



Genetic Fact Sheets for Parents

Other Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – <http://www.newbornscreening.info>

Disorder name: Alpha Thalassemia

Acronym: α thal

- What is alpha thalassemia?
- What causes alpha thalassemia?
- What are the symptoms of alpha thalassemia?
- What is the treatment for alpha thalassemia?
- What causes the decreased production of alpha globin?
- How is alpha thalassemia inherited?
- Is genetic testing available?
- What other testing is available?
- Can you test during pregnancy?
- Can other members of the family have alpha thalassemia or be carriers?
- Can other family members be tested?
- How many people have alpha thalassemia?
- Does alpha thalassemia happen more often in a certain ethnic group?
- Does alpha thalassemia go by any other names?
- Where can I find more information?

This fact sheet has general information about alpha thalassemia. Every child is different and some of this information may not apply to your child specifically. Certain treatments may be recommended for some children but not others. If you have specific questions about alpha thalassemia and available treatments, you should contact your doctor.

What is alpha thalassemia?

Alpha thalassemia is an inherited blood disorder. People with alpha thalassemia have a low number of red blood cells and red blood cells that are smaller than normal. Normal red blood cells have a component called hemoglobin that carries oxygen to the body's tissues. Some people with alpha thalassemia experience anemia, or low levels of hemoglobin. Taking iron supplements to treat the anemia caused by alpha thalassemia will not improve symptoms and is discouraged.

There are several types of alpha thalassemia. Ranging from mild to severe, the types of alpha thalassemia are:

1. Silent carrier
2. Alpha thalassemia trait
3. Hemoglobin H
4. Alpha thalassemia major

People with all types of alpha thalassemia can pass it on to their children. Additionally, parents with a mild form of alpha thalassemia can have a child with a severe form. It is important to find out which type of alpha thalassemia your child has and talk with your doctor about what that might mean for your other children and future pregnancies.

What causes alpha thalassemia?

Thalassemias are a group of inherited conditions caused by decreased production of hemoglobin. Hemoglobin is the protein in red blood cells that carries oxygen from the lungs to the rest of the body. Hemoglobin is made up of two components, called alpha globin and beta globin. Both of these components are needed for hemoglobin to do its job efficiently. When a person does not produce enough alpha globin, they have alpha thalassemia. Having too little alpha globin makes the cells smaller than normal. People with the milder types of alpha thalassemia produce more alpha globin than people with the more severe types, leading to milder symptoms.

Alpha thalassemia is not contagious. You cannot get alpha thalassemia from living with, touching, or spending time with a person with alpha thalassemia. Likewise, there is no way to "cure" alpha thalassemia.

What are the symptoms of alpha thalassemia?

Alpha thalassemia causes minimal effects in most people and more serious problems in others. Symptoms can be temporary or chronic (on-going or long-term). It is important to be educated about the symptoms so that they can be

properly managed. The symptoms that a person has depend on the type of alpha thalassemia they have.

Alpha thalassemia will not affect a child's growth, development, intelligence, or ability to learn. You cannot tell that someone has alpha thalassemia by looking at them.

Anemia

People with alpha thalassemia may experience varying degrees of anemia.

The baseline level of anemia will remain constant throughout a person's lifetime. It is not expected to get worse or improve.

If your child has a blood test done, it will likely show that he or she has small red blood cells. The expected size of the cells depends on which type of alpha thalassemia a person has.

Even with mild anemia, children with alpha thalassemia can usually participate in sports and other physical activities.

Silent Carrier and alpha thalassemia trait

Alpha thalassemia silent carrier and trait are not illnesses and will not make your child sick in any way. It is not expected that individuals with silent carrier and alpha thalassemia trait have fatigue, low energy, or lack of stamina.

Hemoglobin H

Most children with hemoglobin H are generally healthy. Some may experience mild to moderate anemia. Signs of anemia include:

- Fatigue (feeling tired or weak)
- Shortness of breath
- Dizziness
- Headaches
- Cold hands or feet
- Pale skin
- Chest pain

Contact your doctor if you think that your child might be experiencing signs of anemia.

Hemoglobin H will not cause a child to get sick more frequently than other children. However, illnesses may last longer or make a child more sick than other children. Also, during times of illness, they may require a blood transfusion. Some children with hemoglobin H have other complications, including gallstones and a larger than expected spleen. Therefore, these children are followed more

closely by their doctor. It is recommended that they see a hematologist, or blood specialist, yearly.

Alpha thalassemia major

Most babies with alpha thalassemia major do not survive until birth or are stillborn. In the rare cases of babies that do survive, they require frequent blood transfusions and medical care. Alpha thalassemia major can be detected on ultrasound in the second-third trimester of pregnancy. A hallmark ultrasound finding that indicates that a baby may have alpha thalassemia major is hydrops fetalis, an accumulation of fluid in the baby.

What is the treatment for alpha thalassmia?

The type of treatment that a child needs depends on the type of alpha thalassemia that he or she has. It is very important that the parents make sure that their family doctor knows that their child has alpha thalassemia.

Iron supplements will not improve anemia caused by alpha thalassemia. If your doctor thinks that your child should take iron supplements, a blood test called ferritin should first be checked. Taking unneeded iron supplements can cause damage to the body.

Silent carrier and alpha thalassemia trait

Children with mild alpha thalassemia do not require any treatment. However, they may have a chance of having a child with more severe alpha thalassemia. When your child reaches an appropriate age, it is important to let him or her know that he or she can pass alpha thalassemia on to their children.

Hemoglobin H

Children with hemoglobin H should have regular medical care to detect and treat any symptoms that may occur. It is recommended that they see the hematologist yearly. Some children with hemoglobin H may need blood transfusions after a severe illness. A small number of children with hemoglobin H need frequent blood transfusions. Other rare complications may require additional treatment.

It is recommended that children with hemoglobin H take folic acid routinely.

Alpha thalassemia major

When alpha thalassemia is detected before birth, pregnancy management options should be discussed with the doctor.

What causes the decreased production of alpha globin?

Genes tell the body how to make proteins. There are four copies of the gene that tells the body how to make alpha globin. When one or more of the alpha globin genes is not working properly, less alpha globin is made.

The number of non-working genes determines what type of alpha thalassemia a person has. The more working genes a person has, the more alpha globin is made.

Type of Alpha thalassemia	Number of working alpha globin genes	Number of non-working alpha globin genes
silent carrier	3	1
alpha thalassemia trait	2	2
hemoglobin H	1	3
alpha thalassemia major	0	4

How is alpha thalassemia inherited?

The inheritance of alpha thalassemia is complex. A child inherits two copies of the alpha thalassemia globin gene from his or her mother and two copies from his or her father. When a parent passes on one or more non-working copies of the alpha globin gene to a child, he or she has alpha thalassemia. If both parents pass on non-working copies of the alpha thalassemia genes, their child might have a more severe type of alpha thalassemia. The type of alpha thalassemia that a baby could have depends on the combination of the four gene copies.

Some parents do not realize that they have alpha thalassemia until their child is diagnosed.

Genetic counseling is available to families who have children with alpha thalassemia. Genetic counselors can answer your questions about how alpha thalassemia is inherited, types of alpha thalassemia that parents could have and how to test other family members. Ask your doctor about a referral to a genetic counselor.

Is genetic testing available?

Genetic testing for alpha thalassemia can be done on a blood sample. Genetic testing, also called DNA testing, looks for the number of working alpha hemoglobin genes. Genetic testing is the only test that can determine specifically what type of alpha thalassemia a person has. Couples who want to find out what type of alpha thalassemia their children are at risk for may choose to have

genetic testing. If you are interested in genetic testing, talk to your doctor about a referral for genetic counseling.

What other testing is available?

Newborn Screening

Newborn screening for alpha thalassemia is now done in all states. A blood spot from a prick on a baby's heel is used to screen for a number of different genetic conditions. Babies with any type of alpha thalassemia can be detected on the newborn screen. However, the newborn screen does not determine what type of alpha thalassemia a baby has.

Additional Testing

Your doctor may recommend that your child have a blood test around one year of age to screen for additional common blood disorders.

Can you test during pregnancy?

DNA testing can be done in future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis. Parents may choose to have testing during pregnancy or wait until birth to have the baby tested. Alpha thalassemia major can often be detected by ultrasound. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

Can other members of the family have alpha thalassemia or be carriers?

The brothers and sisters of a child with alpha thalassemia may also have alpha thalassemia, even if a parent has not noticed any symptoms. Talk to your doctor or genetic counselor about testing your other children for alpha thalassemia.

Each of the parents' brothers and sisters also has a chance to have alpha thalassemia. It is important to talk to other family members about their risk of having alpha thalassemia.

Can other family members be tested?

Other family members can have DNA testing to determine if they have alpha thalassemia. Family members who are interested in testing should talk to their doctor.

How many people have alpha thalassaemia?

Alpha thalassaemia is one of the most common genetic conditions. Because it can be very mild, the number of people who have alpha thalassaemia is unknown.

Does alpha thalassaemia happen more often in a certain ethnic group?

Alpha thalassaemia occurs most often in people of Southeast Asian, Mediterranean, North African, Middle Eastern, Indian, and Central Asian ancestry. However, a person of any ethnic background can have alpha thalassaemia.

Does alpha thalassaemia go by any other names?

Another name for alpha thalassaemia trait is alpha thalassaemia minor.

Hemoglobin H may also be called Hemoglobin H Disease.

Some people may call alpha thalassaemia "low blood".

Alpha thalassaemia is NOT the same as beta thalassaemia. They are two completely different conditions. Information about beta thalassaemia does not apply to alpha thalassaemia.

Where can I find more information?

Cooley's Anemia Foundation
www.cooleysanemia.org

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