How Is Sickle Cell Disease Diagnosed?

Screening Tests
People who do not know whether they make sickle hemoglobin (hemoglobin S) or another abnormal hemoglobin (such as C, β thalassemia, E) can find out by having their blood tested. This way, they can learn whether they carry a gene (i.e., have the trait) for an abnormal hemoglobin that they could pass on to a child.

When each parent has this information, he or she can be better informed about the chances of having a child with some type of sickle cell disease (SCD), such as hemoglobin SS, SC, Sβ thalassemia, or others.

Newborn Screening
When a child has SCD, it is very important to diagnose it early to better prevent complications.

Every state in the United States, the District of Columbia, and the U.S. territories require that every baby is tested for SCD as part of a newborn screening program.

In newborn screening programs, blood from a heel prick is collected in "spots" on a special paper. The hemoglobin from this blood is then analyzed in special labs. Newborn screening results are sent to the doctor who ordered the test and to the child's primary doctor.

If a baby is found to have SCD, health providers from a special follow-up newborn screening group contact the family directly to make sure that the parents know the results. The child is always retested to be sure that the diagnosis is correct.

Newborn screening programs also find out whether the baby has an abnormal hemoglobin trait. If so, parents are informed, and counseling is offered.
Remember that when a child has sickle cell trait or SCD, a future sibling, or the child's own future child, may be at risk. These possibilities should be discussed with the primary care doctor, a blood specialist called a hematologist, and/or a genetics counselor.

Prenatal Screening
Doctors can also diagnose SCD before a baby is born. This is done using a sample of amniotic fluid, the liquid in the sac surrounding a growing embryo, or tissue taken from the placenta, the organ that attaches the umbilical cord to the mother's womb.

Testing before birth can be done as early as 8–10 weeks into the pregnancy. This testing looks for the sickle hemoglobin gene rather than the abnormal hemoglobin.