

## UNIT 3: GENETICS

### SICKLE CELL ANEMIA

- Caused by a mutation of the gene that codes for the alpha-globin polypeptide haemoglobin.
- Most humans have the allele  $Hb^A$ .
- A **missense substitution** that converts the 6<sup>th</sup> codon from GAG to GTG, creating a new codon for a different amino acid →  $Hb^S$ .
- When  $Hb^S$  is synthesised a valine amino acid, instead of glutamic acid, is added in the polypeptide changing its shape.
- This causes haemoglobin molecules to stick together giving red blood cells a sickle shape.
- They become trapped in capillaries reducing blood flow.
- The life of the red blood cells are reduced and they cannot be replaced quick enough.
- This results in chronic developing.



The majority of people with sickle cell anemia.



The  $Hb^S$  allele helps protect against malaria. This is why it has remained in certain populations.

### WHAT IS A GENOME?

- A genome is the entire base sequence of the DNA in an organism.
  - Animals = DNA in the chromosomes + mitochondrial DNA.
  - Plants = DNA in the chromosomes + mitochondrial DNA + chloroplast DNA.
  - Prokaryotes = DNA in circular chromosomes + any plasmids present.
- The **Human Genome Project** sequenced the entire base sequence of the human genome.
- It had a number of important outcomes:
  - Identifying and locating the position of all human genes.
  - Determining protein structures and functions.
  - Discovering evidence for evolutionary relationships and human origins.
  - Finding mutations and genes that cause disease.
  - Developing personalised medicine based on gene sequences.
  - Improving base sequencing techniques, making it quicker and more accurate.



Sequencing is necessary to determine the base sequence of DNA.



There are many ways to sequence DNA. The Human Genome Project used a method called Sanger sequencing.

### CHECKPOINT

#### Multiple Choice

- Various forms of a gene are called:
  - Chromosomes
  - Alleles
  - Mutations
  - Genes
- Which of the following is a significant type of mutation?
  - Base substitution
  - Base multiplication
  - Base insertion
  - Base deletion

#### Applications

- Using the table on the previous page, discuss the hypothesis that the larger the number of genes an organism has the more complex it is.

#### Structured Answer

- Explain the causes and effects of sickle cell anemia. [3]
- Explain the significance of the Human Genome Project. [4]

# Ib Biology Revision Guide

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