BETA THALASSEMIA TESTING – LETTER TO RELATIVES

Dear _____,

As you may already know, you have a family member, _____, who has beta thalassemia _____ (trait or disease). This letter is to give you information about what this may mean for you and other family members. It was prepared by the Children's Hospital Oakland Thalassemia Center.

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

Beta thalassemia disease is one type of thalassemia disease, which causes anemia. There are different types of beta thalassemia disease, including hemoglobin E/beta thalassemia, beta thalassemia intermedia, and beta thalassemia major. Beta thalassemia disease results in chronic anemia, meaning a person's blood cells are smaller than normal and less able to circulate oxygen and nutrients to all parts of the body. In some cases, blood transfusions may be necessary to help manage the anemia.

Having a relative with beta thalassemia may affect your own and other family members' health care. Beta 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 beta thalassemia disease or trait may themselves have beta thalassemia trait**. Beta thalassemia trait will *not* cause health problems and will *never* become beta thalassemia disease. But if two parents both have trait, there is a 25% chance each time they have a child that he or she will be born with beta thalassemia disease. So it is common for a person beta 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, and that future children could be born with beta thalassemia disease if both their parents have trait.

Special blood tests can help tell if you have beta 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, additional testing may be necessary. Your doctor or genetic counselor will help arrange this special DNA (gene) testing, if needed.

If you learn you have beta thalassemia trait, you can consider other testing options. You may want to have your *partner* tested. If your partner also has beta thalassemia trait, there will be a 25% chance that each of your children will be born with beta thalassemia disease. Blood testing will also identify other types of benign hemoglobin traits. If your partner is found to have hemoglobin E trait, there will be a 25% chance for hemoglobin E/ beta thalassemia (a disease similar to beta thalassemia) in each pregnancy. If your partner is found to have hemoglobin S trait (sickle cell trait), there will be a 25% chance for hemoglobin S/ beta thalassemia in each pregnancy. This is a type of sickle cell disease characterized by anemia, painful events, decreased resistance to infection, and other complications. There are tests that a woman can have *early in pregnancy* to find out if her baby is affected with one of these conditions. 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,

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Additional messages:

prepared by Jennifer Bojanowski, MS, Hematology Genetic Counselor, Children's Hospital Oakland, 04/98