

ALPHA THALASSEMIA TESTING – LETTER TO RELATIVES

Dear _____,

As you may already know, you have a family member, _____, who has alpha thalassemia _____ (trait or disease). This letter is to give you information about what this may mean for you and other family members.

Alpha thalassemia trait is a benign condition that does not cause any health problems. An alpha thalassemia trait carrier may experience a very slight anemia that is not correctable with iron supplementation.

Alpha thalassemia disease is one type of thalassemia disease, which causes anemia. There are different types of alpha thalassemia, including hemoglobin H disease and alpha thalassemia major. Hemoglobin H disease results in chronic anemia, as well as an intolerance to certain medications and other exposures. These exposures can cause the anemia to worsen, even become life threatening, and may lead to the need for blood transfusion in order to correct the anemia. Alpha thalassemia major is a more severe disease that most often leads to miscarriage or stillbirth in affected fetuses. However, a small number of individuals are long-term survivors with alpha thalassemia major. These individuals most often need special treatment before birth in order to survive. After birth, individuals with alpha thalassemia major are dependent on blood transfusions every few weeks.

Having a relative with alpha thalassemia may affect your own and other family members' health care. Alpha thalassemia is not something one “catches” or “outgrows.” It is *inherited*, meaning it's caused by changes in specific instructions (or *genes*) passed down to us from our parents through the egg and sperm. People who are related to one another share some of the same instructions, which explains why family members often resemble one another. This also explains why **anyone related by blood to a person with alpha thalassemia disease or trait may themselves have alpha thalassemia trait.** There are different types of alpha thalassemia trait: 1) “cis” type of alpha thalassemia trait, 2) “trans” type of alpha thalassemia trait, 3) silent alpha thalassemia trait, and 4) Constant Spring trait. Alpha thalassemia trait will *not* cause health problems and will *never* become alpha thalassemia disease. But if two parents both have trait, there is a chance each time they have a child that he or she will be born with alpha thalassemia disease. So it is common for a person with alpha thalassemia disease to be the *first and only* person in his or her family with the disease. But it's important to understand that other family members may have trait, or could be born with alpha thalassemia disease if both their parents have trait.

Special blood tests can help tell if you have alpha thalassemia trait:

- ✓ hemoglobin electrophoresis
- ✓ quantitative hemoglobin A2
- ✓ quantitative hemoglobin F
- ✓ complete blood count (can substitute hemogram)
- ✓ FEP level (can substitute ferritin)

All of these tests can be performed on *one single blood sample*. Your doctor or genetic counselor can help arrange these tests, which are fairly inexpensive and covered by most types of insurance.

Sometimes, the tests listed above do not give a complete answer as to whether a person has some sort of alpha thalassemia trait, which could lead to alpha thalassemia disease in his or her children. In these cases, **additional DNA tests (gene tests) are needed**. Your doctor or genetic counselor can recommend whether this type of follow-up testing is necessary. This type of testing is costly, so it is important to check with your insurance company ahead of time to see if your policy covers it.

If you learn you have alpha thalassemia trait, you can consider other testing options. You may want to have your *partner* tested. If your partner also has trait, there will be a chance that each of your children will be born with alpha thalassemia disease. There are tests that a woman can have *early in pregnancy* to find out if her baby has alpha thalassemia disease. A genetic counselor can provide information and help couples with their individual and personal decisions about this type of testing.

I hope you find this information helpful. For more information, please contact your doctor or a genetic counselor in your area.

Signed,

Additional messages: