HOW IS α-THALASSAEMIA TREATED?

Treatment for α-thalassaemia is based on the type and severity of the disorder.

People who are ***carriers of α-thalassaemia*** usually have mild or no symptoms and do not require treatment. However, careful explanation must be provided to carriers concerning the possibility of passing on the trait to their children and, in the case of parents who are both α-thalassaemia carriers, the risk for developing HbH disease or α-thalassaemia major.

HbH disease can have a broad spectrum of severity. Many individuals with HbH disease may not require regular monitoring or treatment. However, some forms of HbH can be severe and require blood transfusions, regular follow-ups, and other medical interventions.

For many years, there were no effective treatments for ***α-thalassaemia major.*** Most pregnancies resulted in stillbirth or in severely ill newborns who passed away shortly after birth. However, recent medical advances have allowed physicians to attempt to improve treatment of the disorder. Such treatment includes administering in-utero (IUT) blood transfusions before birth to the foetus and immediately after birth. In that case, however, the child will require lifelong blood transfusions and special medical care.

Some foetuses with α-thalassaemia major have also been treated by hematopoietic stem cell transplantation (HSCT), using special cells found in the bone marrow. More treatments for this condition are currently under scientific development. Widespread application of such advances has until now been possible only in very few countries across the world.

For more information you can visit www.thalassaemia.org.cy

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FOCUS ON α-THALASSAEMIA

WHAT IS THALASSAEMIA?

Thalassaemia is inherited blood disorder, meaning that it is passed to a child from one or both parents through their genes. The body of an individual with thalassaemia produces less ***haemoglobin*** than normal, a protein in red blood cells enabling them to carry oxygen from the lungs throughout the whole body. This causes a shortage of functional red blood cells and low levels of oxygen supply to the tissues, which lead to a variety of health problems depending on the type and severity of the condition.

There are two main types of thalassaemia:

- **α-thalassaemia,** occurring when a gene or genes related to the α-globin protein production are missing or changed (mutated), and
- **β-thalassaemia,** occurring when similar gene defects affect production of the β-globin protein.

This leaflet is about **α-thalassaemia.**

WHAT ARE THE TYPES OF α-THALASSAEMIA?

Alpha (α-) globin chains are produced by four genes. One or more of those can be changed or missing altogether. Therefore, four kinds of α-thalassaemia can be described:

1. An individual who has only one (out of the four) abnormal or missing α-globin genes is called a ***silent alpha thalassaemia carrier,*** also known as an ***alpha plus (α+) thalassaemia carrier*** or as having the α-thalassaemia trait. Silent alpha thalassaemia carriers have no signs or symptoms of the disease, but are able to pass thalassaemia on to their children. Only very specific laboratory tests, based on DNA analysis, can accurately diagnose a silent carrier of α-thalassaemia.

2. An individual who has two (out of the four) abnormal or missing α-globin genes is an ***alpha zero (α⁰) thalassaemia carrier,*** also known as having ***α-thalassaemia minor*** or the ***α-thalassaemia trait.*** People with this condition may have red blood cells that are smaller than normal (microcytosis) and sometimes very mild anaemia. Other than those, they usually do not have any symptoms at all, but can pass on the trait to their children.

3. The condition whereby a child inherits only one functional α-globin gene and three (3) missing or abnormal ones from its parents is called ***haemoglobin H (HbH) disease***. This is an intermediate form of α-thalassaemia in terms of clinical severity characterized by increased haemolysis, thus destruction of red blood cells. Signs and symptoms can vary, ranging from moderate to severe anaemia, yellowing of the eyes and skin (jaundice), and enlargement of the spleen.

Consequently, individuals with HbH disease may require blood transfusions and a general check-up annually. These individuals must be looked after by physicians with knowledge and experience on monitoring and addressing complications related to the condition in expert haematology centres or clinics.

4. Four missing or abnormal α -globin genes lead to a condition known as ***Hydrops Fetalis, Hb Bart's syndrome*** or as ***α-thalassaemia major***.

This condition causes a severe anaemia that affects a foetus while still in the womb, before birth, often resulting to the foetus dying prematurely before delivery or to the newborn baby dying shortly after birth. Hb Bart's syndrome can also cause serious complications for women during pregnancy, including dangerously high blood pressure with swelling (preeclampsia), premature delivery, and abnormal bleeding. Therefore, early detection and prevention are essential in this situation.

WHO IS MOST AT RISK FOR α-THALASSAEMIA?

Alpha (α-) thalassaemia can affect individuals of any nationality and ethnicity. Despite being found in most populations worldwide, it is most common in the Middle East, Southeast Asia, and certain Mediterranean countries. Hb Bart's syndrome and HbH disease are primarily recognized in Southeast Asia.

The estimated severe prevalence of Hb Bart's syndrome in Southeast Asia is 1 in 200-2,000[1] births, whilst its prevalence in other parts of the world is unknown to date.

Increased immigration of people from areas with a higher prevalence of α- thalassaemia has led to a steady increase of the alpha-globin disorders in the United States and other Western nations, such as countries of Northern Europe. However, studies have shown that α-thalassaemia may still remain under-recognized and underdiagnosed in these countries.

[1]ORPHAcode: 163596
Source: www.orpha.net/