Gene mutation: sickle-cell shape

Key words

codon
polypeptide chain

Sickle-cell anemia

- Sickle-cell anemia is a disease of the red blood cells caused by an error in one triplet of one of the *polypeptide chains* in beta (β) hemoglobin.
- Sufferers from sickle-cell anemia have red blood cells that are irregular shapes, often crescent moon or sickle shapes, which cannot carry oxygen as well as normal blood cells. This leads to a general lack of energy in the sufferer. Their abnormal shape also means that they tend to get stuck in small blood vessels, leading to painful clots.

Glutamine to valine

- The sickle-cell mutation involves a single triplet substitution from CTC (Cytosine/Thymine/Cytosine) to CAC (Cytosine/Adenine/Cytosine). This change leads to the amino acid valine (VAL) being added to the polypeptide chain in place of glutamine (GLU). This change leads, in turn, to other problems with the three-dimensional shape of the hemoglobin formed, and therefore with its functionality.
- Since the gene mutation is a base substitution, only one of the *codons*, and so one of the amino acids, is affected. In certain areas of the world, sickle-cell anemia offers a degree of protection against the malaria parasite.

Sickle-cell mutation