

4

Genes and Phenotypic Expression

Subject Outline terms and phrases

phenotypic, cellular differentiation, tissue, gene expression, cytosine, methylation, epigenetic, cancer, mutation, cell division, ionising radiation, mutagenic chemicals, viruses, germ cells, somatic cells

1. What is meant by:

(a) phenotypic expression? _____

(b) phenotype? _____

2. Complete the following table of gene products that influence phenotypic expression.

Gene Product	Phenotypic expression
increased EPO	
	diabetes
	increased body size and muscle mass
auxins	
	ripening of fruit
testosterone	
	female secondary sex characteristics

3. Complete the following table of environmental factors that affect transcription and translation, and hence phenotypic expression.

Environmental factor	Phenotypic expression
lack of oxygen in humans	
	change in skin colour
lack of iodine in axoltl diet	
	goitre in humans
malnutrition in children	
	increased plant growth

4. (a) What are **transcription factors**?

(b) State two examples of transcription factors.

(c) State two ways in which transcription factors control gene expression.

5. Define the term cell differentiation, and give four examples of differentiated cells.

6. Explain how methylation of the cytosine nucleotide of a gene can affect the process of transcription.

7. Describe how epigenetic modifications such as changes in DNA methylation can lead to cancer.

8. (a) What is a 'mutation'?

(b) Explain what is meant by the idea that mutations can occur spontaneously.

(c) List three factors that can increase the mutation rate.

9. Complete the following sentence.

A change in the base sequence of _____ can cause a change in the _____ produced or the failure of a _____ to be produced. This may result in the appearance of new _____ in offspring.

10. (a) Explain the meaning of the term 'genetic disease'.

(b) State three reasons why mutations that occur in your cells may have no apparent effect on you.

11. Explain why mutation of DNA in a somatic cell, such as a skin cell causing skin cancer, does not get passed on to the next generation.

12. Explain why mutation of DNA in a germ cell, such as an ovum, can lead to changes in the characteristics of descendants. Give three examples.

13. (a) State two examples of genetic and/or chromosomal abnormalities that result in disease in humans.

(b) Describe the effects of these diseases.
