

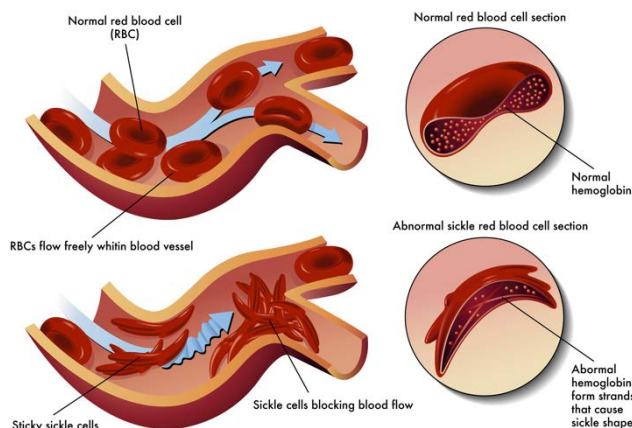
## SICKLE CELL ANEMIA

### SYMPTOMS

Sickle cell anemia, also called sickle cell disease, is a disorder that affects the red blood cells (RBCs).

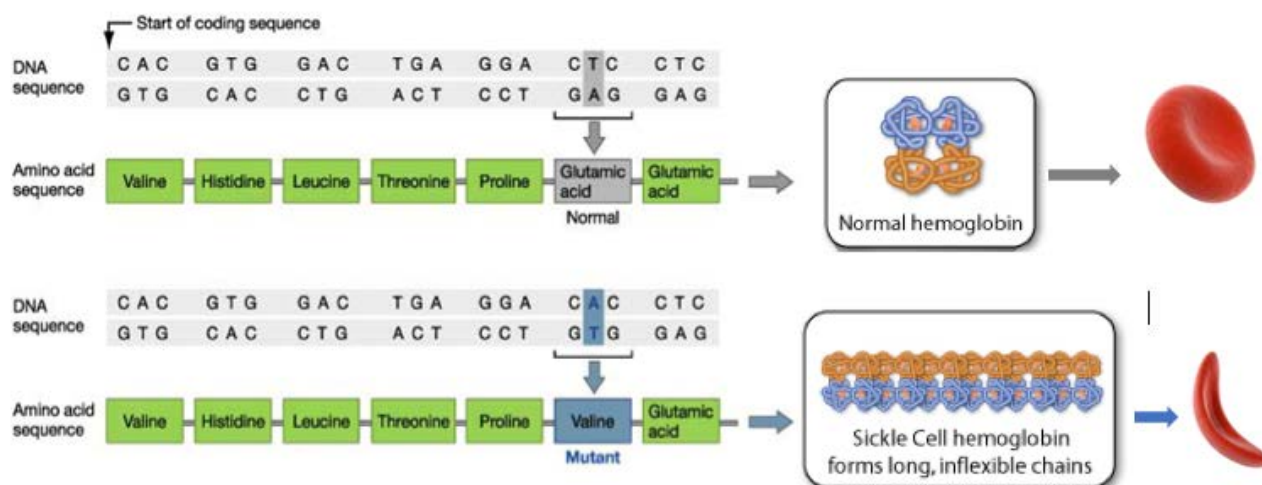
Individuals with this disease produce abnormal hemoglobin, the protein in red blood cells that carries oxygen. Instead of having the streamlined shape of normal RBCs, cells with abnormal hemoglobin flatten and bend into a crescent shape. As a result, they do not move smoothly through small blood vessels but often

clump up and block flow, preventing tissues from getting adequate oxygen. This causes fatigue, severe pain, and often damage to vital organs like the lungs, spleen, kidneys and brain. Other symptoms include frequent infections, delayed growth, and vision problems. In countries where modern medicine is available, children with the disease can sometimes be helped with bone marrow transplants, and symptoms can be medically managed. However, in countries without ready access to modern medicine, 50-80% of individuals with sickle cell disease die in childhood.



### CAUSE

Normal hemoglobin is produced by the HbA gene. The sickle cell allele, HbS, results when an adenine is substituted for a thymine at a critical place in the HbA gene. As a result of this single nucleotide mutation, the hemoglobin produced has a completely different shape than the normal hemoglobin produced by the HbA allele. This in turns causes RBCs containing the abnormal hemoglobin to be misshapen.



Name: \_\_\_\_\_ Period: \_\_\_\_\_ Date: \_\_\_\_\_

## INHERITANCE

Individuals who have two normal alleles (**HbA, HbA**) are *normal*. Their red blood cells are normally shaped.

Individuals with two sickle cell alleles (**HbS, HbS**) have *sickle cell disease (SCD)*. Their blood cells are misshapen.

Individuals who are heterozygous (**HbA, HbS**) are said to have “*sickle cell trait*” (*SCT*). Some of their red blood cells are normal, but some are sickled. They do not typically have severe symptoms of the disease, but can experience symptoms under low oxygen conditions (for example, in muscles during high exertion).

