Public Health England

NHS

NHS Sickle Cell and Thalassaemia Screening Programme

Information and choices for women and couples at risk of having a baby with thalassaemia major

Who is this leaflet for?

We have given you this leaflet because the baby you are expecting may have thalassaemia major.

We know this because your blood tests showed one of the following results.

1. You and the baby's father both have one unusual haemoglobin gene and one normal haemoglobin gene.
2. You have one unusual haemoglobin gene and we don't have test results for the baby's father.

Haemoglobin is a substance in the blood that carries oxygen around the body. People who have one unusual gene are known as carriers.

This leaflet explains:

- what it's like to live with thalassaemia major
- the chances of your baby inheriting thalassaemia major
- possible follow-up tests
- the choices available to you

This information should support, but not replace, the discussions you have with your healthcare professional. Your healthcare professional will support you throughout your choices.

Thalassaemia major

Thalassaemia major affects haemoglobin. Thalassaemia major is most common in people with a Pakistani, Cypriot, Italian, Greek, Indian, Bangladeshi, Chinese or other South East Asian family background. About 20 babies are born with thalassaemia major every year in England.

People with thalassaemia major cannot make enough normal red blood cells and cannot produce enough haemoglobin. Thalassaemia major is a serious and lifelong condition, but long-term treatment can help manage many of the symptoms. People with thalassaemia major can lead long, active and fulfilling lives if they manage their condition well and have the right care and support.

There are different types of thalassaemia major. The most common and serious is beta thalassaemia major. Babies can only inherit beta thalassaemia major if both parents are beta thalassaemia carriers. If one parent is a beta thalassaemia carrier and the other carries another type of unusual haemoglobin, the baby could inherit a different type of thalassaemia major.

Thalassaemia intermedia

Thalassaemia intermedia is not usually as serious as thalassaemia major. If the condition is mild, individuals may only need treatment occasionally or not at all. Your healthcare professional will be able to give you more information about this condition as it varies from person to person.

Symptoms

The main symptoms of untreated thalassaemia major are:

- life-threatening anaemia due to low level of haemoglobin in blood
- shortness of breath
- pale skin
- yellowing of the skin and eyes (jaundice)

Other symptoms include delayed growth, reduced fertility, weak bones (osteoporosis) and gallstones.

Treatment

Treatment aims to correct the anaemia caused by a lack of haemoglobin in the red blood cells.

People with thalassaemia major need blood transfusions every 3 to 5 weeks throughout their life. Regular blood transfusions cause a build-up (or overload) of iron in the body. Medicine to remove this extra iron is needed throughout the person's life. This is called iron chelation therapy.

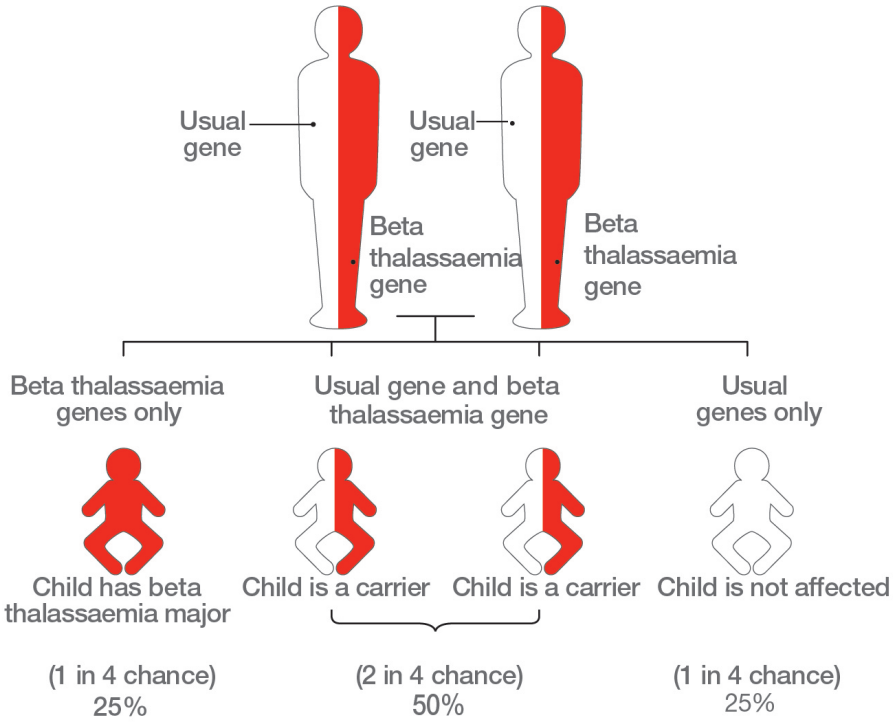
It is important to treat any complications that occur due to anaemia or iron overload, such as delayed growth, heart or bone conditions.

Currently, the only cure for thalassaemia major is a bone marrow or stem cell transplant. This is a complicated and risky procedure which is only suitable for individuals with thalassaemia major who have a matching donor.

What are the chances your baby will inherit the disease?

If you and the baby's father are both carriers then there is a 1 in 4 (25%) chance the baby will inherit thalassaemia major. The diagram below shows how genetic inheritance works.

Both parents in the diagram are carriers. They are drawn in 2 colours to show they have one usual haemoglobin gene (white) and one beta thalassaemia gene (red).



These chances are the same in each and every pregnancy when both parents are carriers.

Follow-up tests

You can choose if you want a test to find out for sure if the baby has inherited thalassaemia major or not. This is called pre-natal diagnosis (PND). It is your decision to have this test or not.

If you decide not to have PND then you will still be offered the newborn blood spot test when your baby is 5 days old. This should detect if your baby has beta thalassaemia major.

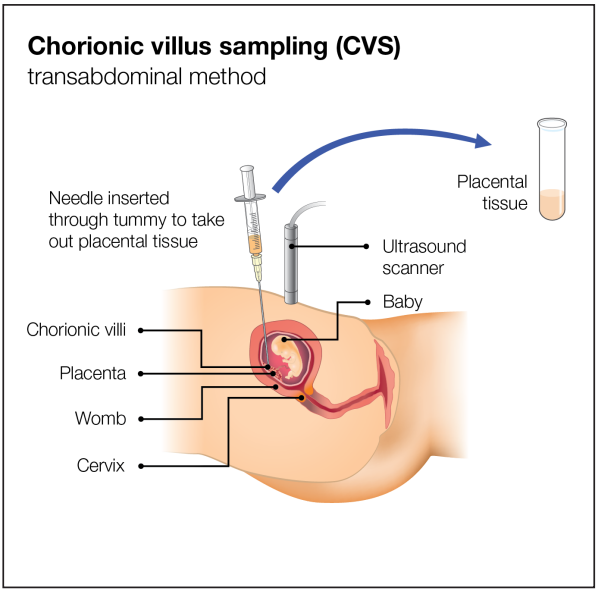
There are 2 main types of PND test depending on the stage of your pregnancy – **chorionic villus sampling** (CVS) and **amniocentesis**. Up to one out of every 100 women who have a CVS or amniocentesis will miscarry due to the test.

CVS is usually done from 11 to 14 weeks of pregnancy but can be done later. CVS can be performed in 2 ways:

- through the abdomen (tummy) – transabdominal
- through the cervix (neck of the womb) – transcervical

Amniocentesis is usually done between 15 and 20 weeks of pregnancy but can be done later.

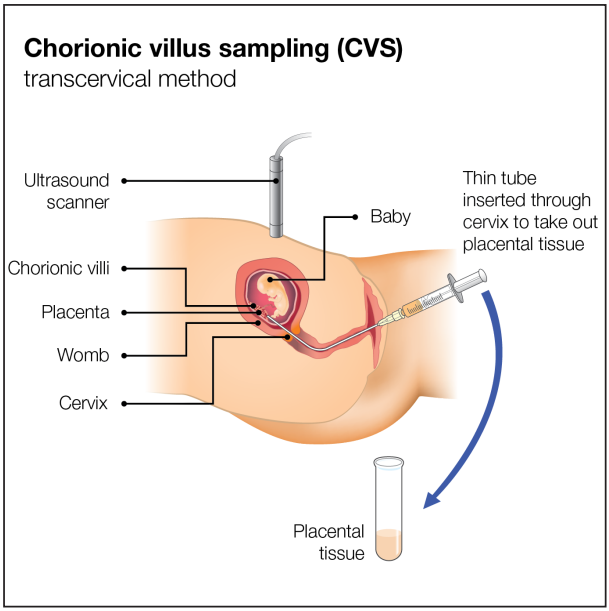
Transabdominal CVS



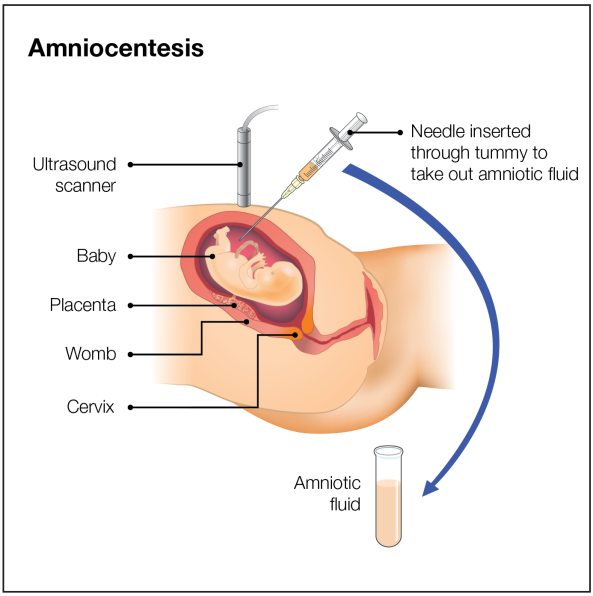
We clean the abdomen with antiseptic and may use a local anaesthetic injection to numb a small area. We insert a fine needle through the abdomen and into the uterus to take the sample. We use an ultrasound probe to guide the direction of the needle.

Transcervical CVS

We insert a thin tube attached to a syringe or small forceps through the vagina and cervix, and guide it towards the placenta using the ultrasound scan. We collect a tiny sample of placental tissue through the cervix.



Amniocentesis



We take a small amount of amniotic fluid (the water around the baby inside the uterus) for testing. The sample contains some of the baby's cells, which contain genetic information.

We clean the abdomen with antiseptic and may use a local anaesthetic injection to numb a small area. We insert a fine needle through the abdomen and into the uterus to take the sample. We use an ultrasound probe to guide the direction of the needle.

Occasionally, for **fewer than 7 in every 100** women, we cannot take enough fluid at the first attempt and have to re-insert the needle. This is usually due to the position of the baby.

If a second attempt fails, we will offer an appointment to have the amniocentesis again on another day.

After taking the needle out, the baby is observed for a short time on ultrasound.

Possible results

There are 3 possible results from PND. Your baby could:

- **inherit thalassaemia major (1 in 4 or 25% chance)**
- **be a carrier of thalassaemia major (2 in 4 or 50% chance)**
- **have normal haemoglobin (1 in 4 or 25% chance)**

In rare cases the screening laboratory cannot give a result. If this happens, you will be contacted and offered a repeat PND test.

Possible choices

If the PND test shows your baby has normal haemoglobin or is a carrier then your pregnancy will continue as usual.

If the PND test shows the baby has inherited thalassamia major, your healthcare professional will talk to you and offer support. You should also have the opportunity to talk to a specialist.

You may choose to:

- **continue with your pregnancy**
- **end the pregnancy (have a termination)**

If you choose to **continue with the pregnancy** your healthcare professional will provide you with information and advice about caring for a child with thalassaemia major, along with sources of support. The UK Thalassaemia Society provides support and information and may be able to put you in touch with local support groups and parents who have children with the same condition.

Your baby will be offered routine newborn blood spot screening (the heel prick test) at 5 days old, which should confirm beta thalassaemia major but will not detect beta thalassaemia intermedia. You should get the newborn screening test result before your baby is 28 days old. Your baby will then be referred to a hospital specialist for treatment and care within 90 days of birth.

If you choose to **end the pregnancy** you will be given as much support as you need to help you come to terms with this difficult decision.

Future pregnancies

If you want to have another baby with the same partner and you are both carriers then the chances of the baby inheriting thalassaemia major will be exactly the same as now, 1 in 4 or 25%.

These chances are the same in each and every pregnancy when both parents are carriers.

However, there are some choices you can make for any future pregnancies.

You can ask your GP, midwife or specialist counsellor for pre-natal diagnosis early in the pregnancy and this can be performed after 11 weeks. This gives you more time to consider your choices if the baby has thalassaemia major. You'll need to see your GP or midwife as soon as you know you are pregnant.

You can consider pre-implantation genetic diagnosis (PGD). This means having an assisted pregnancy using in-vitro fertilisation (often known as IVF). You can ask to be referred to a genetic counsellor to discuss this option.

You can consider an assisted pregnancy using either donated eggs or sperm from people who are not carriers. This means either you or your partner will not be the biological parent of your child. You can discuss this with your healthcare professional.

You can decide not to have any more children.

More information and support

NHS Choices: www.nhs.uk/sct

UK Thalassaemia Society: www.ukts.org
E: office@ukts.org T: 020 88820011

Antenatal Results and Choices: www.arc-uk.org
E: info@arc-uk.org T: 0845 0772290; 020 77137356

Contact a Family: www.cafamily.org
E: info@cafamily.org.uk T: 0808 8083555

For information on how NHS screening programmes use patient information safely and securely, visit
www.gov.uk/phe/screening-data

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Photo credit(s): PHE Screening

More information about sickle cell and thalassaemia screening: www.nhs.uk/sct

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