**Mastery Matrix: Topic 8 – Controlling Gene Expression**

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| **Topic Title** | **Topic Number** | **Sub-Topic Title** | **Sub-Topic Number** | **Paper** | **Learning Statement** | **Statement Type** |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Understand that, while all cells within an organism carry the same genetic information, they only translate part of it | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Understand that the control of translation gives rise to specialised cells | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | State the types of gene mutation which can arise during DNA replication | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | State the effect of mutagenic agents on mutation rate | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Recall what is meant by a 'frame shift' | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Explain how the degenerate nature of the triplet code can lead to silent mutations | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Describe the effect of gene mutation on polypeptide structure/function | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Define a totipotent cell | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Identify the sources of totipotent cells in animals & plants | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Compare the properties and sources of pluripotent, multipotent and unipotent cells | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Examine cardiomyocytes as an example of unipotent cells | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Explain how protein transcription factors can be used to produce induced pluripotent stem cells (iPS cells) | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Evaluate the use of stem cells in treating human disorders | AT |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Produce tissue cultures, using explants of cauliflower | AT |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Describe the impact of specific transcriptional factors on gene expression in eukaryotic cells | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Describe the role of oestrogen in initiating transcription | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | Describe the impact of epigenetic factors on eukaryotic gene expression | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | Understand that epigenetics involves heritable changes in gene function, without changes to DNA base sequence | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | State the ways in which epigenetic changes are introduced | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | Discuss the importance of epigenetics in the development and treatment of disease (e.g. cancer) | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | State the role of RNA interference (RNAi) in inhibiting mRNA translation | K |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | Interpret data from investigations into gene expression | AT |
| Controlling Gene Expression | 8 | Epigenetic Control | 8.2 | 2 | Evaluate the relative phenotypic impacts of genetic and environmental factors, based on appropriate data | AT |
| Controlling Gene Expression | 8 | Cancer | 8.3 | 2 | Compare the characteristics of benign and malignant tumours | K |
| Controlling Gene Expression | 8 | Cancer | 8.3 | 2 | Describe the roles of tumour suppressor genes and oncogenes in tumour development | K |
| Controlling Gene Expression | 8 | Cancer | 8.3 | 2 | Explain the effects of abnormal methylation and increased oestrogen concentration on tumour development | K |
| Controlling Gene Expression | 8 | Cancer | 8.3 | 2 | Evaluate evidence for a correlation between genetic/environmental factors and various forms of cancer | AT |
| Controlling Gene Expression | 8 | Cancer | 8.3 | 2 | Apply an understanding of tumour suppressor/oncogenes to the treatment and curing of cancers | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | State what is meant by a 'sequencing project' | K |
| Controlling Gene Expression | 8 | Stem Cells & Gene Expression | 8.1 | 2 | Define the proteome | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Understand that the sequencing of an organism's genome provides information on its protein sequence | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Explain why the presence of non-coding DNA inhibits understanding of the proteome | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | State, briefly, how sequencing methods have changed over time | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Describe what is meant by 'recombinant DNA technology' | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Explain how the universal nature of the genetic code facilitates recombination | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | State what is meant by a 'transgenic' organism | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Describe, in detail, the methods for producing DNA fragments | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Describe how the polymerase chain reaction (PCR) can be used to amplify DNA fragments | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Explain how promoter/terminator regions, restriction endonucleases and ligases can be used to amplify DNA fragments *in vivo* | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | State how marker genes can be used to detect genetically-modified cells | K |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Interpret information relating to the use of recombinant DNA technology | AT |
| Controlling Gene Expression | 8 | Recombinant DNA Technologies | 8.4 | 2 | Evaluate the ethical, financial and social issues surrounding recombinant DNA technology | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Outline, briefly, the process of electrophoresis | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Explain the association between recombinant DNA technology and gene therapy | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Explain how DNA probes/hybridisation can be used to locate specific alleles | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Explain how labelled DNA probes can be used to screen patients for heritable disease/health risks | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Outline what is meant by 'genetic counselling' and 'personalised medicine' | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Evaluate information relating to genetic screening | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | State the definition and uses of variable number tandem repeats (VNTRs) | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Describe and explain the steps involved in genetic fingerprinting | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | State the applications of genetic fingerprinting | K |
| Controlling Gene Expression | 8 | Genetic Screening & Fingerprinting | 8.5 | 2 | Interpret data showing the results of gel electrophoresis | AT |

**Topic 8.1 – Stem Cells & Gene Expression: Key Knowledge**

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| Define a genetic mutation | Any change to the quantity or structure of an organism’s DNA. |
| Identify the 6 types of gene mutation which can occur | Insertion, duplication, deletion, inversion, substitution, translocation |
| Describe the three potential consequences of a substitution mutation | (i) No change at all (a ‘silent’ mutation); Minor change (i.e. substitution of a single a.a.); Significant changes (e.g. premature stop codon is introduced) |
| Define a mutagenic agent | A physical or chemical agent which disrupts the structure or copying of DNA, increasing the frequency of mutations above a natural, background level |
| Explain why an insertion mutation doesn't always lead to a frameshift | A multiple of three bases may be added, which doesn’t impact the reading of other codons |
| Explain why an individual's muscle and nerve cells have such differing properties, despite containing the same DNA | They differ in terms of the genes which are expressed/'switched on' |
| State the advantage to a specialised cell of transcriptionally silencing a particular gene | If not needed, silencing a gene's expression helps to reduce energetic expenditure |
| Define what is meant by a 'totipotent' stem cell | An undifferentiated cell, which can give rise to any type of bodily cell |
| Identify the source of totipotent stem cells | (early) embryos/zygote |
| State the two sources of multipotent stem cells | Adult bone marrow, umbilical cord blood |
| Describe the properties of an iPS cell | Induced pluripotent stem cells are unipotent cells, which have had silenced genes 'switched back on'. They are similar to pluripotent stem cells & exhibit self-renewal |
| Name the molecules which trigger transcription of a specific gene into mRNA | Transcriptional factors |
| State the function of oestrogen in the cell | Promotes the transcription of specific genes |
| Explain how oestrogen promotes gene expression | Binds to recognition sites on transcription factors, causing their active sites to change shape, so they can bind to the DNA |
| Name the small, double-stranded molecules of RNA which can be used to 'knock out' expression of target genes | Small interfering RNAs (siRNAs) |

**Topic 8.2 – Epigenetic Control: Key Knowledge**

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| Define epigenetics | The process by which environmental factors cause heritable changes to gene function or expression, without changing the base sequence of DNA |
| Name three types of species which contribute to the cell's epigenome by interacting with the DNA | Chemical tags, associated proteins, organic molecules/functional groups |
| Describe how the sources of epigenetic factors vary over an individual's lifetime | Early factors come from mothers during gestation, as we get older, lifestyle choices exert a larger influence |
| Name the protein molecules around which the DNA wraps itself, during condensation | Histones |
| Describe the link between the strength of DNA-histone association and the ease with which transcription can take place. | The more tightly-held/wound the DNA, the less readily it can be transcribed |
| Identify two epigenetic factors which regulate the transcription of certain genes | (i) Methylation of the DNA, (ii) Acetylation of the histones |
| Describe the process of: (i) acetylation, (ii) methylation | (i) Acetyl groups are transferred to the histone molecules, decreasing their positive charge ; (ii) Methyl groups are added to the cytosine bases of DNA, inhibiting the binding of transcriptional factors |
| Explain the impact of increasing acetylation on gene expression | Increasing acetylation decreases the positive charge on the histones, decreasing their attraction to DNA's phosphate group. As a result, the DNA is less-tightly held and can be more easily transcribed |
| Explain the impact of increasing methylation on gene expression | Increasing methylation prevents the binding of transcriptional factors to the DNA, inhibiting transcription of these genes |
| State what is mean by a change to the epigenome being 'heritable' | It can be passed on from one generation to the next |

**Topic 8.3 – Cancer: Key Knowledge**

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| Define 'cancer' | A family of disease, caused by uncontrolled proliferation of somatic cells |
| Identify the two types of gene associated with tumour development | Tumour suppressor genes, Oncogenes |
| State the name given to the family of genes which promote healthy DNA replication and cell division | Proto-oncogenes |
| Identify three ways by which tumour suppressor genes act to reduce the risk of tumour formation | (i) Slowing the rate of cell division, (ii) repairing mutations in the DNA, (iii) Triggering apoptosis (programmed cell death) in mutated cells |
| Describe two mechanisms by which oncogenes may act to promote cancer formation | (i) Permanently activating receptor proteins on the cell’s surface membrane, so it continues to divide; (ii) Promoting synthesis of growth factors inside the cell |
| Describe the effect of 'hypermethylation' on a particular gene sequence | Hypermethylation = increased binding of methyl groups, making it harder for transcriptional factors to bind and inhibiting gene expression |
| Name the fundamental cellular process which leads to tumour formation, when uncontrolled | Mitosis |
| Name the two types of tumour which can form within the body | Benign & malignant |
| Define metastasis | The process of cancer cells breaking off from the primary tumour and spreading to other parts of the body |
| Identify the two routes by which malignant tumours can metastasise | The blood & the lymphatic system |
| Explain why benign tumours are unable to metastasise | Cells are held together by adhesion molecules and a dense, surrounding capsule |
| Name the three treatment options used to tackle a malignant tumour | Surgical removal, radiotherapy, chemotherapy |
| Explain how benign tumours may still cause large-scale damage to the body | As they grow, they can press up against vital organs, disrupting their function |
| Define a carcinogenic factor | An environmental stimulus which drastically increases an individual’s risk of developing cancer |
| Identify four examples of carcinogens | Smoking, Diet/Obesity, Physical Activity, Sunlight/UV radiation |

**Topic 8.4 – Recombinant DNA Technologies: Key Knowledge**

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| Define a single nucleotide polymorphism (SNP) | A single-base variation, associated with a particular disease. Screening for these loci helps doctors to predict and prevent illnesses in their patients. |
| Name the enzyme used to generate cDNA molecules from mRNA | Reverse transcriptase |
| State the function of restriction endonuclease enzymes | Cut DNA molecules at a specific recognition site |
| Name the two types of 'ends' produced by restriction endonucleases and explain which is more 'useful' | Sticky ends & blunt ends - sticky ends are more useful, as they allow for complementary base pairing between 'overhanging' sequences |
| Name the enzyme which catalyses the formation of phosphodiester bonds between the sugar-phosphate backbones of DNA fragments | DNA ligase |
| Name the five steps involved in producing a functional protein via in vivo gene cloning | 1) Isolation of the target gene, 2) Insertion into a vector (plasmid), 3) Transformation (i.e. transferring into host cell), 4) Identification of ‘successful’ transfers, 5) Growth/propagation of protein-producing hosts |
| State the two conditions required for uptake of plasmids by bacterial cells | Gentle heating/warmth ; presence of calcium ions |
| State three reasons why transformation of a bacterial host may be unsuccessful | 1) Bacterial cells fail to take up the plasmid, altogether ; 2) Plasmid ‘closes up’ again, before target DNA is inserted ; 3) Ends of target DNA fragment join to each other, or attach to the plasmid in the wrong direction |
| Define a marker gene | An unrelated sequence, located on the same plasmid as our target gene, which triggers an easily-identifiable phenotype. |
| Identify three types of gene which might be used as a marker | Antibiotic resistance genes ; fluorescence genes ; enzyme-encoding genes |
| Explain why scientists tend to use two marker genes, when carrying out in vivo gene cloning | Reduces the risk of false positives (i.e. allows differentiation between cells which have taken up a transgenic plasmid and those which have taken up a non-transgenic plasmid) |
| Define replica plating | The process of pressing bacterial colonies against a fresh agar plate. This transfers some cells, creating a mirrored copy of the original |
| Describe the advantage of carrying out replica plating before exposing bacterial hosts to an antibiotic | Allows us to identify and propagate those colonies which have been successfully transformed |
| Name the three stages which comprise PCR and the temperatures at which each stage takes place | Separation (95°C), Annealing (55°C), DNA synthesis (72°C) |
| Name the substances required for successive rounds of PCR to take place | A specific DNA fragment; taq polymerase; short, single-stranded primers and a readily available source of DNA mononucleotides |

**Topic 8.5 – Genetic Screening & Fingerprinting: Key Knowledge**

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| Define a DNA probe | A short, single-stranded molecule of DNA, with an easily-identifiable label (or ‘marker’) attached |
| Identify two common examples of labels used in DNA probes | (i) Fluorescent compounds ; (ii) Unstable isotopes/sources of (ionising) radiation |
| Describe the method for assessing whether a radioactively-tagged probe has hybridised with a piece of sample DNA | Expose x-ray film to the sample. If hybridisation has occurred, the film will turn black. |
| Explain why a DNA sample must be washed thoroughly, following hybridisation with a DNA probe | Removes any unhybridised DNA probes, ensuring a more accurate diagnosis/fewer ‘false positives’ |
| Describe what is meant by 'personalised medicine' | A pattern of treatment, based on genetic screening, which is tailored to meet a patient’s unique care needs. |
| Identify two characteristics of medical treatment which might be impacted by genetic screening | The type of treatment a patient receives & the optimal dosage given |
| State the possible consequences of giving a patient a dose of medicine which is (i) too low, (ii) too high | (i) Treatment will be ineffective, (ii) Unnecessarily expensive & patient risks overdosing |
| Describe what is meant by 'genetic fingerprinting' | A method for comparing samples of DNA, in order to identify an individual, based on their unique sequence of bases |
| Identify the types of genetic sequence analysed in genetic fingerprinting | Variable number tandem repeats (VNTRs) |
| Explain why genetic fingerprinting uses VNTRs as a means of comparing individuals' genetic information | VNTRs vary in number & length, from person to person, creating unique patterns |
| Name the technique used to generate an individual's unique genetic fingerprint | Gel electrophoresis |
| Explain why scientists must always use the same restriction endonuclease, when generating an individual's genetic fingerprint | Ensures that the DNA is always cut at the same recognition sites, generating DNA fragments of the same size, each time |
| State and explain the direction in which DNA fragments move during gel electrophoresis | Fragments move from the cathode to the anode, as the phosphate groups in the DNA's backbone are negatively-charged |
| State the effect of increasing mass of a DNA fragment on its distance moved through the gel medium | The larger a fragment's mass, the shorter the distance moved (this is due to the fact that fragments with a larger mass move more slowly) |
| Explain how genetic fingerprinting can be used to estimate the genetic diversity within a population | Select two individuals, at random from the population ; generate genetic fingerprints for these two individuals ; compare the two genetic fingerprints ; the more similar their genetic fingerprints, the lower the diversity within the population |