**Mutations**

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| 1 | Mutation | Any change to the quantity or structure of the DNA of an organism |
| 2 | Gene mutation | A change to one or more of the nucleotide bases or their rearrangement |
| 3 | Substitution mutation | If a nucleotide is changed in the DNA sequence |
| 4 | Nonsense mutation | If the base change results in the formation of a stop codon |
| 5 | Mis-sense mutation | If the base change results in a code for a different amino acid completely |
| 6 | Silent mutation | If the base change still codes for the same amino acid as before (as code is degenerate) |
| 7 | Deletion | If one or more nucleotides is lost from the DNA sequence, normally resulting in a ‘frame shift’ to the left in translation |
| 8 | Mutagen (mutagenic agent) | A material or other factor which increases the normal mutation rate eg high energy radiation, chemicals |
| 9 | Translocation | A mutation in which a section of a chromosome is moved from one position to another, either within the same chromosome or to another chromosome. |
| 10 | Inversion | A mutation in which a segment of a chromosome breaks off and is reinserted in the same place but in the reverse direction relative to the rest of the chromosome. |
| 11 | Addition | A mutation in which one or more nucleotides is inserted into a DNA sequence, normally resulting in a ‘frameshift’ to the right in translation. |
| 12 | Duplication | A mutation in which one or more pieces of DNA are copied. |

**Gene Expression and Cancer**

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| 13 | Totipotent (or omnipotent) | Cells which can mature into any type of body cell |
| 14 | Pluripotent | Cells which can differentiate into almost all types of cells eg in a blastocyst |
| 15 | Multipotent | Cells which can differentiate into a related family of cells eg blood or muscle cells. Used by body to repair and replace damaged tissue |
| 16 | Induced pluripotent stem cells  (iPSCs) | normal, specialised adult cells that have been genetically reprogrammed to become undifferentiated, pluripotent stem cells. iPSCs are a new development, still at the research stage, but they may solve some of the problems of both adult and embryo stem cells. |
| 17 | Explant | Small samples of plant used for tissue culture/micro-propagation. |
| 18 | Callus | A mass of undifferentiated plant cells grown from individual cells from a plant. A callus can be stimulated to form a plantlet |
| 19 | PGRs | Plant Growth Regulators- added to callus to allow them to grow into plantlets for propagation |
| 20 | Adult stem cells | Multipotent cells still existing in adult animals. Difficult to find and culture as usually only multipotent |
| 21 | Embryonic stem cells | Pluripotent cells existing in embryos ie before they have differentiated. From ‘spare’ IVF embryos- therefore ethically debatable |
| 22 | Transcriptional factors | Specific molecules which move from cytoplasm to nucleus to stimulate transcription |
| 23 | siRNA | Small interfering RNA- small double-stranded sections of RNA which prevent gene expression by bonding to complementary base pairs to ‘block’ transcription |
| 24 | Epigenetics | Epigenetics is the study of cellular and physiological traits (and their underlying mechanisms) that are heritable by daughter cells and which result from changes in gene expression that are not due to an alteration in the DNA nucleotide sequence). |
| 25 | Methylation | Methyl groups are added to the DNA which represses transcription as it leads to the DNA becoming more tightly packed. |
| 26 | Acetylation | Acetyl groups are added to histones which allows transcription as it leads to the DNA becoming more loosely packed. |
| 27 | Heterochromatin | Tightly packed DNA – strong association between histones and DNA. |
| 28 | Euchromatin | Loosely packed DNA – weak association between histones and DNA. |
| 29 | Tumour | A tumour is mass of identical cells (clones) formed by uncontrolled cell division. |
| 30 | Malignant tumour | Tumour which grows quickly and spreads throughout the surrounding tissue, affecting its normal function and so causing harm (e.g. lung cancer reduces elasticity of alveoli). More difficult to treat without damaging the whole tissue. |
| 31 | Benign tumour | Tumour which grows slowly, remains encased in a capsule and does not spread far eg wart. Removable by surgery or chemotherapy |
| 32 | Metastasis | Tumours which spread to the bloodstream or lymphatic system and can spread to other body parts, causing secondary tumours there-the most difficult to treat. |
| 33 | Proto-oncogene | Gene which controls a cell’s division, by stimulating it |
| 34 | Tumour suppressor gene | Gene which controls a cell’s division, by slowing it down |
| 35 | Two-hit hypothesis | Mutation of both alleles necessary to inactivate tumour suppressor genes ie the reason cancers are often associated with old age (mutation rates are slow so over a longer time, increased chance of two ‘hits’) |

**Genome Projects**

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| 36 | Genome | All of the genetic material in an organism. |
| 37 | Complete proteome | The full range of proteins coded for by the genome. (Identified by gel electrophoresis and mass spectrometry). |
| 38 | Cellular proteome | refers to the proteins produced by a given type of cell under a certain set of conditions. |
| 39 | Epigenome | The sum of all the epigenetic changes in a cell. |
| 40 | Sanger Sequencing | Initial method of sequencing genomes that requires terminator nucleotides, free nucleotides, primers, DNA polymerase and result in multiple fragments of varying length, which allows the DNA sequence to be determined. |
| 41 | Human Genome Project | International scientific research project to determine the nitrogenous base pair sequence which make up human DNA, identifying and mapping all the genes of the human genome |
| 42 | Next Generation Sequencing (NGS) | Faster, more recent sequencing methods that are continually developing and becoming more efficient, powerful and cost effective. |
| 43 | Whole-genome (shotgun) sequencing | Focuses on sequencing all of the DNA in an organism’s genome by cutting the DNA into many small, easily sequenced sections then uses computer algorithms to align overlapping segments to assemble the entire genome. |
| 44 | DNA electrophoresis | Method of separating out negatively charged DNA fragments by applying a current. The smaller fragments will move quicker towards the positive anode. |

**Recombinant DNA Technology**

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| 45 | Reverse transcriptase | Enzyme which can manufacture DNA from RNA |
| 46 | Retrovirus | Eg HIV, a virus containing RNA as genetic material which can replicate by manufacturing complementary DNA. |
| 47 | Recombinant DNA Technology (genetic engineering) | Processes by which genes are manipulated, altered or transferred from organism to organism. |
| 48 | Recombinant DNA | DNA of two different organisms combined as a result of gene transfer |
| 49 | Promoter | Region of DNA required to allow transcription of the gene to take place. |
| 50 | Terminator | Region of DNA required to stop transcription at the appropriate point |
| 51 | DNA polymerase | An enzyme which manufactures DNA by joining nucleotides (using a complementary strand as a blueprint).  NB it does NOT cause complementary base pairing |
| 52 | Restriction endonuclease | An enzyme which recognises and cuts DNA at a specific sequence of bases |
| 53 | Gene machine | A method of producing a gene by feeding the desired nucleotide sequence into a computer. |
| 54 | Oligonucleotide | Short sequence of DNA |
| 55 | Genetically modified organism (GMO) | An organism resulting from gene transfer from one organism to another, which has recombinant DNA |
| 56 | Vector | A carrier eg a plasmid or virus |
| 57 | Gene transfer/cloning stages | Isolation of DNA; insertion into vector; transformation into host; identification by gene markers; growth/cloning of host cell population |
| 58 | Complementary DNA | cDNA made with nucleotides lined up which are complementary to the mRNA template strand |
| 59 | Palindromic sequence | nucleic acid sequence on double-stranded DNA or RNA where reading the 5' to 3' forward on one strand matches the sequence reading backward 5' to 3' on the complementary strand |
| 60 | Sticky ends | The sequence of nucleotides exposed following an oblique ‘cut’ by a restriction endonuclease |
| 61 | DNA ligase | An enzyme which can join the phosphate-sugar framework of two sections of DNA eg joining sticky ends |
| 62 | Transformation | Reintroduction of plasmids back into host bacterial cells, by mixing them in a medium containing Calcium ions to increase their permeability |
| 63 | Marker genes | Ways to identify whether a gene has been taken up by a bacterial cell eg using antibiotic resistance, fluorescence or specific enzyme presence. |
| 64 | Replica plating | Method to identify bacterial cells with plasmids carrying antibiotic resistance genes by plating on antibiotic-rich medium |
| 65 | Polymerase chain reaction | Automated method of *in vitro* cloning in which fragments of DNA are copied very quickly and many billions of times |
| 66 | Primer | A short sequence of nucleotides with a set of bases complementary to those at one end of each of the two DNA fragments |
| 67 | Annealing | Joining of the primers to their complementary bases at the end of the DNA fragment |
| 68 | Thermocycler | A computer-controlled machine that varies temperatures precisely over a period of time |
| 69 | GM crops | Genetically modified crops changed by insertion of gene eg for resistance, over-fast softening of fruit |
| 70 | (Atryn) Anti-thrombin | Trade name for world’s first anticoagulant to be made from a genetically modified animal (a goat via its milk) |
| 71 | Gene therapy | Using defective gene replacement using genes cloned from healthy individuals |
| 72 | Germ-line gene therapy | Therapy involving replacing or supplementing the defective gene in the fertilized egg, so all of the cells in the new organism develop normally. Currently prohibited. |
| 73 | Somatic-line gene therapy | Therapy involving targeting the damaged tissue itself, so needs to be repeated periodically as cells die and need replacement. |
| 74 | CFTR | Cystic fibrosis trans-membrane-conductance regulator – chloride ion channel protein controls transport of Chloride ions across epithelial membranes. |
| 75 | adenovirus | Viruses which infect the respiratory tract, by injecting their DNA into epithelial cells of the lungs, so are useful vectors for gene transfer |
| 76 | Gene replacement | Defective gene is replaced with a healthy gene |
| 77 | Gene supplementation | One or more copies of the healthy gene (which are dominant alleles) are added alongside the defective gene, so the effect of the defective gene is masked |
| 78 | liposome | A lipid molecule wrapped around a gene, used to allow it across the cell-surface membrane |
| 79 | SCID | Severe Combined Immunodeficiency- an example of a genetic disorder which can be helped using gene therapy |
| 80 | Adenosine deaminase | The enzyme which destroys toxins which would otherwise kills leucocytes (white blood cells). The ADA gene is one which has been treated using gene therapy |
| 81 | DNA probe | Short, single stranded length of DNA linked to an easily identifiable label eg radioactive or fluorescent probes |
| 82 | DNA hybridisation | Combination of separated DNA strands with the probe, by binding it to the complementary bases on one of the strands. |
| 83 | DNA sequencing | Methods to determine the exact sequence in which the nucleotides are lined up in a piece of DNA eg Sanger sequencing |
| 84 | Cycle sequencing | A modified automated version of the Sanger method (12000 bases per min). The four deoxynucleotides are fluorescently labelled, polymerisation in a single tube, resulting mixture separated using capillary electrophoresis in a single narrow tube gel, then read by laser beam. Colour sequence is converted to DNA sequence by computer. |
| 85 | Gel electrophoresis | Method to separate the radioactively-labelled fragments of DNA after PCR by applying a voltage across a gel matrix, followed by detection using photographic film |
| 86 | Restriction mapping | Cutting DNA with a series of different restriction endonucleases (eg HindIII, BamHI, NotI), then separating the fragments. Distance between recognition sites can be discovered by the patterns of fragments produced. |
| 87 | Genetic screening | Checking for individuals in a family for a mutant allele eg sickle-cell anaemia |
| 88 | Genetic counselling | Advice for people at risk of genetic conditions i.e. when in family history, to discover risk to them and their family of its inheritance |
| 89 | Genetic fingerprinting  (genetic profiling) | Technique to determine the genetic identity of an organism eg in forensics, paternity cases, diagnostics, breeding programmes in conservation. It depends on an organism’s genome containing repetitive, non-coding introns, which have core sequences unique to the individual. |