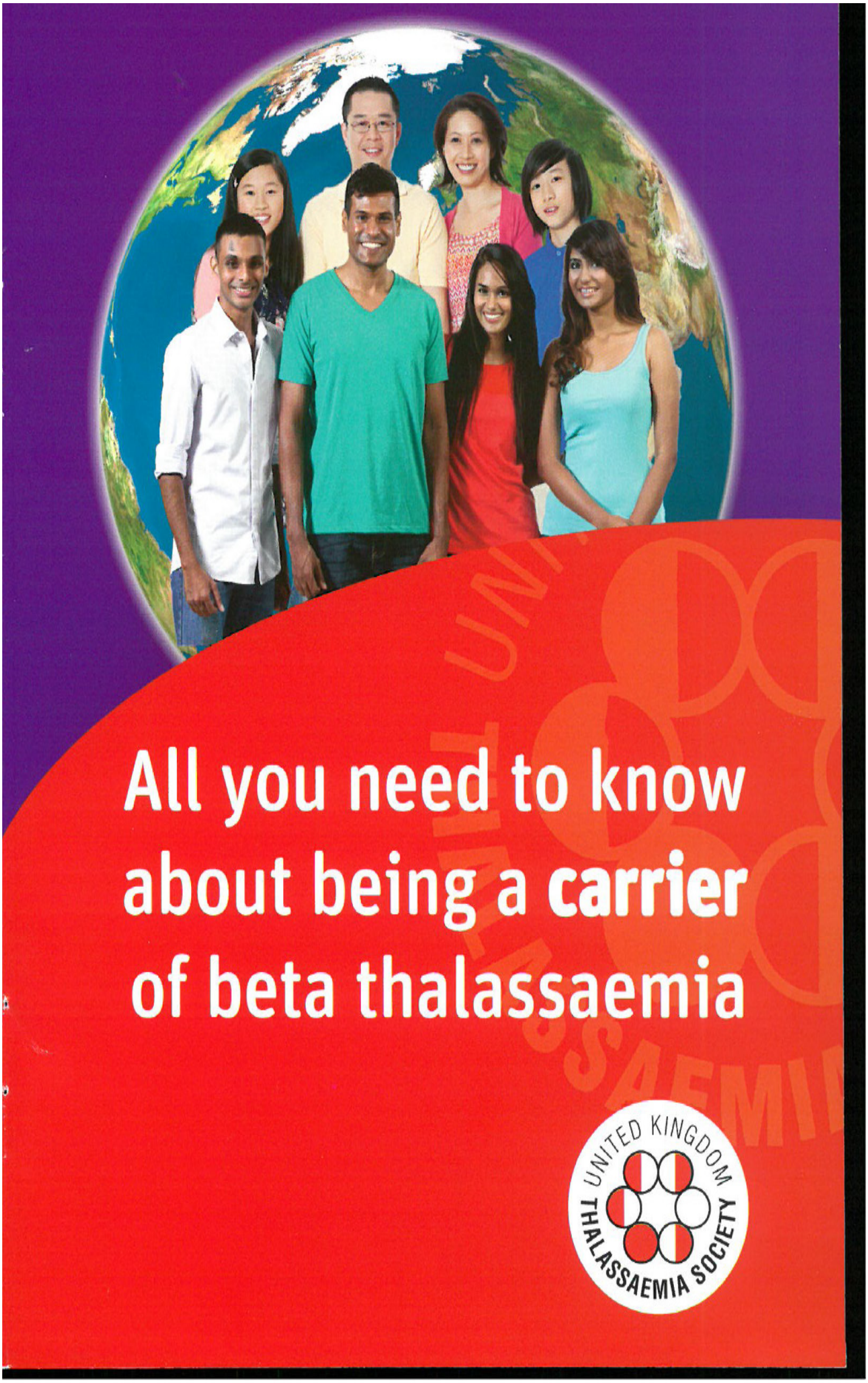

All you need to know about being a **carrier** of beta thalassaemia

UNITED KINGDOM THALASSAEMIA SOCIETY

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amendments by Dr Mary Petrou)

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There are two forms of thalassaemia: alpha thalassaemia and beta thalassaemia.

This is a booklet for people who have had a blood test that shows they carry **BETA THALASSAEMIA**. If you have picked the booklet up by chance, please read it anyway. You might find that you should have a blood test to see if you carry thalassaemia yourself.

Note:

Carrying beta thalassaemia is often called "having beta thalassaemia trait" or "thalassaemia trait" for short. In order to be completely clear, we talk about "carrying beta thalassaemia" throughout this booklet.

The United Kingdom Thalassaemia Society was formed by the parents of thalassaemia patients in Britain. The aim of the Society is to help affected families keep in touch with each other, to raise funds to support research, to improve treatment facilities, to encourage blood testing, counselling and screening programmes. If you would like to give a donation or if you want to find out more about thalassaemia please contact the Society.

Dear Reader,

You have been given this booklet because you have been told that you carry beta thalassaemia or because you are interested in it. This booklet will give you a lot of information about beta thalassaemia, but these are the points that matter most:

1. Carrying beta thalassaemia is not an illness and does not affect your own health. However, it could affect the health of your future children.

2. Do not forget that you carry beta thalassaemia. Keep this booklet and your blood test results, if you have them, among your personal documents.

3. There is nothing bad about carrying beta thalassaemia. There is no need to feel embarrassed or ashamed about it. Instead, talk about it with your partner and family and if they haven't already had a blood test, persuade them to go for one.

4. If your partner (present or future) does not carry beta thalassaemia, or an unusual haemoglobin, there is no problem. However, your children may be carriers like yourself. They should have their blood tested at some time before they have children of their own.

5. If both you and your partner carry beta thalassaemia, there is a risk for your future children, but you can avoid it by planning your family carefully with help from the doctors.

* If you want more information after you have read this booklet, ask your doctor to arrange a visit to a specialist Sickle Cell and Thalassaemia Counsellor or a Genetic Counsellor.

Take this booklet with you if you go to see your doctor about carrying beta thalassaemia.

Other forms of thalassaemia carriers

This booklet is all about carrying **beta thalassaemia**, but there are also other groups of thalassaemia:

Delta-beta thalassaemia and **Haemoglobin Lepore** are very similar to beta thalassaemia. If you carry either of these, all the information in this booklet applies to you.

Alpha thalassaemia is very different from beta thalassaemia. It only rarely causes any illness in children. This booklet does not apply to alpha thalassaemia carriers. A separate booklet for people who carry alpha thalassaemia is available from our Society.

In addition to the thalassaemias there are four important forms of unusual haemoglobins. These are:

- Haemoglobin S
- Haemoglobin C
- Haemoglobin E
- Haemoglobin O Arab

If someone who carries beta thalassaemia chooses a partner who carries one of these unusual haemoglobins, there is a risk that some of their children could have a serious blood disorder, like thalassaemia major (see page 4).

What is beta thalassaemia?

Beta thalassaemia is a condition of the blood that is found in many countries around the world, and particularly in people of Mediterranean, Middle Eastern or Asian origin. It is rare in Northern Europeans.

There are two forms of beta thalassaemia:

1. **Carrying beta thalassaemia.** People who carry beta thalassaemia are perfectly healthy themselves but if their partner also carries beta thalassaemia, there is a chance that some of their children could have beta thalassaemia major. There are about 200,000 people who carry beta thalassaemia in Britain. They are sometimes called "healthy carriers of thalassaemia".

2. **Beta thalassaemia major.** This is a very serious blood disorder that children are born with. It begins to show in early childhood. Children who have beta thalassaemia major cannot make enough haemoglobin in their blood. They need frequent blood transfusions and other medical treatment.

Every year at least 75,000 children worldwide are born with beta thalassaemia major. In Britain there are about 1000 people with the disorder.

Beta thalassaemia major is sometimes called **"Mediterranean anaemia"**, **"Cooley's Anemia"** or **"homozygous beta thalassaemia"**.

Blood and anaemia

To understand more about beta thalassaemia, you need to know 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. Each red blood cell only lives for about 4 months. It is then broken down. New red blood cells are being made all the time. Blood cells are replaced very quickly – that's why people can give blood often.

Blood is red because the red cells contain a substance called haemoglobin. Haemoglobin is very important because it carries oxygen from your lungs to wherever it is needed in the body.

Haemoglobin contains a lot of iron. When your red blood cells are broken down, most of the iron from the haemoglobin is used again to make new haemoglobin. You lose some iron from your body every day and you make up for it with the iron in the food you eat. 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 do not have enough haemoglobin because their diet contains too little iron.

Beta thalassaemia major is a different kind of anaemia. It is caused by not having enough haemoglobin, but it has nothing to do with the amount of iron you're getting from your food. It is an inherited blood disorder.

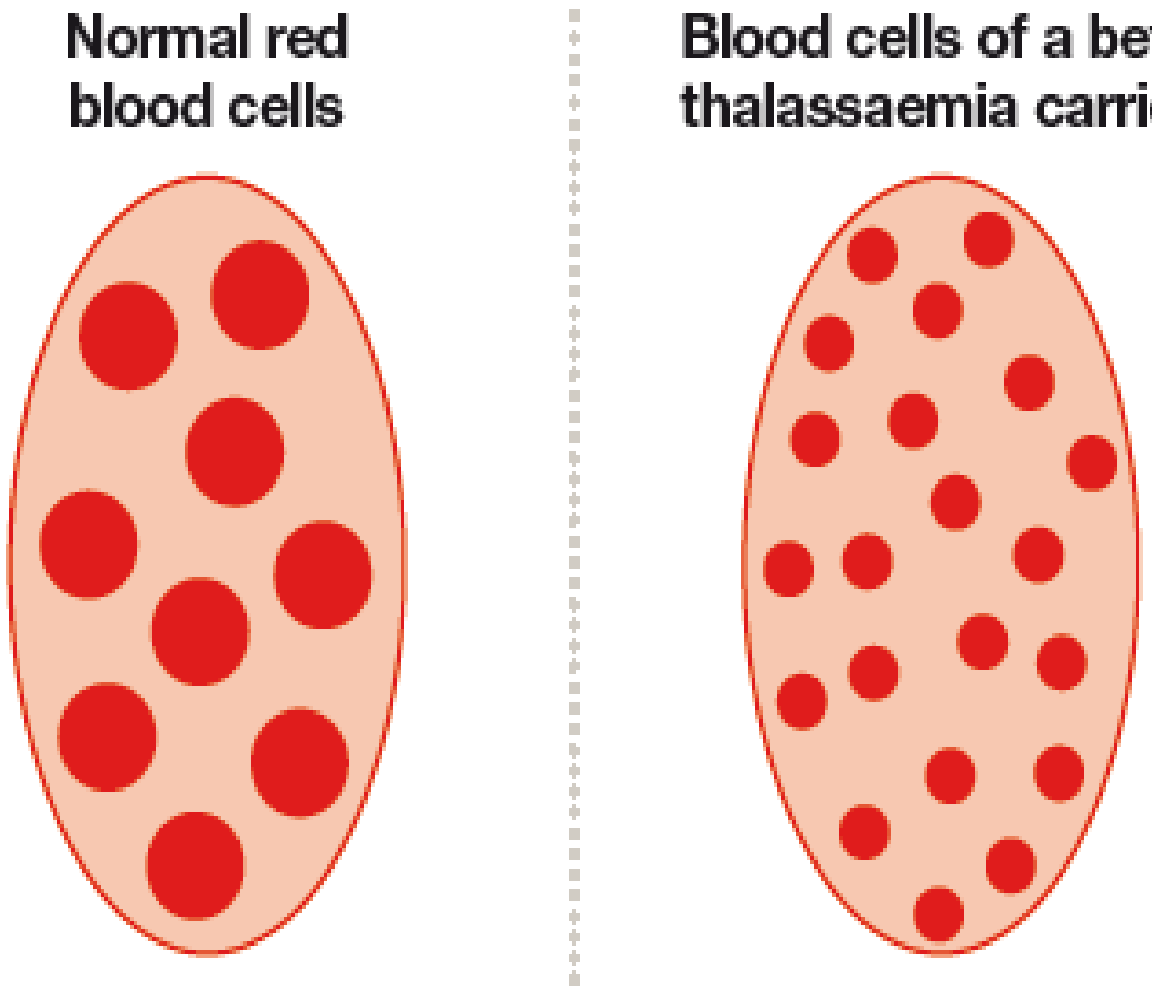
Carrying beta thalassaemia

What is it?

People who carry beta thalassaemia are not ill. They are absolutely healthy and normal but some of them have slight anaemia.

Most people who carry beta thalassaemia do not know that they have it. You only discover it if you have a special blood test, or if you have a child with beta thalassaemia major.

People who carry beta thalassaemia have smaller red cells than usual. They also have slightly more of a kind of haemoglobin called haemoglobin A2 in their blood.



The fact that people who carry beta thalassaemia have unusually small red blood cells does not matter. However, it sometimes causes confusion if a doctor thinks that the small red cells are due to iron deficiency.

Beta thalassaemia is present at birth. It remains the same for life and it can be handed on from parents to children. That means it is inherited.

Why does it matter if you carry beta thalassaemia?

Sometimes people who carry beta thalassaemia can have children with beta thalassaemia major, a serious blood disorder.

How do you find out if you carry beta thalassaemia?

You have to have a special blood test called a "haemoglobinopathy screen". The test measures the size of your red blood cells and how much haemoglobin A2 you have in your blood. **N.B.** Do not assume that beta thalassaemia would have "shown up" if you previously had a blood test for some other reason. The only way to be sure whether or not you are a carrier of beta thalassaemia is to have a haemoglobinopathy screen which tests specifically for thalassaemia.

Is a beta thalassaemia carrier ill?

No. So there is no need for any special medical treatment.

Is a beta thalassaemia carrier more likely to get other illnesses?

No.

Is beta thalassaemia infectious?

No. It is inherited; that means it can only be passed on from parents to their children.

Is a beta thalassaemia carrier physically or mentally weak?

No.

Does carrying beta thalassaemia affect the sort of work you can do?

No.

Can any treatment change being a carrier of beta thalassaemia?

No. If you are born being a carrier of beta thalassaemia you will always be one.

- ***Can carrying beta thalassaemia turn into beta thalassaemia major?***

  No.

- ***Can a beta thalassaemia carrier donate blood?***

  Yes.

- ***Do beta thalassaemia carriers ever need iron medicine?***

  Yes, they sometimes do, but not because they are carriers of beta thalassaemia. It is important that you only have iron medicine if you really do need it.

  The best way to tell whether a beta thalassaemia carrier needs iron is by a special blood test called a "ferritin test" that measures the amount of iron in your blood. If you don't have this test, the doctor may think that you are short of iron simply because you have small red cells and slight anaemia, and may advise you to keep taking extra iron when you really 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 who carry beta thalassaemia need extra iron just as much as any other pregnant woman.

- ***Why have we never heard of beta thalassaemia before?***

  Because tests for beta thalassaemia have only been widely offered recently. Any carriers of beta thalassaemia in your family would not have known about it, as special tests for beta thalassaemia may not have been available to them. Now everybody can have a test to make sure.

Why is beta thalassaemia mainly found in people from certain countries?

People who carry beta thalassaemia are less likely to die if they catch malaria, so carrying beta thalassaemia can be a great advantage. In the past, in countries where malaria was very common, carriers of beta thalassaemia often survived malaria when other people died. They passed thalassaemia on to their children, so as time passed it became more common in countries where malaria was common. Now we can usually cure or prevent malaria, but beta thalassaemia does not go away when malaria disappears. **N.B.** This does not mean that beta thalassaemia carriers are protected against malaria. They need anti-malarial drugs just as other people do when visiting parts of the world affected by malaria.

Does carrying beta thalassaemia have any other advantage?

Yes. It seems that people who carry beta thalassaemia are less likely than other people to suffer from heart attacks when they get older.

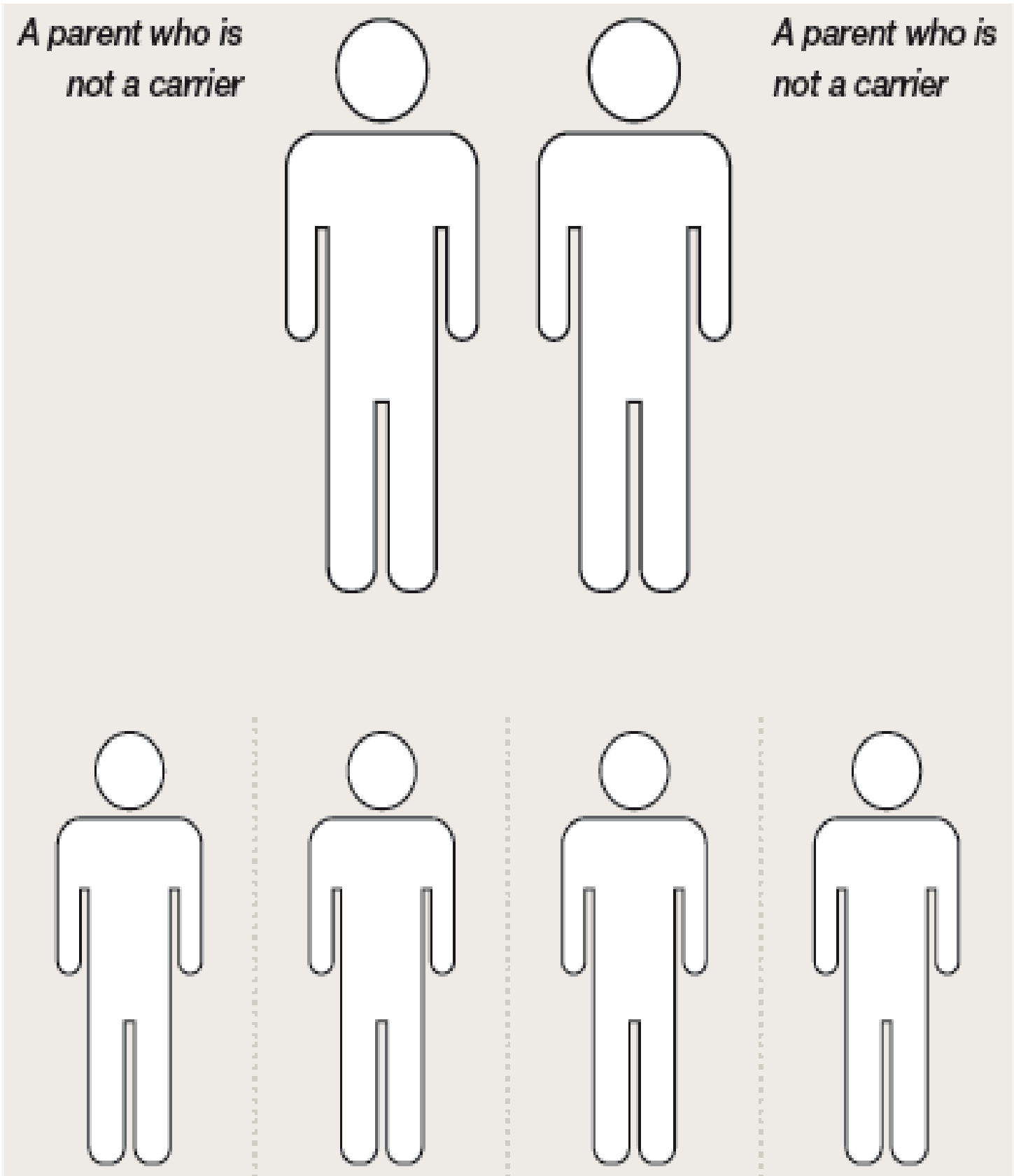
How common is carrying beta thalassaemia?

Very many countries used to have malaria; and all these countries now have quite a large number of people carrying beta thalassaemia. For instance, in Cyprus one in seven people are carriers of beta thalassaemia, in Greece one in twelve, in Italy and all of the Middle East and Asia, including India, Pakistan, Southern China, Hong Kong and Vietnam, the number of people carrying beta thalassaemia varies from one in fifty to one in ten in different areas. In West Africa and the West Indies about one in fifty people carry beta thalassaemia. About one in every thousand people of Northern European origin carries it.

How is beta thalassaemia passed on from parents to their children?

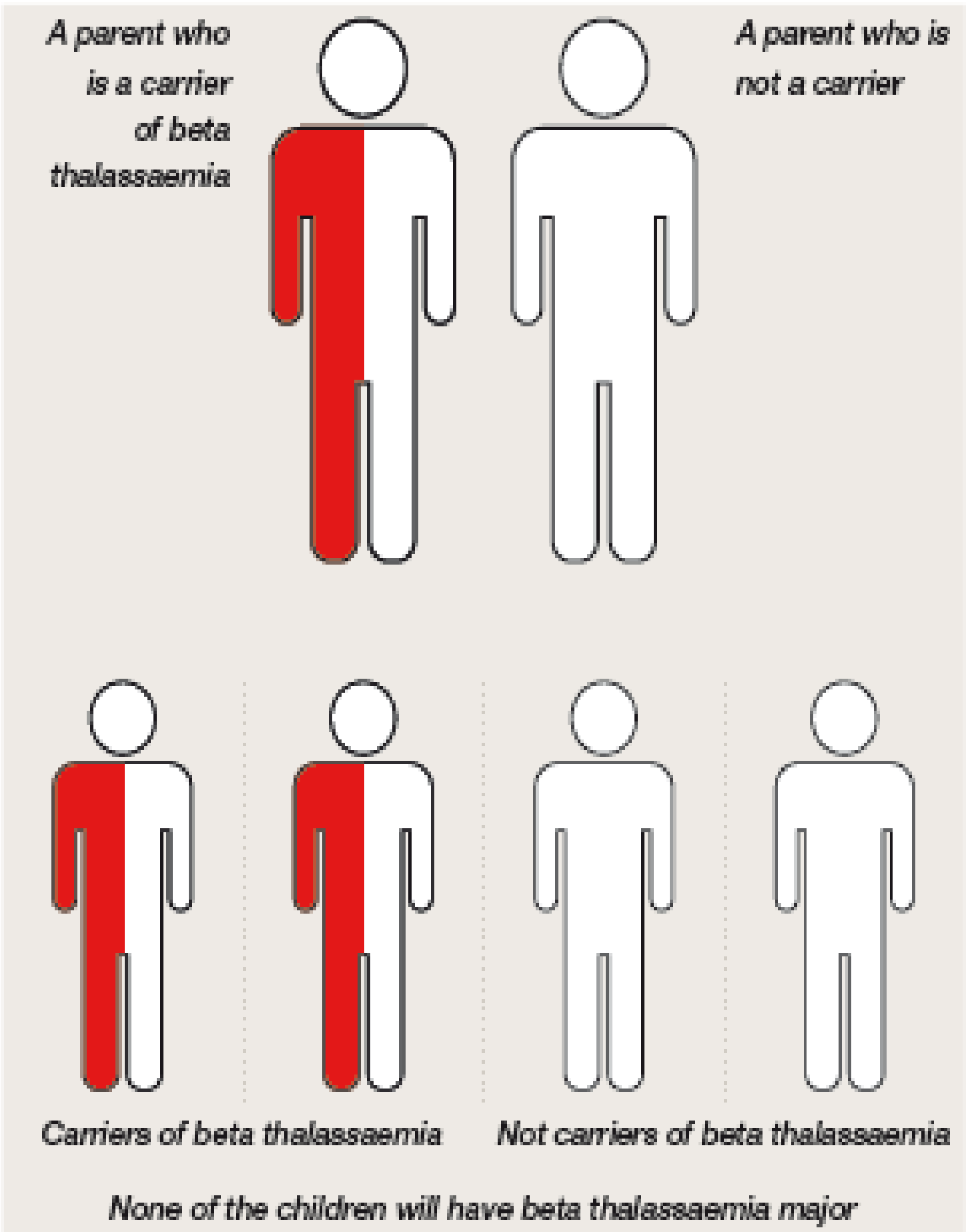
Let us consider three sorts of couples.

1. If both parents are not carriers, they cannot possibly pass on beta thalassaemia to their children.



None of the children will carry beta thalassaemia or have beta thalassaemia major.

2. If one parent is a carrier of beta thalassaemia and one is not a carrier there is a one in two (50%) chance that each of their children will carry beta thalassaemia. None of their children can have beta thalassaemia major.

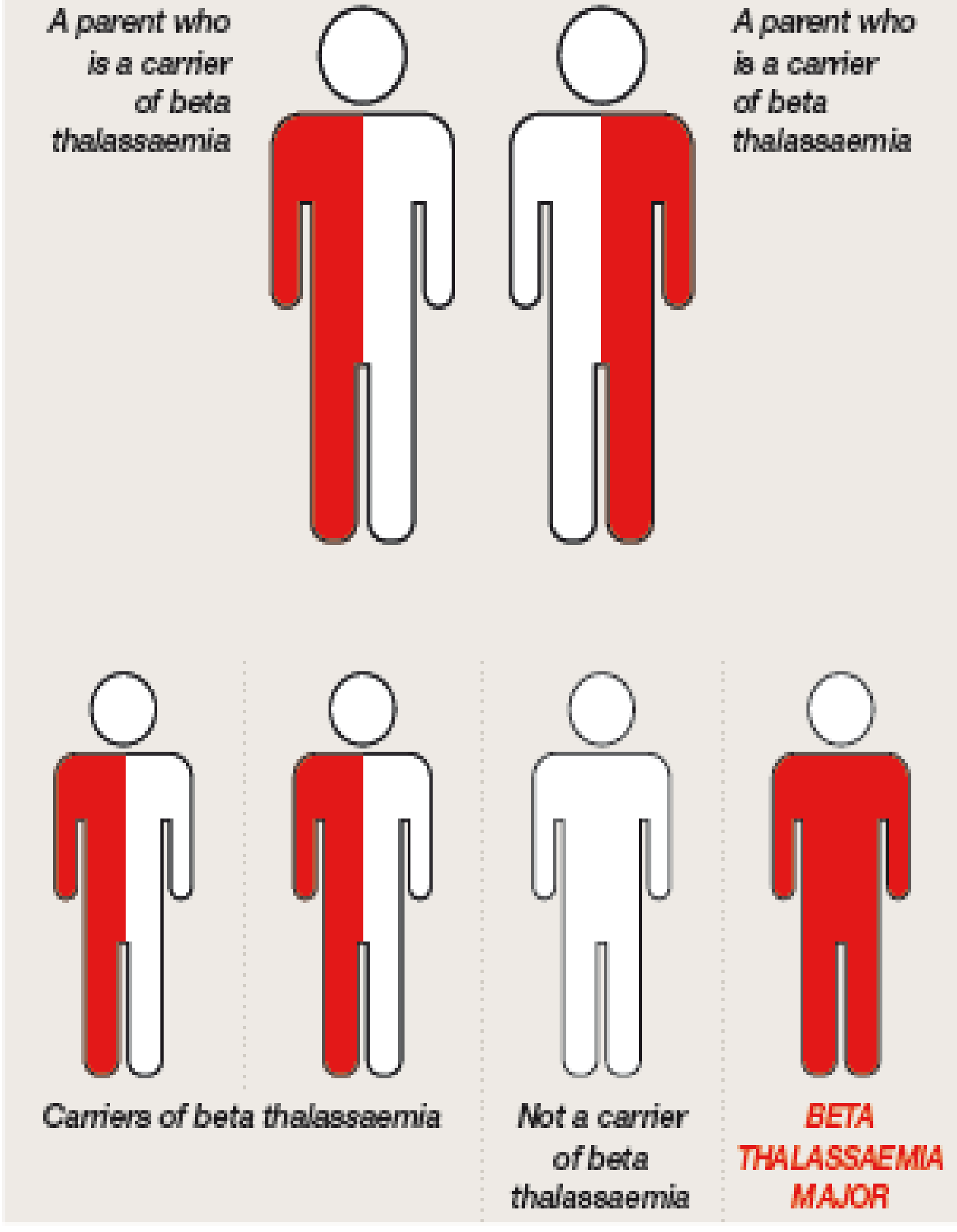


Beta thalassaemia can be passed on through many generations without anybody realising that it is "in the family". If you are a carrier, this is probably the situation in your family. One of your parents must also be a carrier. This means that your brothers and sisters also have a 1 in 2 (50%) chance of being a carrier.

3. **If both parents carry beta thalassaemia then the situation is more complicated.**

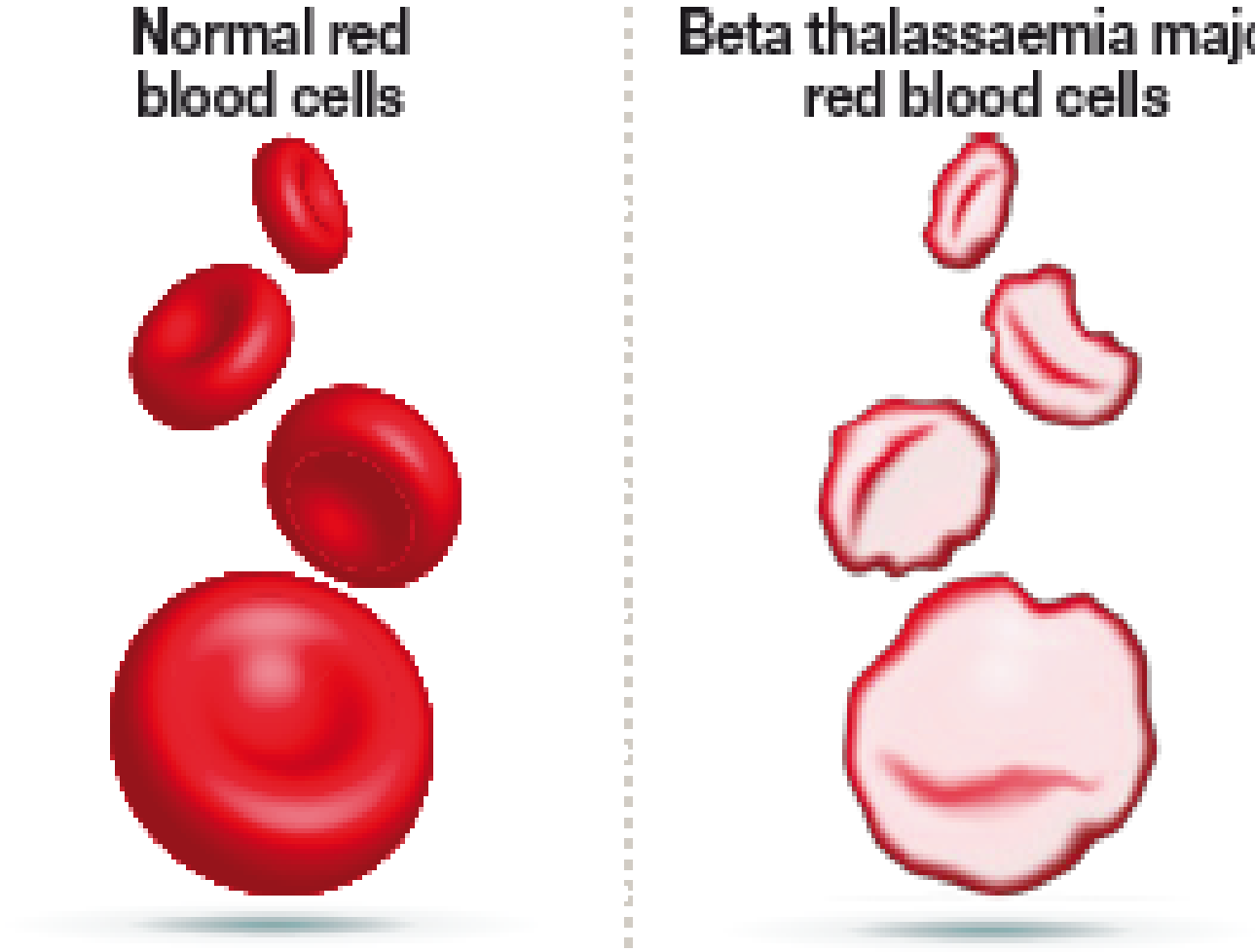
In each pregnancy there is the following possibility:

- 1 in 4 (25%) chance that the child will not be a carrier
- 2 in 4 (50%) chance that the child will carry beta thalassaemia
- 1 in 4 (25%) chance that the child will have beta thalassaemia major



What is beta thalassaemia major?

Beta thalassaemia major is a serious inherited anaemia. Children with beta thalassaemia major cannot make enough haemoglobin, so their bone marrow cannot produce enough red blood cells. The red blood cells that are produced are nearly empty.



Children with beta thalassaemia major are normal at birth but become anaemic, usually between the age of three months and eighteen months. They become pale, do not sleep well, do not want to eat and may vomit their feeds. If children with beta thalassaemia major are not treated, they have miserable lives and usually die between one and eight years of age.

What is the treatment for beta thalassaemia major?

The only possible treatment for most people with beta thalassaemia major is regular blood transfusions, usually every three to four weeks, for the rest of their life. Most children who have these transfusions grow normally and live quite happily into their early teens, but to live longer they need other treatment as well.

After each blood transfusion the red blood cells in the new blood are broken down slowly over the next four months. The iron from the red blood cells stays in the body. If it is not removed, it builds up and can damage the liver, the heart and other parts of the body. If this damage is not prevented most people with beta thalassaemia major die when they are about twenty years old.

To remove the extra iron "iron chelating medicines" must be used. Three different iron chelating medicines are available. Doctors can use different combinations to meet the needs of individual patients.

- *Desferrioxamine (Deferoxamine, Desferal®)* is injected under the skin over 8-12 hours on most nights, using a small portable pump. It has been used since 1964. Patients and their families often find "the pump" very burdensome.

- *Deferiprone (Ferriprox, L1, Kelfer®)* is taken by mouth three times a day. It has been used since 1987. It is particularly good at removing iron from the heart.

- *Deferasirox (Exjade®)* is taken by mouth, once a day as a liquid. It has been used since 2005.

This treatment is very successful and most patients treated with blood transfusions and iron chelation therapy can now lead fairly normal, healthy lives. People with thalassaemia treated in this way are expected to have a near normal length of life. When they grow up they are able to work, marry and more and more are able to have children. But even with the correct medical treatment, there is still the risk of complications and the treatment can be unpleasant and often upsetting.

A different sort of treatment called "bone marrow transplantation" is possible for **some** children with beta thalassaemia major. When it is successful the person does not need any more blood transfusions. However, it is not a simple solution. In each case there is a risk that the bone marrow transplantation may not work, or the patient may die or have serious complications. We hope that these problems will be gradually overcome in the future. We are looking for better treatment all the time.

How can we prevent beta thalassaemia major?

If you carry beta thalassaemia, when you have children there are two possibilities:

I. If your partner does not carry any unusual haemoglobin **there is no chance that any of your children could have beta thalassaemia major**, although they could be carriers of beta thalassaemia.

II. If both you and your partner are carriers of beta thalassaemia, then **in each pregnancy there is a one in four (25%) chance that you will have a child with beta thalassaemia major**.

When both partners carry beta thalassaemia, there are several ways to avoid having children with beta thalassaemia major. For instance, the doctors can test for beta thalassaemia major very early on in pregnancy. Many couples who carry beta thalassaemia decide to test each pregnancy to find out if the baby has beta thalassaemia major. If it has, they may decide to stop the pregnancy and to try again for a child that does not have beta thalassaemia major. There are several other ways to avoid having children with beta

thalassaemia major such as adoption, artificial insemination, egg donation or pre-implantation genetic diagnosis (PGD). PGD involves testing an embryo before it has settled in the womb. It is a long and very complex process that combines in-vitro fertilisation (IVF) techniques with DNA testing.

Should my relatives have a blood test for beta thalassaemia as well?

Yes. The fact that you carry beta thalassaemia means that your relatives also have a high chance of carrying beta thalassaemia because it is "in the family".

Your father and mother, brothers and sisters have a 1 in 2 (50%) chance of carrying beta thalassaemia.

Your uncles and aunts have a 1 in 4 (25%) chance of carrying beta thalassaemia.

Your cousins have a 1 in 8 (12.5%) chance of carrying beta thalassaemia.

You should tell other members of your family about your beta thalassaemia and show them this booklet. Encourage them to have a blood test to see if they also carry beta thalassaemia. If they are carriers they should ask to be referred to a specialist thalassaemia counsellor.

They can arrange for a thalassaemia test through their GP (family doctor), or by contacting the UK Thalassaemia Society.

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