

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. $P(\text{affected aa}) = 1/4$. Add a second gene: parents Bb × Bb at an independent locus. $P(\text{affected at first AND carrier at second}) = 1/4 \times 1/2 = 1/8$. 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]
