

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 6th 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 mutation of 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 sequencing.
 - 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 genomes. Long-read sequencing is used for the Human Genome Project.

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 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

Mary Jones, Matthew Parkin



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