2.4 Stem cells, specialisation and phenotype

Name: _____ Class: _____ Date: _____ **Total: 21 marks**

KEY WORDS totipotent • pluripotent • differential gene expression • post-transcriptional modification • polygenic

Objective

Practise statements 3.17-3.21: stem cells and potency, differential gene expression, post-transcriptional modification, genotype-environment interaction and polygenic inheritance.

1 Key knowledge and worked examples

■ Potency vocabulary

Totipotent – can form a whole organism plus placenta (zygote's first divisions). **Pluripotent** – any body tissue, from the **morula** and **blastocyst** 's inner mass. Every differentiation event narrows the options; a mature cell is locked in.

■ Same genome, different proteins – different cells

Differentiation is **differential gene expression** : transcription factors switch specific genes on or off. Even one gene can give several proteins: **post-transcriptional modification** splices the pre-mRNA differently, so exon combinations differ between tissues (e.g. the Dsx gene in fruit flies makes different proteins in males and females).

■ Phenotype = genotype × environment

Height needs genes AND nutrition; skin colour genes set a range the sun shifts; **polygenic** traits (many genes, small additive effects) give the bell-curve of continuous variation, with multiple alleles at each gene.

■ Worked example: a probability ladder

Two carriers of a recessive allele (both Aa) expect a child. P(affected aa) = 1/4. Add a second gene: parents Bb × Bb at an independent locus. P(affected at first AND carrier at second) = 1/4 × 1/2 = 1/8. Multiply only when events are independent – alleles of different loci segregating independently.

2 Practice

2.1 State the difference between a totipotent and a pluripotent stem cell. [2]

2.2 Explain why every cell in an organism has the same genes but different proteins. [3]

2.3 Give one example of a human phenotype affected by both genotype and environment. [1]

3 Exam-style questions

3.1 The Dsx gene of fruit flies is transcribed in every cell, but the pre-mRNA is spliced one way in males and another in females. Explain how this produces two different proteins from one gene, and name the level at which this control acts. [4]

3.2 Skin melanin amount is controlled by several genes (polygenic) and increases with sun exposure. Explain why skin colour in a population shows continuous variation rather than distinct categories, and what the environment adds to the genetic range. [5]

3.3 Stem cells from a blastocyst can be used to treat spinal injuries in trials. Describe the property of these cells that makes treatment possible, explain why rejection remains a risk, and give one ethical argument for and one against using embryonic stem cells. [6]

Solutions

Original-task marking: M1 method, A1 accuracy, B1 reasoning.

2.1 Totipotent cells can differentiate into any cell type including placenta and a whole organism; pluripotent cells can form any tissue of the body but not extra-embryonic tissue.

2.2 All cells inherit the same genome at division; each cell type switches on only a subset of its genes via transcription factors, so different mRNAs and proteins are made.

2.3 Any correct example with both components: height, skin colour, weight.

3.1 The same pre-mRNA transcript has its introns removed and exons joined differently in the two sexes, producing mRNA molecules with different exon sequences, which translate into proteins with different amino-acid sequences and functions. The control acts after transcription – at RNA processing/splicing (post-transcriptional).

3.2 Several genes each contribute a small, additive amount of melanin, so combinations of alleles give many slightly different levels – a continuum, not categories. Sun exposure adds environmental darkening on top of the genetic potential, so even identical genotypes differ in phenotype across environments.

3.3 Pluripotency – they can differentiate into the lost neuronal cell types. Rejection: the cells carry another individual's surface antigens, which the patient's immune system recognises as foreign. For: treatments can relieve untreatable suffering using embryos that would otherwise be discarded. Against: the embryo is destroyed, which those who grant it moral status cannot accept.