

# SICKLE CELL TRAIT AND DISEASE AND BONE MARROW TRANSPLANTS

## SICKLE CELL DISEASE

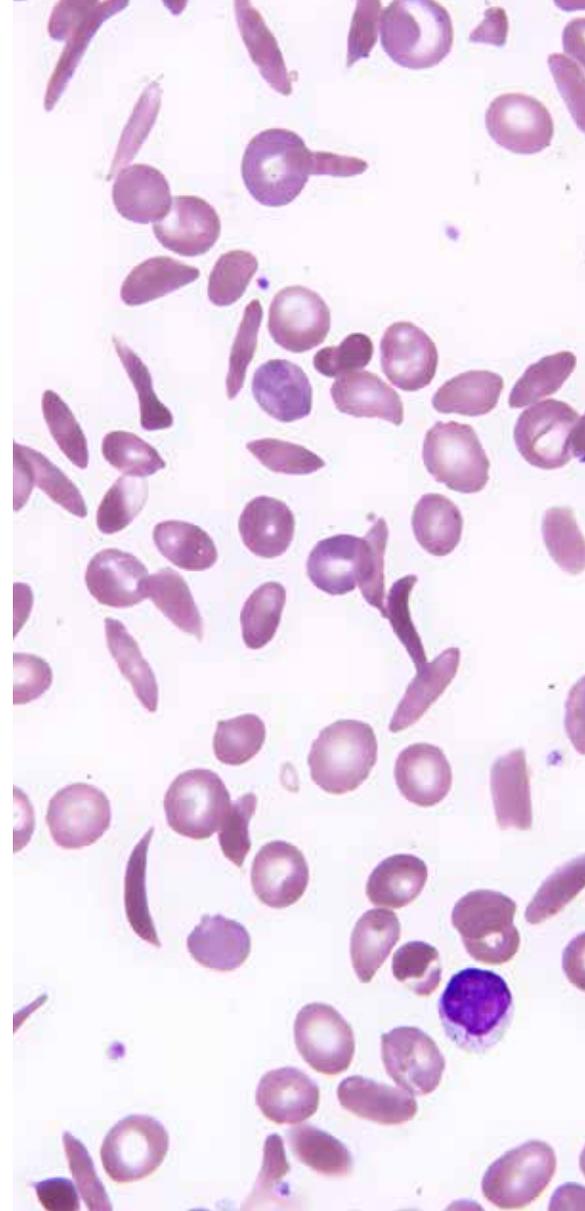
Sickle cell disease (also known as sickle cell anemia) is a genetic condition in which red blood cells take the shape of a sickle and are rigid and unbending (unlike regular red blood cells). The lifespan of these abnormal red blood cells is about one-tenth of regular blood cells, which leads to chronic anemia. Patients with sickle cell disease can suffer from many medical problems, including infections, kidney disease, neurologic complications, and acute pain crises. Acute pain crises occur when the rigid, sickle-shaped cells clog the flow of blood in small vessels, particularly within bones.

Patients with sickle cell disease have two abnormal (mutated) copies of a particular hemoglobin gene. The mutated copy is called hemoglobin “S” and the normal copy is called hemoglobin “A”. One copy of the gene is inherited from the mother, and one is inherited from the father. When both of the inherited genes are mutated (hemoglobin “SS”), the hemoglobin protein will be structurally abnormal and this results in sickled red blood cells.

All newborns in the United States are screened for sickle cell disease at birth. Patients with sickle cell disease will need close clinical monitoring throughout life. Therapies include hydroxyurea, transfusions and red blood cell exchange, as well as treatments targeted towards clinical manifestations - for example, antibiotics for infection or pain medications for acute crises. Some patients may undergo bone marrow transplantation, which can be curative. Gene therapy is also an area of active investigation.

## SICKLE CELL TRAIT

When only one of the copies of the hemoglobin gene is mutated (hemoglobin “AS”), a patient is said to have sickle cell *trait*. Unlike patients with sickle cell disease, patients with sickle cell trait usually have no clinical manifestations. Approximately 3 million people in the United States have sickle cell trait. Sickle cell trait is most prevalent in African-Americans, affecting approximately 10% of the population.



## UNDER THE MICROSCOPE

This pathology slide is a peripheral blood smear from a patient with sickle cell disease demonstrating many elongated red cells with tapered ends, typical of “sickled” cells. In addition to these shapes, these sickled cells are rigid, which makes them block and clog small vessels.

# LABORATORY TESTS

## SICKLE CELL TRAIT AND DISEASE

Sickle cell disease is a chronic illness. This means that in addition to the tests that help detect the disease, there are also routine follow up tests and specialized tests. Patients with sickle cell trait do not need monitoring, but if a pregnant mother is found to have sickle cell trait the father will also be tested for sickle cell trait to determine if the child is at risk for sickle cell disease (see “Gene Inheritance” in sidebar).

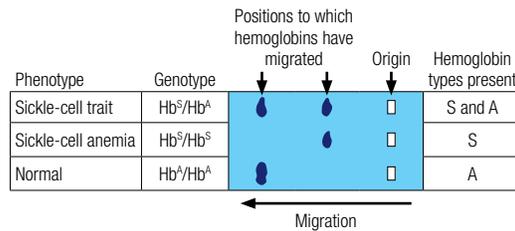
### DETECTION TESTS

#### Hemoglobin Electrophoresis:

This blood test can identify different abnormal hemoglobin species based on how quickly they move on a gel (right).

Normal hemoglobin (“A”) runs very fast, whereas hemoglobin “S” and other types

of abnormal hemoglobins run slower. In a patient with sickle cell trait, about half of the hemoglobin will be detected at the “A” position on the gel, and half will be at the “S” position on the gel. In patients with sickle cell disease, nearly all of the hemoglobin will be at the “S” position.



**High Pressure Liquid Chromatography (HPLC):** This is a more sensitive blood test to measure the identity and quantity of different hemoglobin species. This test is used as an adjunct to hemoglobin electrophoresis, mostly commonly to measure additional hemoglobin species that are normally present at low levels in all patients (hemoglobin A2 and F).

**Hemoglobin Solubility:** This blood test is a qualitative test that identifies abnormal, insoluble hemoglobin. Insoluble hemoglobin will cause red blood cells to sickle. This test is important because it confirms that the hemoglobin detected at the “S” position on the hemoglobin electrophoresis gel is indeed one that will cause sickling.

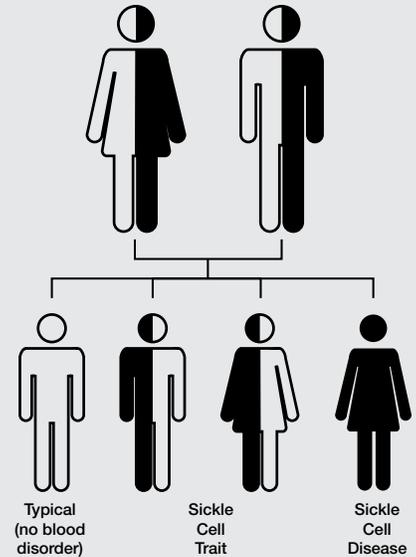
**Sodium Metabisulfite:** This blood test measures sickling of red blood cells by exposing them to a chemical and visually evaluating for sickled cells with a microscope. This test is an alternative to the hemoglobin solubility test.

### ROUTINE TESTS

**Hemoglobin electrophoresis plus HPLC** is performed routinely in patients with sickle cell disease to measure hemoglobin levels, particularly the amount of hemoglobin “S” as well as the amount of hemoglobin “A”. The hemoglobin “A” that is detected comes from red blood cells that have been transfused into the patient from a normal donor. The amount of hemoglobin “S” in the blood can help determine when treatment is needed, or how effective treatment has been.

### SPECIALIZED TESTS

**DNA Sequencing:** Although not necessary for most patients with sickle cell disease, sequencing of DNA can be used to more precisely identify gene mutations in cases that are difficult to diagnose using the tests listed above.



### GENE INHERITANCE

If one parent has sickle cell trait, there is a 25% chance that their child will have sickle cell trait.

If both parents have sickle cell trait, there is a 50% chance their child will have sickle cell trait and a 25% chance their child will have sickle cell disease.

# LABORATORY TESTS

## BLOOD TRANSFUSIONS

**ABO Testing:** This test is performed on the blood of both patients and donors, and determines if A, B or neither antigen is present on the surface of red blood cells (note: this “A” has nothing to do with hemoglobin “A”). This is important because patients have naturally-occurring antibodies against antigens that aren’t on their own red blood cells. For example, if a patient is blood type B, and receives blood from a type A donor, the patient’s naturally-occurring anti-A antibodies will destroy the donated red blood cells.

**Rh Testing:** Some patients have Rh antigen on their cell surface (Rh-positive), and some do not (Rh-negative). Rh-negative patients will develop antibodies to Rh antigen if they are exposed to cells with Rh antigen, and these antibodies will attack Rh-positive cells on any subsequent transfusion. Therefore, Rh-negative patients should only receive transfusions from Rh-negative donors.

**Antibody Screen:** This antibody screen tests the recipient’s blood for antibodies to other types of antigens (besides A, B and Rh) that can occur on a donor’s red blood cells. If the screen detects antibodies to one or more antigens, it will be necessary to find units from donors who do not have these antigens on the surface of their red blood cells.

**Crossmatch:** This test is performed right before a transfusion, and it measures if the recipient’s blood is compatible with the unit of donor blood that is about to be transfused. This is often done by mixing a small portion of the donated blood with the recipient’s serum in the lab and looking for formation of red blood cell aggregates, which indicates that the recipient has antibodies against the donor’s red blood cells.

### Blood Type Compatibility:

		Donor's Blood Type							
		0-	0+	B-	B+	A-	A+	AB-	AB+
Patient's Blood Type	AB+	✓	✓	✓	✓	✓	✓	✓	✓
	AB-	✓		✓		✓		✓	
	A+	✓	✓			✓	✓		
	A-	✓				✓			
	B+	✓	✓	✓	✓				
	B-	✓		✓					
	0+	✓	✓						
	0-	✓							

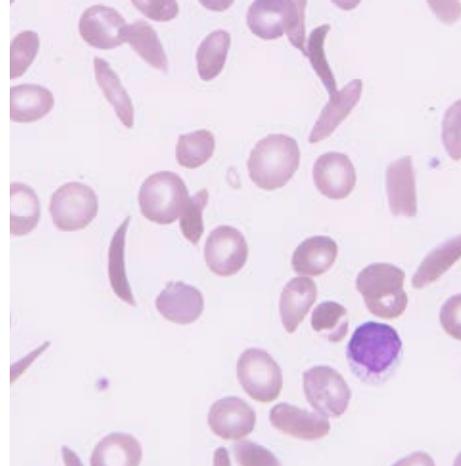
## BONE MARROW TRANSPLANTS

**Human Leukocyte Antigen (HLA):** HLA genes are inherited and the gene products (antigens) are expressed on the surface of cells. Donors are chosen for bone marrow transplantation based on how closely their HLA genes match those of the recipient. Strong matches promote engraftment and decrease the chance and severity of graft-versus-host disease. Patients have a 1 in 4 chance of having identical HLA antigens as their sibling, but it is possible to find strong matches in unrelated people as well.

## ASK YOUR DOCTOR

- How will the lab test results impact my treatment plan?
- How often will I need to get labs in order to check the status of my disease progression?
- What other lab values are we looking at to monitor my health?

# SICKLE CELL TRAIT AND DISEASE AND BONE MARROW TRANSPLANTS: MEET MALIK



Malik (left) was born with sickle cell disease. His mother, Belinda, was devastated when she heard the results of his routine blood tests indicating he had Sickle Cell Disease. The journey that Malik and his family endured through the process of managing his diagnosis was difficult, but thanks to a bone marrow transplant from his brother Michael (right), Malik no longer has sickle cell disease.

**To learn more and to watch a video about Malik, go to [www.ascp.org/patients](http://www.ascp.org/patients).**

*“For every visit that we went to at the hospital, we were sitting with bated breath waiting for the lab results. The results determined the course of every day and the actions that the doctors would take.”*

—Belinda, Malik’s mother

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