

## 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 a genome. The Human Genome Project used a method called Sanger sequencing.

### CHECKPOINT

#### Multiple Choice

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

#### 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.
- Explain the significance of the Human Genome Project.

# Ib Biology Revision Guide

**ML Yell**



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