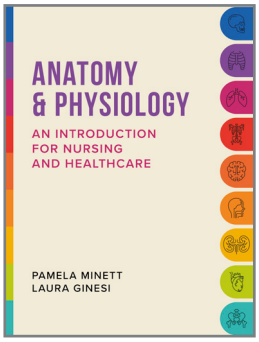
Lantern



Questions to accompany *Anatomy and Physiology*

CHAPTER 13 THE HUMAN GENOME

Multiple Choice Questions (MCQs)

Each question consists of a stem statement or question, and 5 options.
You must pick the one correct answer.

1. A gene is defined as:

A. the science of inheritance and mechanisms by which traits pass from one generation to the next
B. the code for a protein that plays a part in structure or function of the body
C. the complete set of instructions for the human body
D. a macromolecule that makes the chromatin for a cell
E. a molecule made from building blocks and folded into a 3-dimensional structure

2. Deoxyribonucleic acid (DNA) molecules are composed of building blocks called nucleotides. Which of the following bases is NOT found in nucleotides of DNA?

A. uracil
B. cytosine
C. guanine
D. arginine
E. thymine

3. Which is the correct sequence of events in the cell cycle?

A. $G_0 \rightarrow S \rightarrow G_1 \rightarrow G_2 \rightarrow M$
B. $M \rightarrow S \rightarrow G_1 \rightarrow G_2$
C. $G_1 \rightarrow S \rightarrow G_2 \rightarrow M$
D. $S \rightarrow G_1 \rightarrow M \rightarrow G_0$
E. $G_0 \rightarrow G_1 \rightarrow G_2 \rightarrow M$

4. DNA replication takes place during which phase of the cell cycle?

A. G_0
B. M
C. G_1
D. G_2
E. S

5. A change in the sequence of DNA is called a:

A. mutation
B. translation
C. transcription
D. replication
E. restriction

6. The haploid number of human chromosomes is:

A. 46
B. 23
C. 92
D. 12
E. 43

7. Genetically identical cells are produced through which process?

A. genesis
B. mitosis
C. meiosis
D. apoptosis
E. necrosis

8. Dominant alleles in a pair of genes:

A. mean that children always have the same eye colour as their parents
B. have the same effect as recessive alleles
C. mask the effect of recessive alleles
D. only have an effect when two copies are present
E. are donated by the male parent

9. When a cell divides by mitosis, the correct sequence of events is

A. metaphase → telophase → anaphase → prophase
B. prophase → metaphase → telophase → anaphase
C. anaphase → telophase → metaphase → prophase
D. prophase → metaphase → anaphase → telophase
E. telophase → metaphase → prophase → anaphase

10. The observable physical characteristics of an individual are known as their:

A. alleles
B. phenotype
C. genotype
D. dominant traits
E. recessive traits

11. If one parent is a homozygous tongue roller and the other parent is a homozygous non-tongue roller, what proportion of their children would be tongue rollers?

A. 5%
B. 25%
C. 50%
D. 100%
E. 0%

12. Which of these conditions has a genetic cause?

A. Marfan syndrome
B. Huntington's disease
C. thalassaemia
D. all of the above
E. none of the above

13. Cystic fibrosis is caused by an inherited allele that affects function of:

A. endocrine glands
B. bone marrow
C. neurocrine glands
D. exocrine glands
E. haemoglobin

14. Sickle cell anaemia is an inherited disorder that affects the structure of:

A. endocrine glands
B. bone marrow
C. neurocrine glands
D. exocrine glands
E. haemoglobin

Critical thinking: ARQs (assertion reasoning questions)

These questions consist of two statements:

- an assertion, and
- a reason.

You must first determine whether each statement is *TRUE* or *FALSE*.

- If both statements are true, you must next determine whether the reason correctly explains the assertion. The answer will be option 1 or option 2.
- If one statement is true and the other is false then the answer is option 3 or option 4, depending on which of the statements is correct.
- If both statements are false, then the answer is option 5.

  There is one option for each possible outcome.

Question 15

A = the Assertion	R = the Reason
Mutations can be beneficial when they provide the genetic material for natural selection and survival of the fittest	Mutations may result in chromosomal abnormality or inherited disease
Options	
1) Both A and R are true and R is the correct explanation of A	
2) Both A and R are true but R is NOT the explanation of A	
3) A is true but R is false	
4) A is false but R is true	
5) Both A and R are false	

Question 16

A = the Assertion	R = the Reason
When gametes (sperm and eggs) are formed, the alleles separate so each gamete has two alleles for each characteristic (trait)	Two sets of chromosomes, and therefore many pairs of alleles, are inherited by a zygote
Options	
1) Both A and R are true and R is the correct explanation of A	
2) Both A and R are true but R is NOT the explanation of A	
3) A is true but R is false	
4) A is false but R is true	
5) Both A and R are false	

Question 17

A = the Assertion	R = the Reason
Mitochondria have their own DNA which contains 37 chromosomes	Monogenic inheritance refers to the way genes are passed from one mitochondrion to another
Options	
1) Both A and R are true and R is the correct explanation of A	
2) Both A and R are true but R is NOT the explanation of A	
3) A is true but R is false	
4) A is false but R is true	
5) Both A and R are false	

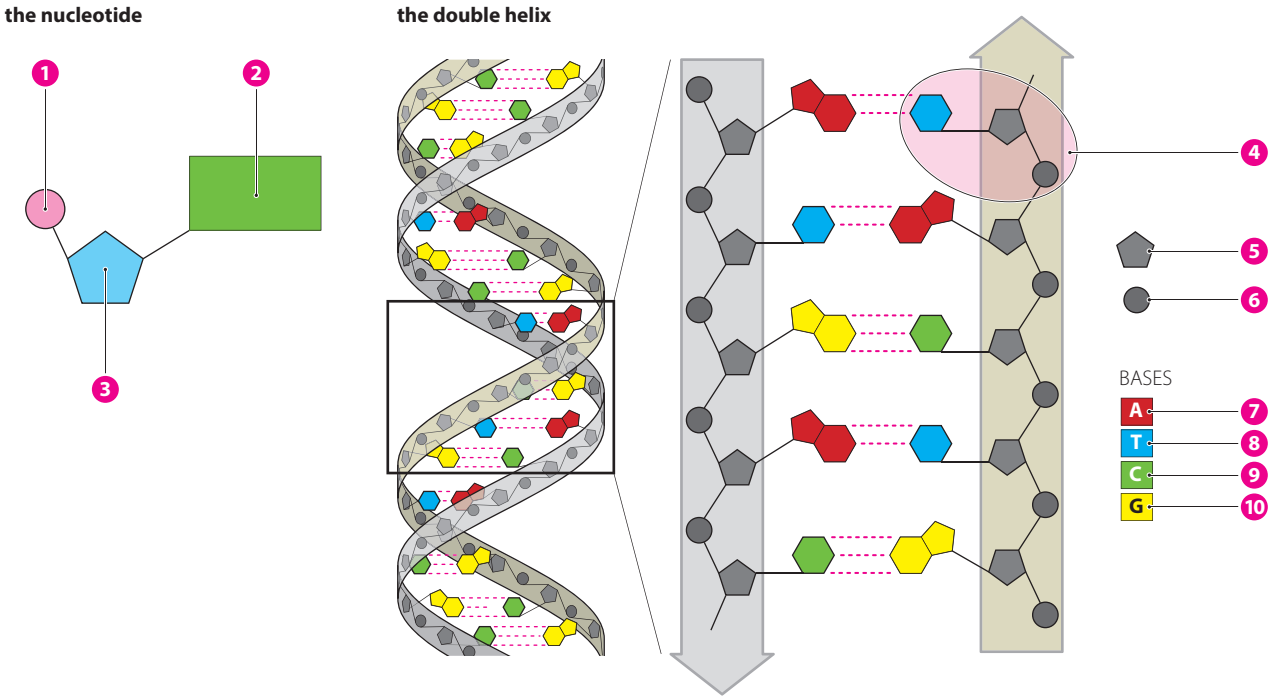
Question 18

A = the Assertion	R = the Reason
Transcription of a gene takes place in the cytoplasm of a cell	Ribosomes “decode” the information stored in mRNA to build polypeptides by assembling amino acids in the correct order
Options	
1) Both A and R are true and R is the correct explanation of A	
2) Both A and R are true but R is NOT the explanation of A	
3) A is true but R is false	
4) A is false but R is true	
5) Both A and R are false	

Putting it all together

Question 19

DNA is a macromolecule and genetic instructions are encoded in its structure. Label parts 1–10 of the figures below to demonstrate your understanding of the way in which nucleotides are arranged to form the double helix.



1 _____

2 _____

3 _____

4 _____

5 _____

6 _____

7 _____

8 _____

9 _____

10 _____

Question 20

Choose one of the following conditions that was explored in *Chapter 13*:

- Haemophilia
- Cystic fibrosis
- Sickle cell anaemia
- Thalassaemia
- Huntington's disease

A. In genetics, what is meant by the term "carrier"?
B. Explain, in your own words, how affected children inherit the disorder from carrier parents. You may find it helps to use figures and diagrams to support your explanation.

Answers to questions

Answers are supplied to most, but not all questions. Some may require you to carry out further research using the book.

1. **A gene is defined as:**
   B. the code for a protein that plays a part in structure or function of the body

2. **Deoxyribonucleic acid (DNA) molecules are composed of building blocks called nucleotides. Which of the following bases is NOT found in nucleotides of DNA?**
   A. uracil

3. **Which is the correct sequence of events in the cell cycle?**
   C. $G_1 \rightarrow S \rightarrow G_2 \rightarrow M$

4. **DNA replication takes place during which phase of the cell cycle?**
   E. S

5. **A change in the sequence of DNA is called a:**
   A. mutation

6. **The haploid number of human chromosomes is:**
   B. 23

7. **Genetically identical cells are produced through which process?**
   B. mitosis

8. **Dominant alleles in a pair of genes:**
   C. mask the effect of recessive alleles

9. **When a cell divides by mitosis, the correct sequence of events is**
   D. prophase → metaphase → anaphase → telophase

10. **The observable physical characteristics of an individual are known as their:**
    B. phenotype

11. **If one parent is a homozygous tongue roller and the other parent is a homozygous non-tongue roller, what proportion of their children would be tongue rollers?**
    D. 100%

12. **Which of these conditions has a genetic cause?**
    D. all of the above

13. **Cystic fibrosis is caused by an inherited allele that affects function of:**
    D. exocrine glands

14. **Sickle cell anaemia is an inherited disorder that affects the structure of:**
    E. haemoglobin

Critical thinking: ARQs (assertion reasoning questions)

These questions consist of two statements:
- an assertion, and
- a reason.

You must first determine whether each statement is *TRUE* or *FALSE*.
- If both statements are true, you must next determine whether the reason correctly explains the assertion. The answer will be option 1 or option 2.
- If one statement is true and the other is false then the answer is option 3 or option 4, depending on which of the statements is correct.
- If both statements are false, then the answer is option 5.

There is one option for each possible outcome.

Question 15

A = the Assertion	R = the Reason
Mutations can be beneficial when they provide the genetic material for natural selection and survival of the fittest	Mutations may result in chromosomal abnormality or inherited disease

2. Both A and R are true but R is NOT the explanation of A

Explanation

The assertion (A) is *TRUE*. Mutations are changes in the genetic material (DNA) of a cell and can occur as a result of mistakes in copying of DNA or through exposure to environmental factors such as cigarette smoke or UV light.

Mutations are part of both normal and abnormal physiological processes. The resulting change(s) can be a good thing or a bad thing for the cell and may or may not produce observable changes in the organism. Much depends on the way in which every mutation affects gene expression and the way proteins are made and function.

The Reason (R) is *TRUE*. Sometimes, the result of a mutation is an alteration in the structure and function of a protein that plays a critical role in the human body. Since every cell in the body depends on the activity of proteins:

- if a mutation alters one of the proteins that is critical to the human body, then the change may be harmful and result in a disease
- if the mutation is present in the sperm or an egg, then fertilisation may give rise to an embryo that carries the mutation in all of its cells (known as germ-line mutations).

Both statements are *TRUE* but not related, so **option 2 is the correct answer**.

Question 16

A = the Assertion	R = the Reason
When gametes (sperm and eggs) are formed, the alleles separate so each gamete has two alleles for each characteristic (trait)	Two sets of chromosomes, and therefore many pairs of alleles, are inherited by a zygote

4. A is false but R is true

Explanation

The Assertion statement (A) is *FALSE*. Gametes – sperm and eggs – are haploid cells that have been formed through the process of meiosis, which is a reduction division. As a result, each gamete has 23 chromosomes and hence only one allele for each characteristic.

The Reason (R) is *TRUE*. At fertilisation, the sperm and the egg fuse to form a diploid zygote, which has 46 chromosomes. Since each haploid cell contributes 23 chromosomes, the zygote inherits two pairs of every allele, one from maternal chromosomes and one from paternal chromosomes.

The Reason is true but the assertion is false, so **option 4 is the correct one to select**.

Question 17

A = the Assertion	R = the Reason
Mitochondria have their own DNA which contains 37 chromosomes	Monogenic inheritance refers to the way genes are passed from one mitochondrion to another

5. Both A and R are false

Explanation

The Assertion (A) is *FALSE*. Mitochondria do possess a small amount of DNA, but it is not in the form of chromosomes and it contains 37 genes. Most of the mitochondrial genes code for enzymes necessary for aerobic respiration and for ribosomal RNA that helps with assembly of proteins.

The Reason (R) is *FALSE*. Mitochondrial DNA is derived from the mother's cells, including the egg; therefore it is maternally inherited. This is because mitochondria are found in the flagellum (tail) of sperm; the flagellae, including their mitochondria, are required for swimming but are normally lost during fertilisation or degraded by the egg cell.

Since both statements are incorrect, option 5 is correct.

Question 18

A = the Assertion	R = the Reason
Transcription of a gene takes place in the cytoplasm of a cell	Ribosomes "decode" the information stored in mRNA to build polypeptides by assembling amino acids in the correct order

4. A is false but R is true

Explanation

The Assertion (A) is *FALSE*. Transcription is the process of copying a segment of DNA (the gene) to make a single strand of messenger RNA (mRNA) and it takes place in the nucleus of the cell, not the cytoplasm.

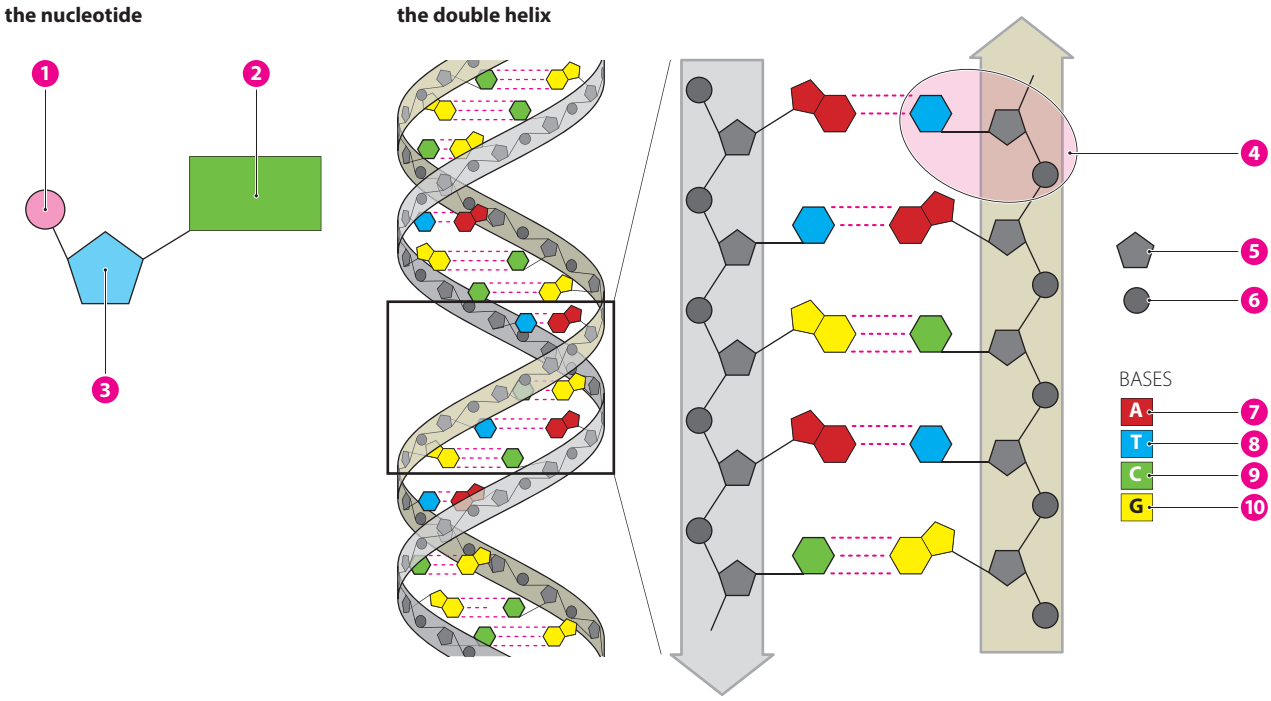
The Reason (R) is *TRUE*. Ribosomes are found in the cytoplasm of cells and their function is the synthesis of proteins such as enzymes, antibodies, hormones and clotting factors. Specifically, ribosomes take part in the translation stage of protein synthesis, when mRNA is used as the template for assembly of amino acids in the correct order.

Since the Assertion (A) is false, and the Reason (R) is true, **option 4 is the correct answer**.

Putting it all together

Question 19

DNA is a macromolecule and genetic instructions are encoded in its structure. Label parts 1–10 of the figures below to demonstrate your understanding of the way in which nucleotides are arranged to form the double helix.



1 phosphate	6 phosphate
2 base (C,G, A or T)	7 adenine
3 sugar (deoxyribose)	8 thymine
4 nucleotide	9 cytosine
5 sugar	10 guanine