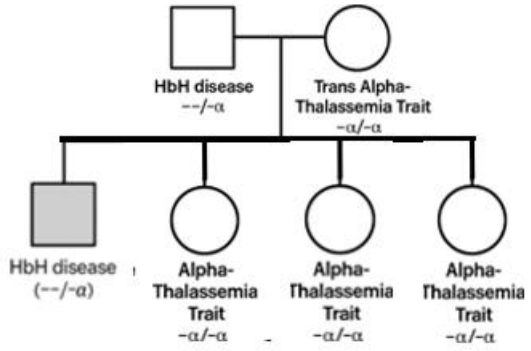
<td> 
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<td>With each pregnancy, this couple has a:
•1 in 4 chance of child with Hb Bart disease
•1 in 4 chance of child with HbH disease
•1 in 4 chance of child with alpha Thal trait
• 1 in 4 chance of child with silent alpha Thal carrier</td>
<td>With each pregnancy, this couple has a:
•1 in 4 chance of child with HbH disease
•3 in 4 chance of child with alpha Thal trait</td>
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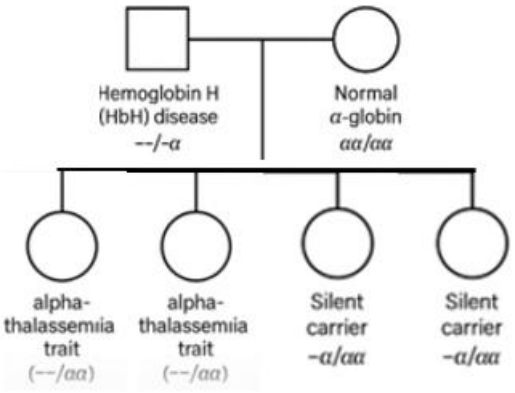
<td colspan="2"> 
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<tr>
<td colspan="2">With each pregnancy, this couple has a:
• 2 in 4 chance of child with alpha Thal trait
• 2 in 4 chance of child with silent alpha Thal carrier</td>
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References:

https://www.tasca.org.au

https://www.thalassemia.org/learn-about-thalassemia/about-thalassemia/#thalassemiabrochures

***For Medical Professionals seeking further information or advice, please contact Pathlab to discuss with one of our Haematologists:* 07 858 0795**