

3133 2

WHO/HDP/EMT/90.2

ORIGINAL: ENGLISH

Distr.: LIMITED

EDUCATIONAL MATERIALS ON THALASSAEMIA

Thalassaemia is not only a disease of *"just a few people"*. You may carry this disease, only you don't know it.

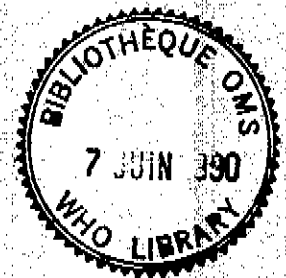
Thalassaemia is a serious illness that is handed on to children by their parents who do not know that they are *"healthy carriers"*.

Thalassaemia can be prevented if information, scientific research and proper medical services can be made available.



WORLD HEALTH ORGANIZATION
HEREDITARY DISEASES PROGRAMME

1990



WHAT YOU NEED TO KNOW ABOUT ALPHA THALASSAEMIA TRAIT

Dear Reader,

There are several kinds of thalassaemia. This booklet is for people who have had a blood test that shows they carry alpha thalassaemia trait (this is usually written α thalassaemia trait).

α thalassaemia trait is not an illness, and will not affect your health.

It is not the same as beta thalassaemia trait (written as β thalassaemia trait).

There are two kinds of α thalassaemia trait:

(a) Alpha-plus (α^+) thalassaemia trait is very common, and is almost always harmless.

(b) Alpha-zero (α^0) thalassaemia trait is uncommon, and could be a problem for your children. So if you have α thalassaemia trait it is important to know which type.

This booklet gives information about both types.

Do not forget that you carry α -thalassaemia trait. Keep the blood test report or thalassaemia card permanently with your medical record.

If you want more information after you have read this booklet, ask your doctor to arrange a visit to a genetic counsellor.

Take this booklet with you if you go to see your doctor about your α -thalassaemia trait.

WHAT IS "THALASSAEMIA"?

Thalassaemia is a peculiarity of the blood that is common among people originating from the Mediterranean area, the Middle East, or Asia. It is rare in North Europeans.

There are two main forms of thalassaemia: alpha thalassaemia and beta thalassaemia (α thalassaemia and β thalassaemia).

When people talk about thalassaemia, they usually mean β thalassaemia, because it causes problems more often than α thalassaemia. You can obtain a separate booklet about β thalassaemia - "Educational Materials on Thalassaemia" (WHO/HDP/EMT/90.1), available free of charge in English (Arabic, Chinese, French, Russian and Spanish translations will be available in autumn 1990) from the Hereditary Diseases Programme, Division of Noncommunicable Diseases and Health Technology, World Health Organization, 1211 Geneva 27, Switzerland.

When a person carries α thalassaemia, they are said to carry α thalassaemia trait.

There are two types of α thalassaemia trait:

(a) Alpha plus (α^+) thalassaemia trait is extremely common, and nearly always completely harmless.

It is carried by about

- one third of people originating from Africa.
- half the people of India and Pakistan.
- many people from the Mediterranean area, particularly Cyprus, Sardinia, Greece or South Italy.
- many people from the Middle East.

(b) Alpha zero (α^0) thalassaemia trait is quite uncommon.

It does no harm to the people who carry it, but it could affect the health of their children.

It is carried by about

- one in thirty people originating from south-east Asia (south China, Hong Kong, Singapore and Thailand).
- one in a hundred people originating from Cyprus or parts of Greece.

One of the problems with α thalassaemia is that it can be quite difficult to distinguish α^+ and α^0 thalassaemia trait.

HOW CAN I BE SURE IF I HAVE α^0 OR α^+ THALASSAEMIA?

If you, or your ancestors, come from Africa, India or Pakistan, you will have α^+ thalassaemia. You are extremely unlikely to have α^0 thalassaemia, and you have nothing to worry about.

But if you have α thalassaemia and you or your ancestors come from Cyprus, Greece, the Middle East, south-east Asia (Thailand, Vietnam, Kampuchea, Laos), South China or Singapore, you could have α^0 thalassaemia trait. This would not do you any harm, but it could affect your children. You may be advised to bring your partner for a test before you have children. If your partner does not have any type of α thalassaemia, there will be no risk for your children, and you have nothing to worry about. But if your partner's blood test result shows any peculiarity, you should see an expert in haemoglobin disorders for further testing, and advice.

If you are in any doubt about the type of α thalassaemia you carry and you need to find out, go to see your doctor, and take this booklet with you.

BLOOD AND ANAEMIA

To explain about thalassaemia, we need to talk a little about normal blood and about anaemia.

What is blood made of?

Blood is made up of a lot of red blood cells in a clear, slightly yellow liquid called plasma. Blood is red because the red blood cells contain a substance called haemoglobin. Haemoglobin is very important because it carries oxygen from your lungs to wherever it is needed in your body. It contains a lot of iron. In fact, the main reason why people need iron in their food is to make haemoglobin.

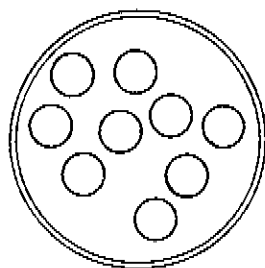
What is anaemia?

Some people have too little haemoglobin in their blood. These people have anaemia. There are many different kinds of anaemia. The most common kind is iron deficiency anaemia. This happens when people are not eating enough of the foods that contain iron. Some people who carry thalassaemia have a very mild anaemia, but it has nothing to do with the amount of iron you are getting from your food. It is inherited.

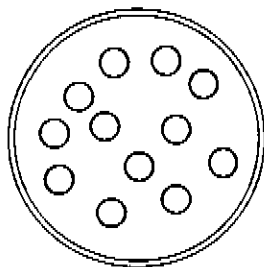
WHAT DOES ALPHA THALASSAEMIA TRAIT MEAN FOR ME?

People with α thalassaemia trait are perfectly healthy: only a few have a slight anaemia. That is why most people with α thalassaemia trait do not know they have it. They only discover it through having a special blood test.

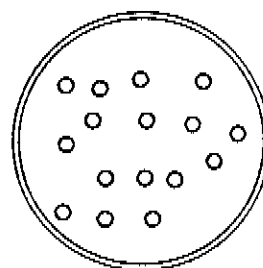
The red blood cells of people with α thalassaemia trait are smaller than the usual kind of red cells. In α^+ thalassaemia the red-cells are about three-quarters the usual size, in α^0 thalassaemia they are about two-thirds the usual size.



Usual red cells



α^+ thalassaemia red cells



α^0 thalassaemia red cells

α thalassaemia trait is present in the fetus before birth, it remains the same throughout life, and can be handed from parents to children. That is, it is inherited.

WHY DO YOU NEED TO KNOW IF YOU CARRY α THALASSAEMIA TRAIT?

Sometimes people with α^0 thalassaemia trait can have babies born with a very severe anaemia. If you have α^0 thalassaemia trait it is important to know about this risk.

A very few people with α^+ thalassaemia trait can have children with a milder anaemia called Hb H disease. If you have α^+ thalassaemia trait, you have a very small risk of having children with anaemia.

IS A THALASSAEMIA CARRIER ILL?

No, so there is no need for any medical treatment.

ARE THERE ANY OTHER PROBLEMS?

No. Thalassaemia carriers are not more likely to get any other illnesses, nor are they weak in any way, or limited in their choice of job.

CAN ANY TREATMENT CHANGE α THALASSAEMIA TRAIT?

No. If you are born with thalassaemia trait, you will always have it.

CAN α THALASSAEMIA TRAIT TURN INTO A SEVERE FORM OF THALASSAEMIA?

No. It cannot.

DO α THALASSAEMIA CARRIERS EVER NEED IRON?

Yes, they sometimes do, but it is important that you only have iron medicine if you really need it. The best way to be sure a thalassaemia carrier needs iron is by a blood test

to measure the amount of iron in your blood. If you do not have this test, the doctor may think that you are short of iron simply because you have small red blood cells and a slight anaemia, and may advise you to keep taking extra iron even when you do not need it. This will do you no good, and in the long-run it could be harmful.

WHAT ABOUT PREGNANT WOMEN?

Pregnant women with thalassaemia trait need extra iron just as much as other pregnant women.

WHY IS α THALASSAEMIA TRAIT FOUND IN CERTAIN COUNTRIES?

People with α thalassaemia trait are less likely to die if they catch malaria. In the past, in countries where malaria was common, α thalassaemia trait was an important advantage because people with α thalassaemia trait survived malaria where other people died of it. These people passed the trait on to their children, so as time passed, it became more common in malarial parts of the world. But now we can usually cure or prevent malaria and thalassaemia trait is no longer an advantage. As it is inherited, it does not go away from a population when malaria disappears.

In every country where malaria is or was common a large number of people have α thalassaemia trait.

OTHER FORMS OF THALASSAEMIA TRAIT

This booklet is about α thalassaemia trait. It is important not to get it mixed up with other forms of thalassaemia.

Beta (β) thalassaemia trait is common in many of the places where α thalassaemia trait occurs. It has a similar effect on the people who carry it, but it causes rather more risk for their children. It is described in a separate booklet "Everything you need to know about thalassaemia trait".¹

Delta-beta- ($\delta\beta$)-thalassaemia trait and haemoglobin Lepore trait are both forms of β thalassaemia trait.

There are also four main types of abnormal haemoglobins. These are:

- HbS
- HbC
- HbD
- HbE

If someone has α thalassaemia trait and chooses a partner who has β thalassaemia trait, $\delta\beta$ thalassaemia trait, haemoglobin Lepore trait, or haemoglobin S, C, D or E, there is no risk that their children could have a severe anaemia. This problem can only ever arise if one α thalassaemia carrier chooses another α thalassaemia carrier as a partner. Even then, problems are not very common.

¹ See WHO information booklet on educational materials on thalassaemia, WHO unpublished document WHO/HDP/EMT/90.1, pages 2-9. Available free of charge in English (Arabic, Chinese, French, Russian and Spanish translations will be available in the autumn of 1990) from the Hereditary Diseases Programme, Division of Noncommunicable Diseases and Health Technology, World Health Organization, 1211 Geneva 27, Switzerland.

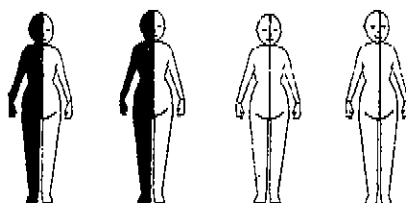
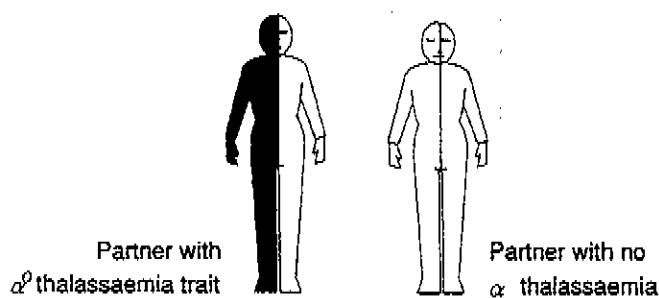
WHAT COULD MY α THALASSAEMIA MEAN FOR MY CHILDREN?

The risk is quite different for carriers of α^+ and α^0 thalassaemia trait. Most problems can arise for carriers of α^0 thalassaemia trait, so we will discuss this first.

WHAT ARE THE POSSIBLE RISKS FOR CARRIERS OF α^0 THALASSAEMIA TRAIT?

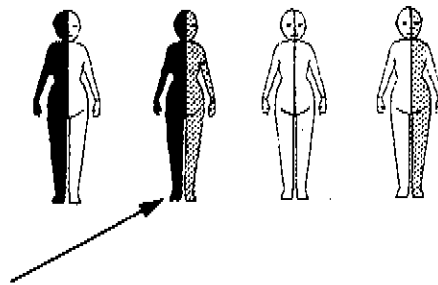
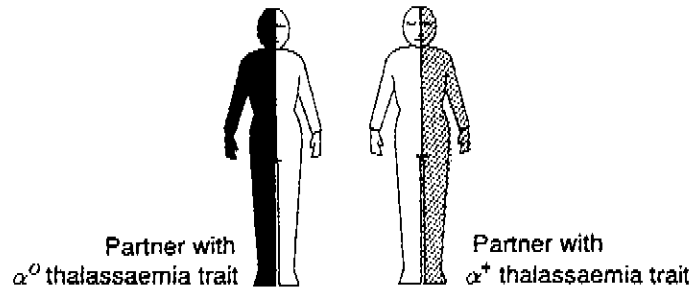
To answer this question, we must see how α thalassaemia is passed on from parents to their children. Let us consider three sorts of couples.

1. If a carrier of α^0 thalassaemia trait chooses a partner who carries no α thalassaemia at all, on average half the children will carry α^0 thalassaemia trait and half will have the usual type of blood. None of them will be ill with an important α thalassaemia. There is no risk for an α^0 thalassaemia carrier whose partner is not a carrier.



Half the children will carry α^0 thalassaemia, and half will not. All are healthy.

2. Sometimes a person with α^0 thalassaemia trait chooses a partner with α^+ thalassaemia trait. Most of their children will be completely healthy (half will carry α^+ thalassaemia and a quarter will not carry any type of thalassaemia). But a quarter (25%) will inherit α^0 thalassaemia from one parent and α^+ thalassaemia from the other. This leads to a type of anaemia called "haemoglobin H disease".



One out of 4 children (on average) may inherit α^0 thalassaemia from one parent and α^+ thalassaemia from the other. This child will have haemoglobin H disease. All the others are healthy.

What is haemoglobin H disease?

Children with HbH disease are anaemic: they have a haemoglobin level of 8-9 grams per decilitre (8-9 g/dl). The normal level is about 11-14 grams per decilitre (11-14 g/dl). So their haemoglobin level is lower than normal.

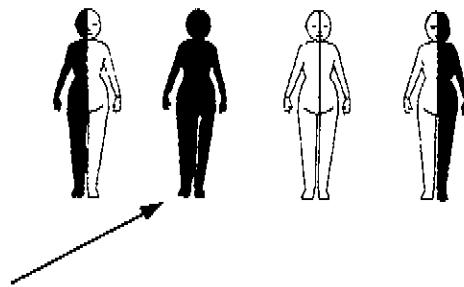
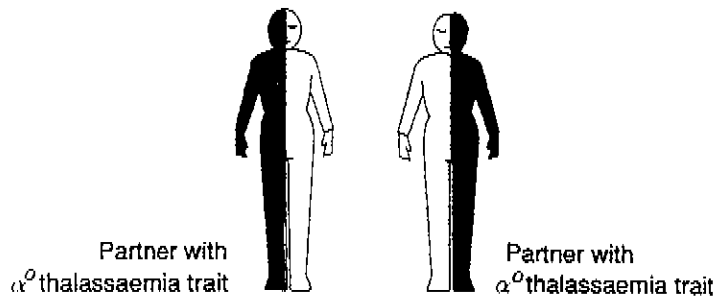
But people with haemoglobin H disease are usually quite well, and can work and have children like other people.

Once couples who could have children with HbH disease understand the situation, they are not really worried.

They usually ask to have the baby tested as soon as it is born, so that they can know the situation.

If the baby does have haemoglobin H disease, the parents are advised to attend a regular paediatric clinic a few times a year, just to check that the baby is developing well, and to make sure that there are no problems.

3. If by chance (rarely) a carrier of α thalassaemia trait chooses a partner who also carries α^0 thalassaemia trait, most of their children will be healthy. (They may carry α^0 thalassaemia trait, or they may have completely normal blood). But a quarter (25%) will inherit α^0 thalassaemia from both parents. They will have α thalassaemia major.



One out of 4 children (on average) may inherit α^0 thalassaemia from both parents.
This fetus will be affected by α^0 thalassaemia hydrops fetalis.
All the other children will be healthy.

In each pregnancy there is a one in four (25%) chance that the child will have normal blood and two in four (50%) chance that the child will have α^0 thalassaemia trait. There is a 1 in 4 (25%) chance that the fetus will have α^0 thalassaemia major.

WHAT IS α^0 THALASSAEMIA MAJOR?

Another name for α^0 thalassaemia major is α thalassaemia hydrops fetalis.

This is a very serious anaemia that develops in the fetus. It can only happen when both parents carry α^0 thalassaemia trait.

The fetus cannot make enough haemoglobin, because its bone marrow cannot produce enough red blood cells. The red blood cells that are produced are nearly empty. As a result, the fetus becomes very anaemic and weak and its heart is not able to pump blood around properly.

The pregnancy seems to go normally up to about five months, sometimes for longer, but then the baby stops growing normally, and the mother may develop high blood pressure. An ultrasound examination may be done. This usually shows that the baby is "oedematous" - which means that it is puffed up, with too much water in it.

Usually the mother starts labour early, between 28 and 36 weeks of pregnancy, and the baby is dead or dying when it is delivered.

There is one in four (25%) chance of the same thing happening in any further pregnancies, so this is one condition that people are very eager to avoid.

This is why it is so important for people to know if they have α^0 thalassaemia trait, and whether their partner also carries it, before they decide to have a family.

CAN α^0 THALASSAEMIA MAJOR BE TREATED?

There is no treatment for α^0 thalassaemia major.

CAN α^0 THALASSAEMIA MAJOR BE PREVENTED?

When both partners carry α^0 thalassaemia trait, there are several ways to avoid having a stillborn baby. It is possible to tell very early on indeed in a pregnancy whether the fetus will be healthy, or suffers from α^0 thalassaemia major. Most couples who both carry α^0 thalassaemia trait ask the doctors to test each pregnancy to find out if the baby has thalassaemia major. This test can be done any time after 8 weeks after the last period. When the fetus is affected, it has no hope of a normal life, so parents usually wish to have the pregnancy terminated. Then they start again with another pregnancy, hoping to have a healthy child next time. Remember, there is a three quarters (75%) chance of a healthy child in each pregnancy!

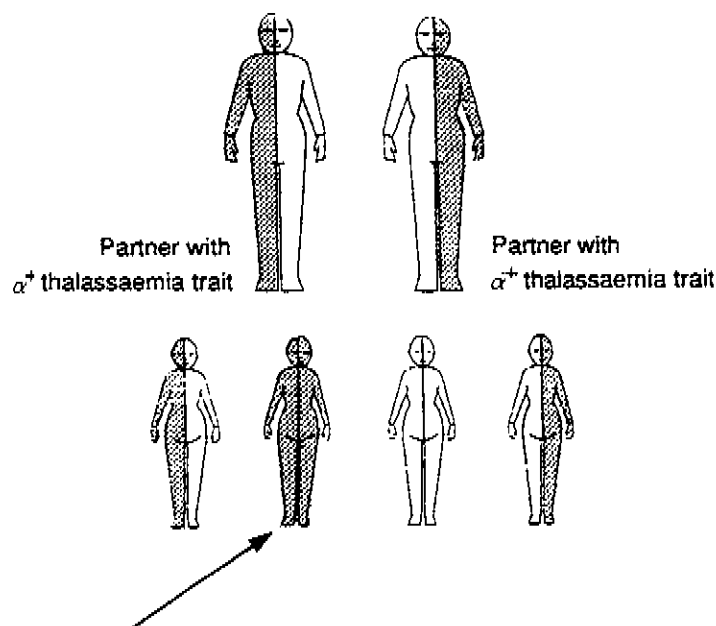
There are several other ways to avoid having children with α thalassaemia major. To find out more, ask your doctor to arrange for you to visit a genetic counsellor.

RISKS FOR CARRIERS OF α^+ THALASSAEMIA TRAIT

The most important risk for carriers of α^+ thalassaemia trait is the risk of a mistake. They could be told they carry α thalassaemia, and then people might think it is the severe form of α^0 thalassaemia.

There is a very small risk indeed that a carrier of α^+ thalassaemia trait will have children with anaemia. Let us consider several situations.

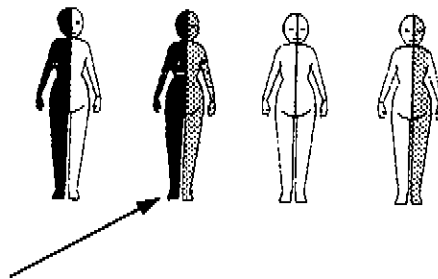
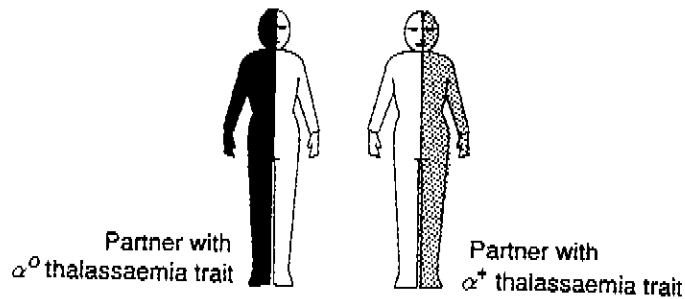
1. A carrier of α^+ thalassaemia chooses a partner who does not carry any form of thalassaemia. On average half the children carry α^+ thalassaemia and half will not, and none should suffer from a severe inherited anaemia.
2. A carrier of α^+ thalassaemia chooses a partner who also carries α^+ thalassaemia. In this case, a quarter (25%) of the children will inherit α^+ thalassaemia from both parents. However, α^+ thalassaemia is so mild, this simply leads to slightly smaller red blood cells, and the person is still perfectly healthy.



One out of 4 children (on average) may inherit α^+ thalassaemia from both parents. This child will be perfectly healthy. All the other children will be healthy also.

3. Rarely a person with α^+ thalassaemia trait chooses a partner who has α^0 thalassaemia trait. Most of their children will be completely healthy (half will carry α^+ thalassaemia and a quarter will not carry any type of thalassaemia). But a quarter (25%) will inherit α^0 thalassaemia from one parent and α^+ thalassaemia from the other. This leads to a type of anaemia called "haemoglobin H disease" (see pages 51-53).

So in conclusion, most people who carry α^+ thalassaemia trait have no need to worry. They should think of themselves as normal in every way.



One out of 4 children (on average) may inherit α^0 thalassaemia from one parent and α^+ thalassaemia from the other. This child will have **haemoglobin H disease**. All the others are healthy.

* * * * *