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Principles of Genetics

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Section no. (1)

Define the following terms

- **gene** is a specific sequence of nucleotide bases, whose sequences carry the information required for constructing proteins, which provide the structural components of cells and tissues as well as enzymes for essential biochemical reactions. The human genome is estimated to comprise more than 30,000 genes

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- DNA is a stable polynucleotide. The double-helix structure of the DNA molecule in terms of: the components of DNA nucleotides; the sugar-phosphate backbone; specific base pairing and hydrogen bonding between polynucleotide strands (only simple diagrams of DNA structure are needed; structural formulae are not required). Explain how the structure of DNA is related to its functions.

Mutations •

New forms of alleles arise from changes • (mutations) in existing alleles

Mitosis •

Mitosis increases cell number in growth and • tissue repair and in asexual reproduction. During mitosis DNA replicates in the parent cell, which divides to produce two new cells, each containing an exact copy of the DNA of the parent cell. Candidates should be able to name and explain the stages of mitosis and recognise each stage from diagrams and photographs.

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Meiosis •

During meiosis, cells containing pairs of • homologous chromosomes divide to produce gametes containing one chromosome from each homologous pair. In meiosis the number of chromosomes is reduced from the diploid number (2n) to the haploid number (n). (Details of meiosis (.not required

Gene Therapy and Cystic Fibrosis •

In gene therapy healthy genes may be •
cloned and used to replace defective
genes. In cystic fibrosis the
transmembrane regulator protein, CFTR,
is defective. A mutant of the gene that
produces CFTR results in CFTR with one
missing amino acid. The symptoms of
cystic fibrosis related to the malfunctioning
of CFTR.