MEGA LECTURE

Definitions and Concepts for AQA Biology A-level

Topic 8 - The Control of Gene Expression

Acetylation: The addition of acetyl groups to histones. Acetylation activates the gene by making it more accessible to transcription factors.

Addition: A form of gene mutation in which one or more nucleotide bases are inserted into a DNA sequence. This may result in a frameshift to the right.

Benign: Describes a tumour that is non-cancerous. Such tumours grow slowly, are enclosed in a capsule and remain at the site of origin. They can usually be removed by surgery.

Cancer: A non-communicable disease resulting from tumour cells that metastasise.

Cellular proteome: The proteins expressed in a given type of cell.

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

Complete proteome: All of the proteins coded for by the genome.

Deletion: A form of gene mutation in which one or more nucleotide bases are removed from a DNA sequence. This may result in a frameshift to the left.

Differentiation: A process in which cells become specialised for function.

DNA hybridisation: The process by which a single-stranded segment of DNA is combined with a complementary fragment of DNA or RNA.

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.

DNA probe: A short, single-stranded segment of DNA that can be fluorescently or radioactively labelled. DNA probes are used to locate specific alleles of genes.

DNA sequencing: Determining the entire DNA nucleotide base sequence of an organism.

Duplication: A form of gene mutation in which one or more nucleotide bases are repeated. This may result in a frameshift to the right.

Epigenetics: The study of changes in gene expression that are not due to alterations in the nucleotide base sequence of DNA.

Frameshift mutation: A form of gene mutation in which the addition or deletion of nucleotide bases alters all subsequent triplet codes in a DNA sequence. This often leads to the production of a non-functional protein.

Gel electrophoresis: A technique that separates fragments of DNA by size using electric current.

Gene machine: A method of artificially manufacturing genes by feeding the desired sequence of bases into a computer.

Gene mutation: A change to at least one nucleotide base in DNA or the arrangement of bases. Gene mutations occur spontaneously and may result in changes to genotype.

Gene therapy: A technique in which a functional gene, cloned from a healthy individual, is inserted into cells that lack the gene.

Genetically modified organism (GMO): 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 fingerprinting: A technique used to genetically identify an organism. It has applications in forensics, paternity testing, diagnostics and the breeding of plants and animals.

Genetic screening: Testing individuals for certain faulty alleles.

Genome: The complete genetic material of an organism.

Hypermethylation: Increased methylation of DNA. This results in the inactivation of tumour suppressor genes and the resulting formation of tumours.

Hypomethylation: Reduced methylation of DNA. This results in the activation of oncogenes genes and the resulting formation of tumours.

Induced pluripotent stem (iPS) cells: Unipotent cells that have been reprogrammed (using transcriptional factors) to become pluripotent stem cells. iPS cells are capable of self-renewal.

Inversion: A form of gene mutation in which a group of nucleotide bases 'break off' from the DNA sequence and reattach in the same position but in the reverse order.

***In vitro*:** Describes a procedure that takes place outside of a living organism in a controlled environment e.g. DNA is amplified using PCR in a thermocycler.

***In vivo*:** Describes a procedure that takes place inside of a living organism e.g. fragments of DNA can be transferred to a host cell (using a vector) where they are amplified.

Malignant: Describes a tumour that is cancerous. Such tumours grow rapidly, are not enclosed in a capsule and can spread to other regions of the body. Treatment involves radiotherapy, chemotherapy or surgery.

Marker genes: 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. fluoresce or provide antibiotic resistance.

Metastasis: The process by which cells break off from a primary tumour and spread to other areas of the body, forming secondary tumours.

Methylation: The transfer of methyl groups to cytosine bases of DNA. Methylation inhibits transcription by making the DNA less accessible to transcriptional factors or preventing transcriptional factors from binding. This deactivates the gene.

Multipotent cells: Stem cells found in mature mammals that can only differentiate into a limited number of cell types (specific to a tissue).

Mutagenic agent: An agent that increases the rate of gene mutations above normal level.

Mutation: A random change in DNA which may result in genetic variants.

Mutation rate: The frequency of mutations per biological unit (e.g. per cell division).

Non-coding DNA: DNA that does not code for a protein but instead controls gene expression.

Oestrogen: A steroid hormone involved in the initiation of transcription. It joins to a receptor site on a transcriptional factor, activating the DNA binding site and stimulating transcription.

Oncogenes: Mutations of proto-oncogenes that are activated continuously.

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

Pluripotent cells: Stem cells found in embryos that have the ability to differentiate into almost all types of cell.

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

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

Promoter: Region of DNA where RNA polymerase binds during transcription.

Proto-oncogenes: Genes that stimulate cell division upon the attachment of growth factors to specific receptor proteins on the cell membrane.

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.

Risk factor: A variable associated with a greater chance of developing a disease or infection.

RNA interference (RNAi): A method of controlling gene expression by breaking down target mRNA molecules, preventing translation.

Silent mutation: A type of substitution mutation that produces the same amino acid due to the degeneracy of the genetic code.

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.

Substitution: A form of gene mutation in which one nucleotide base is exchanged for another.

Terminator: Region of DNA where RNA polymerase is released, ending transcription.

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

Totipotent cells: Stem cells found in early mammalian embryos which have the ability to differentiate into any type of body cell.

Transformation: The reinsertion of plasmids back into bacterial cells to form transgenic bacteria. This involves mixing the plasmids and bacterial cells in a medium containing calcium ions.

Tumour: An abnormal mass of cells formed by uncontrolled cell division.

Tumour suppressor genes: Genes that slow cell division, repair DNA and cause the breakdown of cells with damaged DNA by apoptosis.

Transcriptional factors: Specific molecules which pass from the cytoplasm of a cell into the nucleus, where they bind to complementary base sequences of DNA and initiate transcription.

Transgenic organism: An organism that contains recombinant DNA.

Translocation of bases: A form of gene mutation in which a group of nucleotide bases 'break off' from the DNA sequence on one chromosome and are added to the DNA sequence on a different chromosome.

Unipotent cells: Stem cells found in mature mammals that arise from multipotent cells and can only differentiate into a single cell type.

Variable number tandem repeats (VNTRs): Repeated sequences of non-coding nucleotide bases. It is unlikely that two unrelated individuals will have the same VNTRs.

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

Whole-genome shotgun (WGS) sequencing: A method of sequencing an organism's entire genome. This involves cutting the DNA into small segments and aligning overlapping sections using computer algorithms.