

## WHAT DO YOUR RESULTS MEAN?

Sickle Cell Anemia, Sickle Cell Trait, and other hemoglobin variants can be identified by special laboratory test. These tests identify hemoglobin, which is the substance inside red blood cells. The function of the hemoglobin is to carry oxygen from the lungs to all parts of the body, and is also responsible for the red color of the blood.

Listed below are some of the common hemoglobin types.

### NORMAL

**AA- NORMAL ADULT HEMOGLOBIN.** There is no evidence of sickle cell anemia or sickle cell trait. The red blood cells are normal in shape and function.

### TRAITS

**AC- HEMOGLOBIN C TRAIT.** The inheritance of an **A** gene from one parent and **C** gene from the other parent. Persons with hemoglobin C trait are themselves healthy. There is no anemia, and there are no symptoms. Approximately 1 out of every 50 Black Americans have hemoglobin C trait.

**AD- HEMOGLOBIN AD.** The inheritance of an **A** gene from one parent and **D** gene from the other parent. This hemoglobin does not cause any clinical or hematological abnormalities in the heterozygote. One parent had to carry hemoglobin D gene. If this individual marries a person with an hemoglobin S gene the offspring may be a heterozygote for the hemoglobin SD often confused with hemoglobin SS.

**AF- (HEREDITARY PERSISTENCE OF FETAL HEMOGLOBIN – HPFH).**

**HPFH** is the persistence of fetal hemoglobin into adult life. In the heterozygous condition one parent had to have at least one gene for the **HPFH**. If the heterozygous individual marries there is the possibility that the offspring may inherit the Hb-HPFH. The heterozygotes are not anemic and there are no clinical or hematological abnormalities associated with this condition.

**AG- HEMOGLOBIN AG.** The inheritance of an **A** gene from one parent and **G** gene from the other parent. This hemoglobin does not cause any clinical or hematological abnormalities in the heterozygote. One parent must carry a hemoglobin G gene. If this individual marries a person with a hemoglobin S the offspring may be heterozygous for hemoglobin S and hemoglobin G.

**A+FAST.** The inheritance of an **A** gene from one parent and the **fast moving** hemoglobin from the other parent. In order that we may give a definitive diagnosis on the individual's screening we would need to draw a tube of whole blood to run an additional battery of test to identify the fast moving hemoglobin. In most cases the individual should not experience any health problems.

**AE- HEMOGLOBIN AE.** The inheritance of an **A** gene from one parent and **E** gene from the other parent. This hemoglobin does not cause any clinical or hematological abnormalities in the heterozygote. One parent had to have at least one Hb E gene. If this individual marries a person with an Hb S gene the offspring may be heterozygous for Hb S and Hb E.

**AS- SICKLE CELL TRAIT.** The inheritance of an **A** gene from one parent and an **S** gene from the other parent. There are usually no symptoms associated with sickle cell trait because the red blood cells are normal in shape and function. (Approximately 1 out of every 10 Black Americans (10%) have AS hemoglobin or sickle cell trait). However, the S gene can be passed on to future generations, which in combination with an S gene from another person can produce sickle anemia.

## MORE COMMON DISEASES

**SC- SICKLE CELL HEMOGLOBIN C DISEASE.** The inheritance of an **S** gene from one parent and a **C** gene from the other parent. Persons with SC hemoglobin sometimes experience the same symptoms as persons with sickle cell anemia, although the symptoms may not be as severe. Approximately 1 out of every 1,000 Black Americans have Sickle Cell Hemoglobin C Disease.

### SS- SICKLE CELL ANEMIA.

The inheritance of an **S** gene from each parent. The red blood cells are shaped like sickles and do not carry oxygen as well as they should. General symptoms may include severe anemia, fatigue, and pain in any parts of the body, leg ulcers or yellowing of the whites of the eyes. Swelling of the hands and feet in babies is also common. When these symptoms become very severe, a person is experiencing what is called a “pain episode”. Approximately 1 out of every 400 Black Americans have SS hemoglobin or sickle cell anemia.

**CC- HEMOGLOBIN C DISEASE** The inheritance of **C** gene from each parent. Symptoms may include mild anemia, mild jaundice, and an increased tendency to form gallstones. Painful crisis does not occur. Approximately 1 out of 10,000 Black Americans have CC hemoglobin or Hemoglobin C Disease.

## OTHERS COMMON TYPES OF HEMOGLOBINS

**THALASSEMIAS.** Is a genetic blood group disorders that affects the human body. People with Thalassemia do not produce enough alpha or beta. These are proteins of the hemoglobin and the red blood cells that do not form properly and cannot carry sufficient oxygen. All types of Thalassemias have similar symptoms but they can vary from mild to severe anemia. The Thalassemias are classified according to the chain of amino acid affected. The two principal types are:

**ALPHA THALASSEMIA.** It is found in people from Africa, Middle East, India, Southeast Asia, Southern China, and occasionally the people from the Mediterranean region. It is caused by the elimination of one or more genes of the alpha globin chain.

There are at least four forms of Alpha Thalassemia:

Silent Carrier, Alpha Thalassemia Trait, Hemoglobin H Disease, Hydrops Fetalis. In United States the more common form is:

### **ALPHA THALASSEMIA TRAIT/BARTS (INDICATOR FIRST DAYS OF NEWBORN'S LIFE)**

The inheritance of the normal **A** gene from one parent and the **Alpha Thalassemia** gene from the other parent. Individuals with Alpha Thalassemia Trait (Barts) can be misdiagnosed as being iron deficient, because some of the red blood cells are small in size and pale in color. Alpha Thalassemia Trait is not a disease.

**BETA THALASSEMIA.** It is found in people of Mediterranean descent, and also found in the Arabian Peninsula, Africa, Iran, Southeast Asia and Southern China. It is caused by the mutation of the beta globin chain.

There are three types of Beta Thalassemias that also range from mild to severe in their effect on the body.

Thalassemia Minor or Thalassemia Trait, Thalassemia Intermedia, Thalassemia Major or Cooley's anemia.

## OTHER FORMS OF THALASSEMIA

There are other related disorders that occur when the gene for Alpha or Beta Thalassemia combines with an abnormal or mutant gene.

<b>Hb S/ <math>\beta</math>+ Thal</b>	<b>HEMOGLOBIN SICKLE BETA + THALASSEMIA</b>
<b>Hb C/<math>\beta</math> + Thal</b>	<b>HEMOGLOBIN C BETA + THALASSEMIA</b>
<b>Hb E/<math>\beta</math> + Thal</b>	<b>HEMOGLOBIN E BETA + THALASSEMIA</b>

