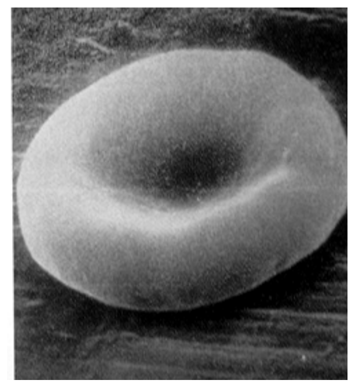
Name ____ Date ______________ Period _______

Genetics of Sickle Cell Anemia

B I O L O G Y

Background: For some genes, neither allele is completely dominant, so a heterozygous individual differs from either type of **homozygous** individual. (A heterozygous individual has two different alleles for the same gene, whereas a homozygous individual has two identical alleles for the same gene. For example individuals that are either **AA** or **aa** are homozygous, whereas individuals that are **Aa** are heterozygous.)

The gene for hemoglobin is an example. One allele codes for normal hemoglobin, while another allele codes for altered hemoglobin, called sickle cell hemoglobin. When a person is homozygous for this sickle cell allele, this causes a serious disease called sickle cell anemia. The sickle cell hemoglobin tends to cause the red blood cells to assume a sickle shape, in contrast to the normal disk-shaped red blood cell shown on the left in the figure below. What problems might be caused by the sickle-shaped red blood cells?



A person who is heterozygous for the sickle cell allele often has very few or no symptoms of sickle cell anemia. In addition, people who are heterozygous for the sickle cell allele are more resistant to malaria, an infection of the red blood cells which is transmitted by mosquitoes in many tropical countries. Thus, in areas where malaria is widespread, people who are heterozygous for the sickle cell allele are less likely to become ill and die. Because of this advantage, the sickle cell allele became relatively common in regions like West Africa where malaria is common. Since African-Americans are descended from populations in which the sickle cell allele was relatively common, African-Americans have relatively high rates of the sickle cell allele (approximately 8% are heterozygous for this allele and 0.16% are homozygous).

Suppose that a person who is heterozygous for the sickle cell allele (**Ss**) marries a person who is also heterozygous for this allele (**Ss**). Draw a Punnett Square to show the expected genetic makeup of their children, and then answer the following questions.

1. What fraction of their children will suffer from sickle cell anemia?

2. What fraction of their children will inherit the sickle cell allele, but will not suffer from sickle cell anemia? (These children will be resistant to malaria.)

3. The presence of Malaria in many countries has been eradicated and even in parts of Africa, the incidence of Malaria has been reduced substantially. Without endemic malaria from Africa, the sickle cell mutation is purely disadvantageous. What do you think will happen to the percentage of people carrying the Sickle Cell gene over time under the pressures of natural selection?

4. Sickle Cell disease is caused by the replacement of a single nucleotide on the β-globin gene found on the short arm of chromosome 11. What do we call this type of mutation?

5. Complete the following genetic crosses. Show genotypes of parents, construct a punnett square, and give the genotypic and phenotypic ratios of the offspring.

a. Normal male x carrier female

b. Sickle-cell male x normal female

c. Sickle-cell male and carrier female