WJEC (Wales) Biology A-level

Topic 4.5 - Application of Reproduction and Genetics

Definitions and Concepts

Anopheles gambiae - The vector of malaria, otherwise known as the mosquito. It has rapidly evolved resistance to many insecticides.

Clones - The genetically identical offspring produced as a result of cloning.

Cloning - A method of producing genetically identical offspring by asexual reproduction.

Complementary DNA (cDNA) - A single strand of DNA complementary to the mRNA template strand.

Cystic fibrosis - A genetic disorder characterised by the production of thick, sticky mucus that causes lung infections and scarring.

DNA ladder - A set of DNA fragments of known size (standards) used to estimate the fragment lengths of a molecule run on a gel during electrophoresis.

DNA ligase - An enzyme that joins the sugar-phosphate backbone of two DNA segments.

DNA polymerase - An enzyme that synthesises a double-stranded molecule of DNA from a single template strand using complementary nucleotides.

Drisapersen - An experimental drug which aims to treat DMD by exon skipping. It introduces a 'molecular patch' over the mutated exon, enabling the gene to be read. A shorter, more functional type of dystrophin is synthesised.

Duchenne muscular dystrophy (DMD) - An X-linked recessive condition that is characterised by muscle degeneration and weakness. It is caused by one or more mutations in the dystrophin gene that prevent the production of dystrophin.

Exon - A region of DNA that codes for an amino acid sequence.

Functional mRNA - The final mRNA product that has had introns removed, as well as having undergone other post-transcriptional changes.

Gel electrophoresis - A technique that separates nucleic acid fragments or proteins by size using electric current.

Gene therapy - A therapeutic technique in which a faulty allele is replaced with a functional allele in order to treat or prevent disease.

Genetically modified (GM) organism - An organism that has had its genome altered.

Genetic counselling - A service that provides information and advice to people affected by or at risk of genetic diseases. This helps individuals and families to make informed decisions.

Genetic engineering - The modification of the genome of an organism by the insertion of a desired gene from another organism. This enables the formation of organisms with beneficial characteristics.

Genetic fingerprinting - A technique used to genetically identify an organism. It has applications in forensics.

Genetic screening - Testing individuals for certain faulty alleles. This can be used to detect disorders such as cystic fibrosis, Huntington's disease and thalassemia.

Genomics - A field of biology that focuses on the evolution, structure, function and mapping of genomes.

Germ line therapy - A type of gene therapy in which a faulty allele is replaced with a functional allele in germ cells or a very early embryo. The effects of this are permanent and can be inherited.

Human Genome Project - An international research project involving thousands of scientists which used Sanger sequencing to successfully map the entire human genome.

Huntington's disease - A genetic disorder characterised by the progressive degeneration of nerve cells in the brain, reducing the affected individual's ability to think, talk and move.

Intron - A non-coding sequence of DNA.

Malaria - A disease caused by the protoctista *Plasmodium* that lives within two hosts, mosquitoes (*Anopheles gambiae*) and humans. It causes recurrent episodes of fever and can be fatal.

Marker gene - An additional gene inserted into a plasmid that is used to aid in the identification of host cells that have taken up the desired gene. Marker genes are easily recognisable e.g. provide antibiotic resistance.

Next-generation sequencing (NGS) - The method of DNA sequencing used in the 100K Genome Project. It is faster, cheaper and more accessible than Sanger sequencing.

Personalised medicine - A form of medical care that enables doctors to provide healthcare customised to an individual's genotype.

Plasmodium sp. - The parasite that causes malaria. It has developed multi-drug resistance.

Polymerase Chain Reaction (PCR) - An *in vitro* technique used to rapidly amplify fragments of DNA.

Post-transcriptional processing - A set of biological processes, including the removal of introns, that modify pre-mRNA to produce functional mRNA following transcription.

Pre-mRNA - The product of transcription before any post-transcriptional regulation.

Primers - Short nucleotide sequences, complementary to one end of each of the DNA fragments.

Recognition sequences - Specific base sequences of DNA that restriction enzymes cut.

Recombinant DNA - A combination of DNA from two different organisms.

Recombinant DNA technology - The process by which segments of DNA are transferred from one organism to another.

Restriction endonucleases - Enzymes that cut DNA molecules at recognition sequences creating sticky ends.

Reverse transcriptase - An enzyme that synthesises DNA from RNA.

Sanger sequencing - The method of DNA sequencing used in the Human Genome Project that involves the formation of DNA fragments of varying lengths. This process takes a long time.

Short tandem repeats (STRs) - Sections of repeated nucleotides within introns that produce variation in individuals.

Somatic cell therapy - A type of gene therapy in which a faulty allele is replaced with a functional allele in affected somatic cells. The effects of this are temporary and cannot be inherited.

Stem cells - Cells that are unspecialised and retain the ability to differentiate into a range of cell types.

Sticky ends - The staggered cut formed by restriction endonucleases in double-stranded DNA.

Taq DNA polymerase - A thermally stable enzyme that synthesises a double-stranded molecule of DNA from a single template strand using complementary nucleotides.

Thalassaemia - A group of genetic disorders that result in reduced haemoglobin production.

Thermocycler - A machine controlled by a computer that varies temperatures at predetermined time intervals.

Tissue engineering - An extension of gene therapy that aims to replace, repair or improve biological function by replacing organs and tissues.

Transformation - The reinsertion of plasmids back into bacterial cells to form transgenic bacteria. This involves mixing the plasmids and bacterial cells.

Vector - A carrier used to transfer a gene from one organism to another e.g. plasmid.

100K Genome Project - A UK Government project that aims to study variation in the human genome amongst 100,000 UK citizens. It uses Next Generation Sequencers (NGS).