Beta thalassemia is most common in people of Mediterranean, Middle Eastern, African, South Asian (Indian, Pakistani, etc.), Southeast Asian and Chinese descent.

People with beta thalassemia trait will usually experience no health problems other than a mild anemia.

You may have beta thalassemia trait and not know it.
Thalassemia is the name of a group of genetic blood disorders. Over two million people in the United States have the genetic trait for thalassemia. You may be one of them.

There are two very important reasons for you to find out if you have beta thalassemia trait:

First, when two people who both have beta thalassemia trait have a child, there is a one-in-four (25%) chance with each pregnancy that the child will be born with a serious blood disorder that requires lifelong blood transfusions and drug treatments.

Second, some doctors may mistake beta thalassemia trait for another condition and prescribe the wrong treatment for you.

Finding out if you have beta thalassemia trait is easy.

The first step to finding out if you have the beta thalassemia trait is to ask your doctor to look at the size of your red blood cells. This is shown by the Mean Corpuscular Volume (MCV) of your Complete Blood Count (CBC). Your doctor may already have a record of your CBC on file.

If your MCV reading is 80 or lower, and you are NOT iron deficient, you may have beta thalassemia trait. Additional testing, including a hemoglobin electrophoresis, quantitative hemoglobin A2 and quantitative hemoglobin F, will be necessary to determine if you have beta thalassemia trait. These tests can be ordered by your doctor.

To understand how thalassemia affects the human body, you must first understand a little about blood.

Blood carries oxygen from your lungs to other parts of your body. Oxygen is carried by a protein called hemoglobin found inside the red blood cells. Hemoglobin is made of two different kinds of proteins, called alpha and beta globins.

Beta globin is made by two genes, one passed on to the baby from each parent. Individuals who have one abnormal beta globin gene have beta thalassemia trait (also known as beta thalassemia minor).

A person with beta thalassemia trait (minor) simply carries the genetic trait for beta thalassemia and will usually experience no health problems other than a mild anemia.

Physicians often mistake the small red blood cells of the person with beta thalassemia minor as a sign of iron deficiency anemia and incorrectly prescribe iron supplements that will not help the anemia.

If you are tested for beta thalassemia trait and are found to be a trait carrier, your spouse should also be tested for beta thalassemia trait.

If both parents carry the beta thalassemia trait, there is a 25% chance with each pregnancy that their child will inherit two abnormal beta globin genes.

In its most severe form, this may cause beta thalassemia major or Cooley’s anemia, a severe blood disorder that causes a life-threatening anemia that requires regular blood transfusions and extensive ongoing medical care.

The inheritance of two abnormal beta globin genes may also cause beta thalassemia intermedia, a moderately severe anemia with significant health problems including bone deformities and enlargement of the spleen.

For more information about thalassemia, please contact: Cooley’s Anemia Foundation at (212) 279-8090 or info@thalassemia.org
You can also visit our web site at www.thalassemia.org

If you and your spouse both carry beta thalassemia trait and plan to have children, you should seek the advice of a genetic counselor.