Sickle cell anemia is an autosomal recessive genetic disorder that affects thousands of people in the United States and millions worldwide. Sickle cell anemia commonly occurs in groups whose ancestors came from Africa, as well as South America, Cuba, Central America, Saudi Arabia, India, and the Mediterranean.

Sickle cell anemia is caused by a change in the hemoglobin protein in red blood cells. Sickle cell anemia results in paleness, fatigue, shortness of breath, and increased heart rate due to a deficiency in the oxygen-carrying component of the blood. When oxygen levels are low in an affected individual, the red blood cells become deformed into a curved, sickle shape. People with sickle cell anemia can experience swelling, pain, infection, and organ damage.

All individuals have two alleles for the gene that codes for the hemoglobin protein (Hb). Individuals with two \( \text{Hb A} \) alleles have normal, round red blood cells. Heterozygous individuals, with one \( \text{Hb A} \) allele and one \( \text{Hb S} \) allele, do not experience symptoms of the disease, but they may produce some sickle-shaped red blood cells. Individuals with two \( \text{Hb S} \) alleles have sickle cell anemia.

The diagrams to the right represent some of the steps in the formation of hemoglobin in two individuals, Y and Z. In these diagrams, only a small part of the hemoglobin gene sequence is represented.

Individual Y has two \( \text{Hb A} \) alleles and therefore produces normal red blood cells. Individual Z has two \( \text{Hb S} \) alleles and therefore produces sickle-shaped red blood cells.
Individual Y
(normal hemoglobin)

ACT CCT GAG GAG
TGA GGA CTC CTC

1

ACU CCU GAG GAG

2

Immature 
red blood cell
DNA

ACU CCU GAG GAG

mRNA
(made from 
bottom DNA 
strand)

1

ACU CCU GUG GAG

2

Amino 
acid 
sequence

Threonine – Proline – Glutamic acid – Glutamic acid

3

Mature 
red blood cells

Individual Z
(sickle cell hemoglobin)

ACT CCT GTG GAG
TGA GGA CAC CTC

1

ACU CCU GUG GAG

2

Amino 
acid 
sequence

Threonine – Proline – Valine – Glutamic acid

3