

WHO/HDP/EM/HB.BT/94.5
ENGLISH ONLY
Distr.: GENERAL

EDUCATIONAL MATERIALS

ON

HAEMOGLOBINOPATHIES:

BETA THALASSAEMIA



**WORLD HEALTH ORGANIZATION
HEREDITARY DISEASES PROGRAMME**

1994

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Dear Reader

You have been given this information because you have been told that you carry beta thalassaemia trait, or because you are interested in it.

This section will give you a lot of information about beta thalassaemia trait, but these are the points that matter most:

- I Your beta thalassaemia trait is not an illness and does not affect your own health. However, it could affect the health of your future children.
- II Do not forget that you carry beta thalassaemia trait. Keep this booklet, and your blood test results if you have them, among your personal documents.
- III There is nothing bad about carrying beta thalassaemia trait. 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.
- IV If your partner (present or future) does not carry thalassaemia, or an abnormal haemoglobin, there is no danger. However, your children may be carriers like yourself. They should have their blood tested at some time before they have children of their own.
- V If both you and your partner carry beta thalassaemia trait, there is a danger 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 "haemoglobinopathy counsellor", or a genetic counsellor.

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

SECTION I: ALL YOU NEED TO KNOW ABOUT BETA THALASSAEMIA TRAIT

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

This booklet is for people who have had a blood test that shows they carry **BETA THALASSAEMIA TRAIT**.

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Note

Beta thalassaemia trait is often called "thalassaemia trait" for short, because it was discovered before alpha thalassaemia trait. This can be misleading, because alpha and beta thalassaemia are the same in some ways, but different in others. In order to be completely clear, we talk about "beta thalassaemia trait" throughout this booklet.

● **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 they're not eating enough of the foods that contain iron, or because they have lost blood by bleeding.

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 disorder.

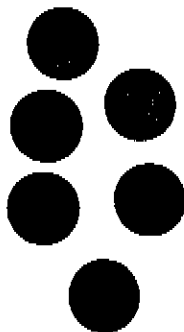
3. **WHAT IS BETA THALASSAEMIA TRAIT ?**

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

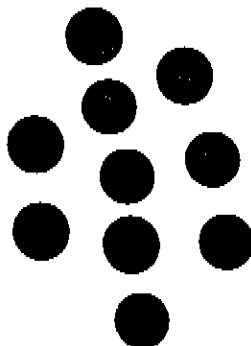
Most people with beta thalassaemia trait do not know that they have it. You only discover it if you have a special blood test, or if you have a child with beta thalassaemia major.

People with beta thalassaemia trait have smaller red cells than usual.

Normal sized red blood cells



Beta thalassaemia trait red blood cells



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The fact that people with beta thalassaemia trait 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.

1. 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:

- **Beta thalassaemia trait.** People with beta thalassaemia trait are perfectly healthy themselves, but if their partner also has beta thalassaemia trait, there is a chance that some of their children could have **beta thalassaemia major**. There are about 200,000 people with beta thalassaemia trait in Britain. They are sometimes called "healthy carriers of thalassaemia". Beta thalassaemia trait is sometimes called thalassaemia minor.
- **Beta thalassaemia major.** This is a very serious blood disorder that begins 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 100,000 children are born in the world with beta thalassaemia major. In Britain, there are about 500 young people with the disorder.

Beta thalassaemia major is sometimes called **Mediterranean Anaemia, Cooley's Anaemia, or Homozygous Beta Thalassaemia.**

2. 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 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.

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.

People with beta thalassaemia trait also have slightly more of a kind of haemoglobin called **haemoglobin A₂** in their blood.

Beta thalassaemia trait 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 Trait ?**

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

● **How Do You Find Out If You Have Beta Thalassaemia Trait ?**

You have to have a special blood test. The doctors can tell by measuring the size of your red blood cells and how much haemoglobin A₂ you have in your blood.

● **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 Beta Thalassaemia Trait Affect the Sort of Work You Can Do ?**

No.

● **Can Any Treatment Change Beta Thalassaemia Trait ?**

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

● **Can Beta Thalassaemia Trait Turn Into Beta Thalassaemia Major ?**

No.

● **Do Beta Thalassaemia Carriers Ever Need Iron Medicine ?**

Yes, they sometimes do, but not because of their beta thalassaemia trait. 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 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 with beta thalassaemia trait need extra iron just as much as any other pregnant women.

● Why is Beta Thalassaemia Trait Found in Certain Countries ?

People with beta thalassaemia trait are less likely to die if they catch malaria, so beta thalassaemia trait can be a great advantage. In the past, in countries where malaria was very common, people with beta thalassaemia trait survived malaria when other people died. They passed the trait on to their children, so as time passed it became more common in malarious parts of the world. Now we can usually cure or prevent malaria, but beta thalassaemia trait does not go away when malaria disappears.

● Does Beta Thalassaemia Trait 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 Beta Thalassaemia Trait ?

Very many countries used to have malaria and all now have quite a large number of people with beta thalassaemia trait. For instance, in Cyprus one in seven people have beta thalassaemia trait (both Turkish and Greek Cypriots), and in Greece one in twelve people have beta thalassaemia trait. In Italy and all of the Middle East and Asia, including India, Pakistan, Southern China, Hong Kong and Vietnam, the number of people with beta thalassaemia trait varies from one in fifty to one in ten in different areas. In Africa and the West Indies about one in fifty people have beta thalassaemia trait. About one in every thousand people of British origin have the trait.

● Other Forms of Thalassaemia Trait

This booklet is all about beta thalassaemia trait, but there are other forms of thalassaemia trait:

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

Alpha-thalassaemia trait is very different from beta-thalassaemia trait. It only rarely causes any illness in children. People with alpha-thalassaemia trait do not carry beta-thalassaemia trait. This booklet does not apply to them. A separate leaflet, called "Information for People who Carry Alpha Thalassaemia Trait" can be obtained from the UK Thalassaemia Society, at the address at the end of this booklet.

In addition to the thalassaemias there are three important forms of abnormal haemoglobins. These are:

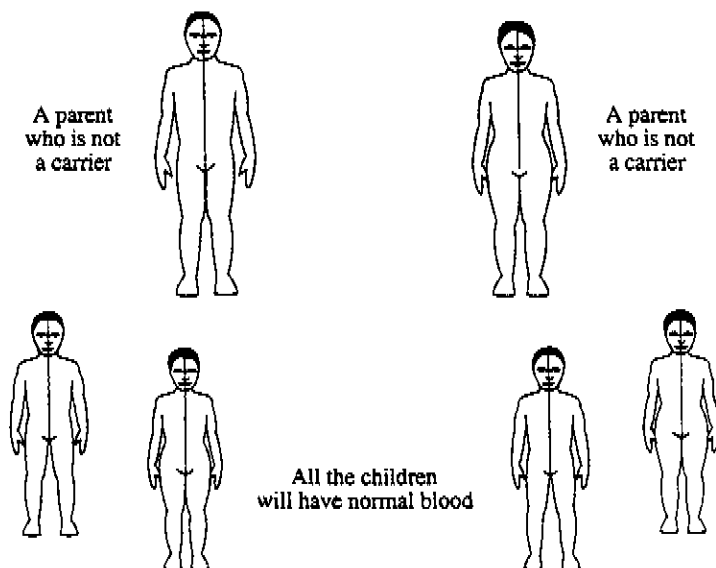
- Haemoglobin S
- Haemoglobin C
- Haemoglobin E
- Haemoglobin O Arab

If someone who carries beta-thalassaemia chooses a partner who carries one of these abnormal haemoglobins, there is a risk that some of their children could have a serious anaemia, like thalassaemia major.

4. HOW IS BETA THALASSAEMIA TRAIT PASSED ON FROM PARENTS TO THEIR CHILDREN ?

Let us consider three sorts of couples.

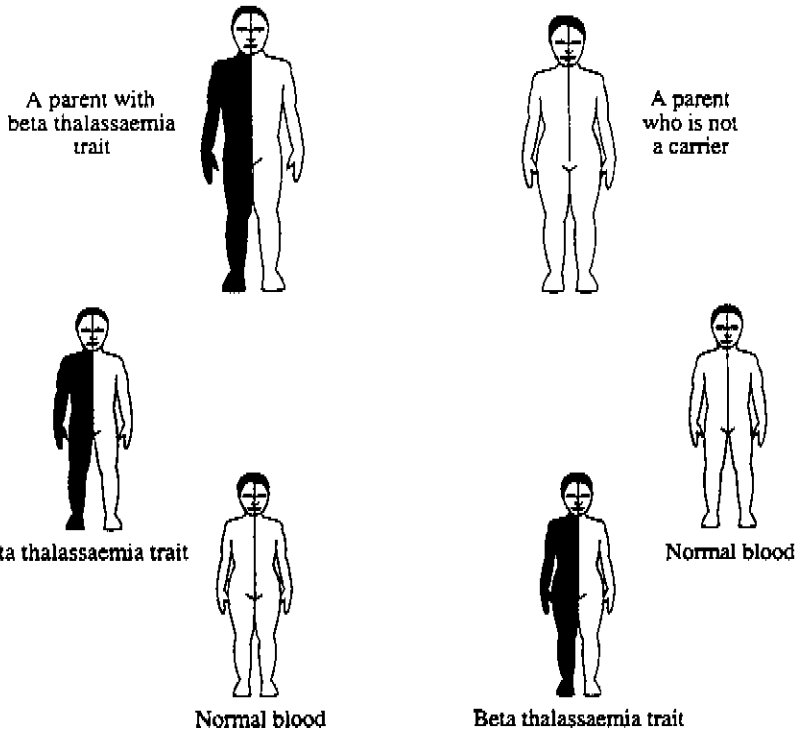
- (a) If both parents are not carriers, they cannot possibly pass on beta thalassaemia trait or beta thalassaemia major to their children. All their children will have normal blood.



None of the children will have beta thalassaemia trait or beta thalassaemia major

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



None of the children will have beta thalassaemia major

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People with beta thalassaemia trait, like you, are completely healthy. They can pass on the trait through many generations without anybody realizing that it is "in the family".

This is probably the situation in your family. As you have beta thalassaemia trait, 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. They also may have a risk of having a child with beta thalassaemia major.

- (c) If both parents carry beta thalassaemia trait, their children may have beta thalassaemia trait or they may have completely normal blood, or they may have beta thalassaemia major.

In each pregnancy there is a one in four (25%) chance that their child will have normal blood, a two in four (50%) chance that the child will have beta thalassaemia trait, and a one in four (25%) chance that the child will have beta thalassaemia major.

A parent with
beta thalassaemia
trait



A parent with
beta thalassaemia
trait



Beta
thalassaemia
trait



Normal
blood



BETA
THALASSAEMIA
MAJOR



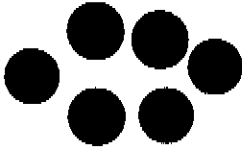
Beta
thalassaemia
trait



5. WHAT IS BETA THALASSAEMIA MAJOR ?

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

Normal sized red blood cells



Beta thalassaemia major red blood cells



Children with beta thalassaemia major are normal at birth but become anaemic 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. They usually die between one and eight years of age.

● Can Beta Thalassaemia Major Be Treated ?

The only treatment for beta thalassaemia major is regular blood transfusions, usually every four weeks for the rest of their lives. Most children who have these transfusions grow normally and live quite happily into their early teens, but to live longer than twenty, 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.

At present the only way to remove the extra iron from the body is to give injections of a drug called Desferal under the skin from a small pump 5-7 nights of every week. Desferal picks up the iron and carries it out in the urine. This treatment is very successful and most children treated with blood transfusions and Desferal can now lead fairly normal healthy lives. When they grow up they are able to work, most marry, and more and more are able to have children. But the treatment is unpleasant and often upsetting.

A different sort of treatment called "bone marrow transplantation" is now 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 have beta thalassaemia trait, this is, if you are a healthy carrier of beta thalassaemia, when you have children there are two possibilities:

- (a) If your partner has normal blood, **there is no chance that your children could have beta thalassaemia major**, though they could have beta thalassaemia trait.
- (b) If both you and your partner have beta thalassaemia trait, **then in each pregnancy there is a one in four chance that you will have a child with beta thalassaemia major.**

When both partners carry beta thalassaemia trait, there are several ways to avoid having sick children. For instance, doctors can now test for thalassaemia major very early on in pregnancy while the baby is still in the womb. Many couples who both carry beta thalassaemia trait decide to test each pregnancy to find out if the baby has beta thalassaemia major. If it has, they often 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, or artificial insemination. To find out more, ask your doctor to arrange for you to visit a genetic counsellor.

6. **SHOULD MY RELATIVES HAVE A BLOOD TEST FOR THALASSAEMIA AS WELL ?**

Yes. The fact that you carry beta thalassaemia trait 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, and your children 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 trait, and show them this booklet. Encourage them to have a blood test to see if they also carry beta thalassaemia trait, so that they can avoid problems for themselves.

They can arrange for thalassaemia test through their general practitioner, or by contacting one of the thalassaemia and sickle cell centres listed at the end of this booklet.

7. LIST OF CONTACT ADDRESSES

The following addresses have been provided by the Thalassaemia International Federation - TIF, and are as of 1994.

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SECTION II: PRENATAL DIAGNOSIS FOR BETA THALASSAEMIA

This section is addressed to couples who both carry beta thalassaemia. Sometimes beta thalassaemia is written β -thalassaemia.

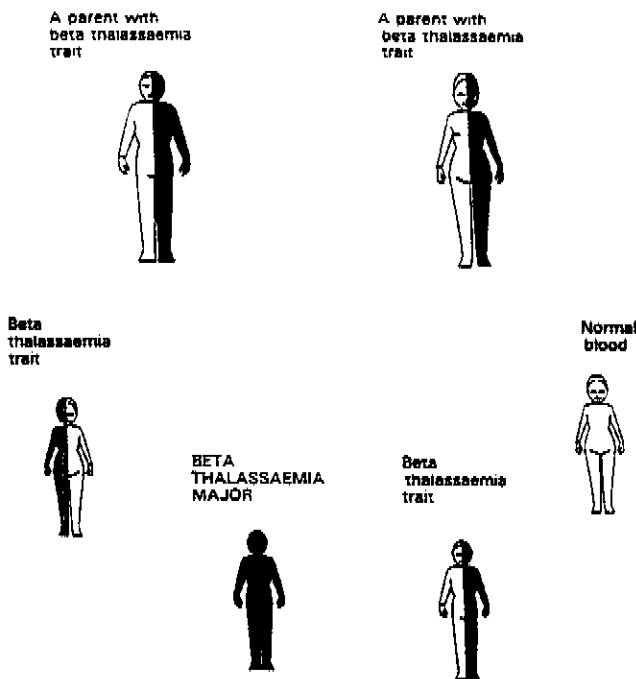
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1. CARRIER COUPLES

When two thalassaemia carriers have a child, there is a 1 in 4 risk that the child will have thalassaemia major. This risk is the same in each pregnancy. You cannot know in which pregnancy or in which order an affected child will come.

Every characteristic of our body, such as eye colour, shape of the face, or haemoglobin production, is determined by genetic material called "genes" which are inherited from our parents. We inherit two genes for every characteristic, one from each parent. We all have two genes that control the haemoglobin in our red blood cells. Being a carrier of thalassaemia means that one of the two "genes" that control the production of haemoglobin in your red blood cells is normal, and one is defective. Since one of the two genes is functioning well, you are healthy. When a child is being formed it inherits one gene for haemoglobin from each parent. When a parent is a carrier, a child can receive either a defective gene or a functioning one from that parent.

The picture below shows that when both parents are carriers, as in your case, there are three possibilities.



1. The baby can receive two functioning genes (one from each parent). In this case it will be normal. In each pregnancy, there is a 1 in 4 chance of a normal baby.
2. The baby can receive one functioning gene and one defective gene. In this case it will be a carrier, like you. In each pregnancy, there is a 1 in 2 chance that the baby will be a carrier.
3. The baby can receive two defective genes (one from each parent). In this case it will have thalassaemia major. In each pregnancy, there is a 1 in 4 chance of a thalassaemic baby.

So, in every pregnancy there is a 3 out of 4 (75%) chance that the child will be healthy, and a 1 in 4 (25%) risk that will have thalassaemia major. In every pregnancy, your chance of having a healthy child is much higher than your risk of having a sick one.

2. THE LIFE OF A CHILD WITH THALASSAEMIA MAJOR

Children with thalassaemia major cannot make enough haemoglobin in this blood. They are normal at birth but usually become severely anaemic at some time between 6 and 18 months of age. The only treatment is by regular blood transfusions every 4 weeks. They need this treatment life-long.

But the transfusions cause another problem. Blood contains a lot of iron, and this gradually accumulates in the body. Iron is poisonous. It can damage the liver, heart and other organs. Unless it is removed it causes death at about 20 years of age. Fortunately, iron can be removed using a drug call Desferal. Unfortunately, Desferal cannot be taken by mouth. It has to be infused under the skin through a fine needle using a little portable pump. Usually the pump is used at night for 8-12 hours while the child sleeps, on 5-6 days a week.

With this combined treatment, people with thalassaemia major can live at least up to 35 years. They can go to school and work normally, and many can have a family. But they find the treatment very burdensome.

Treatment is steadily improving. In the past 10 years or so, an increasing number of thalassaemic children have been "cured" by bone marrow transplantation. However, bone marrow transplantation is not easy. It is possible only for patients with a brother or sister who matches their own tissue type fully, and there are serious risks to life and health.

3. HOW CAN YOU AVOID HAVING AN AFFECTED CHILD ?

If you do not wish to face the problems and pains of having affected children, then you have a number of choices. This booklet is mainly about prenatal diagnosis, but you need to know that there are other possibilities.

Other choices:

- (a) You may decide to have children without interfering with pregnancy at all. Some people make this choice if they cannot accept a termination of the pregnancy under any circumstances, or if they do not want to take the slightest risk of losing the pregnancy as a result of tests. If this is your choice, we will help you as much as we can.
- (b) You may choose to separate and find another partners who is not a carrier, since a carrier and a non-carrier cannot produce an affected child. This choice is open to those not yet fully committed to a partner. In fact, very few couples make this choice.
- (c) You may choose to stay together but not have children, or else adopt a child.
- (d) You may choose to use the techniques of "assisted reproduction" to have an unaffected child. There are two ways to do this.
 - Firstly, you (the woman) may be given sperm from a man who is not your partner. (The donor man must be tested and found not to be a carrier).
 - Secondly, your partner's sperm can be used to fertilise eggs taken from another woman. (The donor woman must be tested and found not to be a carrier). The fertilised eggs can then be placed in your womb.

However, both of these methods are expensive. If you are interested, ask for more information from your genetic counsellor.

Finally, if you wish to have your own healthy children and avoid having an affected child, you can ask for prenatal diagnosis in each pregnancy.

4. WHAT IS PRENATAL DIAGNOSIS ?

This is a test that can be done on the baby before it is born to see whether it is affected by a particular disease or not. If it is affected, the couple may decide to stop the pregnancy, and try again for an unaffected child. If the baby is not affected, then the pregnancy can continue normally.

Remember, for most couples at risk for sickle cell disorder there is a 3 out of 4 chance of a healthy child in each pregnancy, so most pregnancies continue normally after the test.

You will need to go through the test in every pregnancy. There are couples who have built up a family of 3 or 4 healthy children using the test each time.

Material from the baby can be obtained for testing in three different ways, call chorionic villus sampling, fetal blood sampling and amniocentesis. The type of test you have depends on the stage of your pregnancy and the position of the placenta. The test can be done between 10 and 22 weeks of pregnancy - but obviously the earlier the better.

Nowadays the best way to examine the material from the baby is by analysing the haemoglobin genes. This is called "DNA analysis". More than a hundred different alterations in the haemoglobin gene can cause thalassaemia. Therefore it is necessary to study blood from both parents to find out your particular alteration. If we cannot detect your alteration, we may need to study blood from your parents, or from your children if you have any. In order to do the test in a rush, it is best to study both of you, and other family members if necessary, before you start a pregnancy.

5. CHOOSING PRENATAL DIAGNOSIS

Once you have become pregnant, we ask you to attend our clinic as soon as possible. It is important for both partners to come for this first visit, if at all possible. The first visit is necessary for the following reasons.

- To discuss the advantages and disadvantages of the test fully with you, and answer all your questions, so that you can decide if you want a test or not.
- To do an ultrasound scan, so that if you want the test we can plan the best date and method. This will depend on the exact stage of your pregnancy, the position of the placenta, etc. It is useful to drink a lot of water before you come to the hospital, to help with the ultrasound scan.
- To take blood samples from you and your partner if necessary.
- To make up your medical notes.

If you choose to have prenatal diagnosis, you make a separate visit for the test. You come into hospital for a few hours in the morning, with your partner if he wishes to come. We do another ultrasound scan to see exactly where the baby and the placenta are. Then we do the test that is the best for you. We use ultrasound all during the test, so that we can see exactly what is happening. You are not put to sleep. You can go home later the same morning.

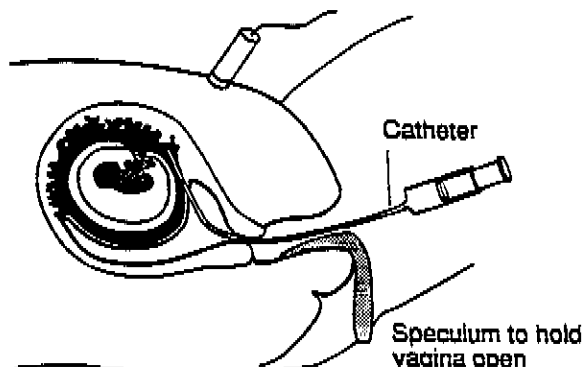
6. CHORIONIC VILLUS SAMPLING

This test is called CVS for short. It can be done early in pregnancy, from about 10 weeks after your last menstrual period. The best timing is between 10 and 12 weeks.

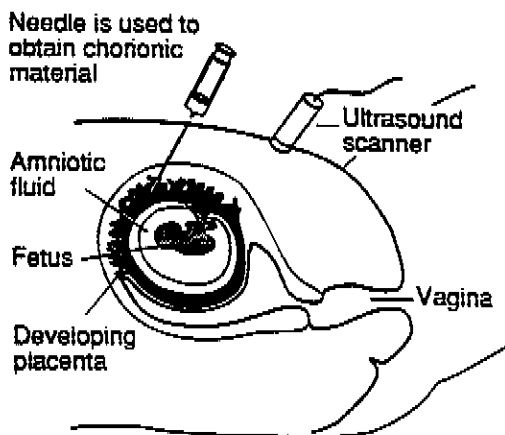
In chorionic villus sampling, a small amount of material is taken from the developing placenta. The placenta is where the baby is attached to the mother. It develops from the tissues of the baby, not of the mother, so it has the same constitution as the baby. It is made up of chorionic villi.

There are two ways to obtain a sample of chorionic villi: either through the vagina or through the abdomen (tummy). Which one we use depends on the position of the placenta. We use an ultrasound scan all the time so that we can see exactly what we are doing.

When it is possible to reach the placenta through the vagina, we do not use a needle. The obstetrician puts a very thin plastic tube through the vagina and then into the womb. It is so thin that most women hardly feel it. It does not touch the baby, or the little bag of water it is lying in. Then we attach a syringe to the end of the tube and suck out a very small sample of chorionic villi from the placenta. The picture below shows how the test is done.



When we cannot reach the placenta through the vagina, we reach it by putting a long thin needle through the abdomen and into the womb. We inject a local anaesthetic into the skin before we insert the needle, to "freeze it". The local anaesthetic stings and there is a brief feeling of pressure when the needle is put in, but you should feel very little pain. When the ultrasound picture shows that the needle is in the right place, the obstetrician fixes a syringe to it and gently sucks some tissue out. The picture below shows how this test is done.



Once some tissue has been sucked out, we immediately look at it under a microscope to check that it is from the placenta. If it is, we stop. If it is not, we move the tip of the tube or needle slightly and try again. Sometimes we have to make 2 or 3 attempts to get the tissue. The test usually takes 10 to 20 minutes.

After the test, we invite you to rest in the hospital for about 30 minutes. Then you can go home. It is wise to take things easy for one or two days. This means you can go about as usual, but should avoid heavy work. Avoid sexual intercourse for 10 days after the test. If the test has been done through the vagina you may see some blood spots for a few days. This is usually harmless. However, if there is a lot of blood or you have pain or fever, you should contact us immediately at the telephone number we have given you.

How do we test the chorionic villus sample ?

The genes responsible for all the characteristics inherited from the parents, including haemoglobin, are made of a material called DNA. All the tissues of our body contain our whole DNA pattern. In the fetus, this includes the placenta. We study DNA from the chorionic villi so see if the baby's genes for haemoglobin are normal, or if an alteration has been passed on from the parents. It takes from 3 to 10 days to analyze DNA, so we usually have the result in about one week. We will tell you immediately we know the result. If the baby is not affected you can continue your pregnancy with confidence.

Is the DNA test accurate ?

DNA analysis is the best method for diagnosing inherited diseases. However, in every medical test there is a small possibility of a mistake. All human beings can make a mistake however careful they try to be. Sometimes nature itself can "play a trick" and make us make a mistake. So there is a chance of a mistake with this test, but it is very very small, less than 1 in 200 (0.5%).

The DNA test depends on studying the parents' DNA and then comparing it with the baby's DNA. The test is not reliable if the man who comes for testing is not the baby's real father. It is very important for the woman to tell us confidentially if there is any possibility that her partner is not the baby's father.

Is CVS safe ?

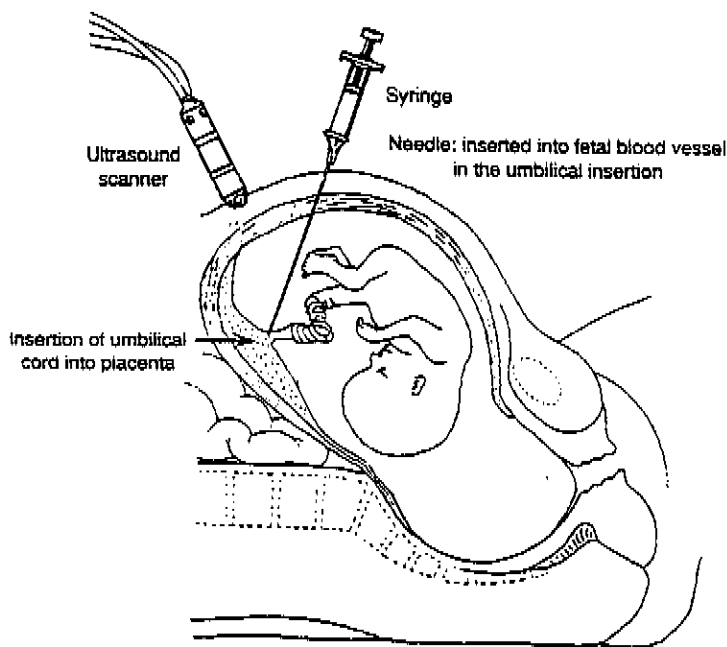
We are still not exactly sure how safe CVS is. There is practically no risk to the mother. The main risk is that the test could cause a miscarriage. We do the test as gently as possible, but anything that interferes with a pregnancy may cause a miscarriage. At the moment, the risk of miscarriage after CVS during the first 12 weeks seems to be around 1 in 50 (2%). However, it is difficult to give an exact figure for the risk of miscarriage after this test because other factors can influence it. For example, the older the mother the higher the risk of miscarriage. When a miscarriage does occur, it can be difficult to tell whether it was due to the test or not, because many miscarriages happen naturally at around 12 weeks of pregnancy without any test.

It also seems that, very rarely, CVS can cause abnormalities in the development of the limbs or fingers. However, this has only happened if the test was done before 9.5 weeks of pregnancy. There is no evidence that CVS can cause any problems when it is done at 10 weeks or later. This is why we prefer to do CVS between 10 and 12 weeks.

7. FETAL BLOOD SAMPLING

In this test we take blood from the fetus. It has to be done at 18-22 weeks after the last menstrual period. We use it when the pregnancy is already quite far advanced and we cannot reach the placenta for CVS. We also use it when we cannot make the diagnosis by DNA analysis.

The obstetrician first injects some local anaesthetic into the skin of the tummy. Then he or she puts a very thin needle through the tummy and takes a few drops of blood from the baby's umbilical cord, where it is attached to the placenta. The needle does not touch the baby itself. We use an ultrasound scan all the time so that we can see exactly what we are doing.



After the test the woman rests for 20-30 minutes in hospital. It takes about a week to get the result.

How do we test the fetal blood ?

When possible we test it by DNA analysis, as described for CVS. If this is not possible we test it by analysing the baby's haemoglobin.

In the womb, babies with normal blood make mainly fetal (baby) haemoglobin (HbF) but they also make a small amount (4-9%) of adult haemoglobin (HbA). So in a baby with normal blood there is a small amount of adult haemoglobin. In a baby with thalassaemia trait there is half that much. In a baby with thalassaemia major there is little or no adult haemoglobin. So to find if the baby is affected, we find out how much adult haemoglobin it has. In a baby that carries sickle cell trait this is half Hb A and half Hb S.

If you have this test, we will be happy to show you your results and explain them to you, if you wish to see them.

Is fetal blood sampling reliable ?

We think the test is very reliable. But in every medical test there is always a small possibility of a mistake. We think there is about a 1 in 100 (1%) chance of a mistake. If the results are doubtful, we may have to repeat the test after 2 weeks.

Is fetal blood sampling safe ?

Every medical test carries a risk. In fetal blood sampling this is very small. There are practically no risks to the mother. In about 1 in 100 cases, however, a miscarriage will occur. This can happen from a few days to a few weeks after the test.

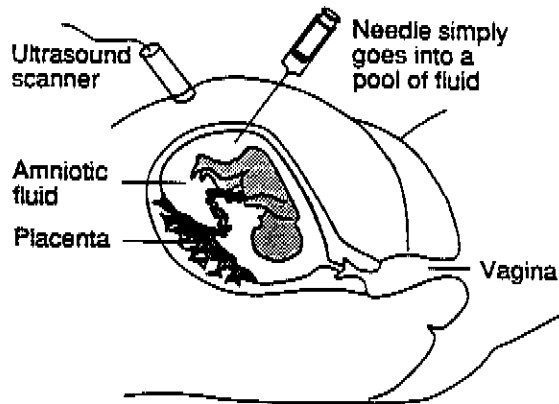
To keep the risk to the minimum, you should take things easy for one or two weeks after the test, avoiding tiring housework and carrying heavy objects like shopping or children. Avoid sexual intercourse for 10 days after the test. If you notice any bleeding or discharge from the vagina, contact the hospital at once.

8. AMNIOCENTESIS

We use this test very occasionally when, for some reason, we cannot do CVS or fetal blood sampling.

How is amniotic fluid obtained ?

Amniotic fluid is the liquid that surrounds the baby in your womb. As shown in the picture below, the obstetrician puts a small needle through the tummy into the womb and takes out a small amount of the fluid from around the baby.



No local anaesthetic is necessary. There is only a brief feeling of pressure when the needle is put in. It usually takes only a few minutes to draw off some fluid.

How do we test amniotic fluid ?

Amniotic fluid can be tested by DNA analysis, as with CVS. However, it cannot be analyzed at once because it contains very little material from the baby. We have to grow it in culture in the laboratory for at least 2 weeks. Then the DNA analysis requires another 5-7 days.

Therefore, the results of this test take at least 3 weeks to come from the laboratory.

Is amniocentesis safe ?

As we said before, every medical test carries a risk, but the risk of this one is very small. It is almost entirely safe for the mother, and there is less than a 1 in 100 chance that it could cause a miscarriage.

Is amniocentesis accurate ?

It is just as accurate as the CVS test.

9. TERMINATION OF PREGNANCY

If the test shows that the baby is affected, you may decide to end the pregnancy. This is done in one of two different ways, depending on the stage of your pregnancy.

Early termination

This is possible if you are less than 14 weeks pregnant. You come into the hospital one evening. The next day you are put to sleep as if you were having an operation. Your womb is emptied through your vagina. The operation is quick and you feel no pain. You can go home the next day.

A termination does not reduce your chance of having another baby. A few months later you can try again to have a healthy child, and you can have another prenatal diagnosis if you wish.

Late termination

We have to use a different method if you are more than 14 weeks pregnant. We inject a substance called prostaglandin into the womb. This brings on labour pains and starts a miscarriage. The procedure lasts about 17 hours. You can have plenty of pain-killers and some anaesthesia, but of course it is much more upsetting than an early termination.

This type of termination does not spoil your chance of having other pregnancies. After some months you can try again to have a healthy child. But next time it is best to have an earlier prenatal diagnosis at about 10 weeks of pregnancy, if possible.

10. PLEASE COME EARLY !

The test should not be done in a rush, so we like to see you and your partner before you become pregnant. If you are planning to have a baby and think you will want to have it tested for haemoglobin disorders, please contact your local counselling service soon. It may be necessary to take small blood samples from you, your partner, and any other children you have. If you are already pregnant and want the baby tested, contact your local counselling services as soon as possible. They will be able to answer all your questions, and will arrange for you to have a test - if you want one. You can contact your local counselling service at:

