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AS Module 2
GENES AND GENETIC ENGINEERING

Introduction

This module explores some of the ways in which the understanding of genes that has been developed over the past half century is being applied. Elucidation of the structure and functions of nucleic acids has helped to explain how genes incorporate coded information which determines the nature of organisms, and how, during the cell cycle and sexual reproduction, genetic information is copied and passed on. The development of techniques for manipulating and transferring genes has opened up opportunities to use microorganisms to synthesise biological compounds on a large scale, to enhance food production and to introduce treatments for human genetic disorders. Candidates are expected to understand the basis of these developments and to explore the practical and ethical issues associated with them.

This module includes part of the knowledge and understanding specified in the mandatory subject criteria for Advanced Subsidiary Biology, as set out in the document issued by QCA in June 1999. It covers sections 3.14, 3.15 and part of 3.16 of that document.

11.1 The genetic code

The gene

Genes are sections of DNA which contain coded information that determines the nature and development of organisms.
A gene can exist in different forms called alleles which are positioned in the same relative position (locus) on homologous chromosomes.

Structure of DNA

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).

Replication of DNA

The semi-conservative mechanism of DNA replication, including the role of DNA polymerase.

The genetic code

How DNA acts as a genetic code by controlling the sequence of amino acids in a polypeptide.

Codons for amino acids are triplets of nucleotide bases.

Role of nucleic acids in protein and enzyme synthesis

The structure of RNA.

The production of mRNA in transcription, and the role of RNA polymerase.

The roles of ribosomes, mRNA and its codons, and tRNA and its anticodons in translation.

Candidates should be able to explain:

- how the structures of DNA and RNA are related to their functions;
- the relationship between genes, proteins and enzymes.

Mutation

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

Gene mutation as the result of a change in the sequence of bases in DNA, to include addition, deletion and substitution.

Mutations occur naturally at random. High energy radiation, high energy particles and some chemicals are mutagenic agents.

Candidates should be able to explain:

- how a change in the sequence of bases in an individual gene may result in a change in the amino acid sequence in the polypeptide;
- how the resulting change in polypeptide structure may alter the way the protein functions;
- how, as a result of mutation, enzymes may function less efficiently or not at all, causing a metabolic block to occur in a metabolic pathway.

11.2 The cell cycle

Mitosis

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. Mitosis increases cell number in this way in growth and tissue repair and in asexual reproduction.

Candidates should be able to name and explain the stages of mitosis and recognise each stage from diagrams and photographs.

Applications of cloning

Genetically identical organisms (clones) can be produced by using vegetative propagation, and by the splitting of embryos.

Given appropriate information, candidates should be able to explain the principles involved in:

- producing crops by vegetative propagation;
- the cloning of animals by splitting apart the cells of developing embryos.

11.3 Sexual reproduction

Gametes and fertilisation

Sexual reproduction involves gamete formation and fertilisation. In sexual reproduction DNA from one generation is passed to the next by gametes.

Differences between male and female gametes in terms of structure, size, number produced and mobility.

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.)

Importance of meiosis	When gametes fuse at fertilisation to form a zygote the diploid number is restored. This enables a constant chromosome number to be maintained from generation to generation. **Candidates should be able to** interpret life cycles of organisms in terms of mitosis, meiosis, fertilisation and chromosome number.

11.4 Applications of gene technology

Principles of genetic Engineering	In genetic engineering, genes are taken from one organism and inserted into another. The use of restriction endonuclease enzymes to extract the relevant section of DNA. The use of ligase enzyme to join this DNA into the DNA of another organism.
The polymerase chain reaction	The process of DNA replication can be made to occur artificially and repeatedly in a laboratory process called the polymerase chain reaction (PCR). The use of PCR, radioactive labelling and electrophoresis to determine the sequence of nucleotides in DNA.
Genetically engineered microorganisms	Microorganisms are widely used as recipient cells during gene transfer. Plasmids are often used as vectors to incorporate selected genes into bacterial cells. Rapid reproduction of microorganisms enables a transferred gene to be cloned, producing many copies of the gene.
Genetic markers	Genetic markers in plasmids, such as genes which confer antibiotic resistance, and replica plating may be used to detect the bacterial cells that contain genetically engineered plasmids.
Large scale culturing	Bacteria containing the transferred gene can be cultured on a large scale in industrial fermenters. Useful substances produced by using genetically engineered microorganisms include antibiotics, hormones and enzymes. (Details of manufacturing processes 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. Techniques that might possibly be used to introduce healthy CFTR genes into lung epithelial cells include: • use of a harmless virus into which the CFTR gene has been inserted; • wrapping the gene in lipid molecules that can pass through the membranes of lung cells.

Genetically modified animals	How animals can be genetically engineered to produce substances useful in treating human diseases, as exemplified by genetically engineering sheep to produce alpha-1-antitrypsin which is used to treat emphysema and cystic fibrosis.
Evaluation of genetic engineering	**Candidates should be able to** evaluate the ethical, social and economic issues involved in the use of genetic engineering in medicine and in food production.