

## TRIAL TEST 6: MUTATIONS



<b>Time allowed:</b> 60 minutes	<b>Section One – Multiple Choice</b>	30 marks
<b>Total marks:</b> 100	<b>Section Two – Short Answer</b>	50 marks
	<b>Section Three – Extended Answer</b>	20 marks

### SECTION 1 – MULTIPLE CHOICE (30 MARKS)

1. Mutations are:
  - (a) lethal.
  - (b) beneficial.
  - (c) benign.
  - (d) any of the above.
2. Mutations can be caused by:
  - (a) chemicals.
  - (b) viruses.
  - (c) high temperatures.
  - (d) all of the above.
3. Most mutations occur during
  - (a) meiosis.
  - (b) mitosis.
  - (c) chromosomal replication.
  - (d) homologous pairing.
4. Which of the following changes is most likely to result in a mutation in an offspring?
  - (a) A change to the DNA in a somatic cell.
  - (b) The deletion of a gene in the DNA of a sperm cell.
  - (c) Crossing over during meiosis.
  - (d) A mistake in the replication of DNA during mitosis.
5. Most mutations result in recessive genes. They are therefore:
  - (a) harmful to the offspring.
  - (b) mostly hidden and therefore neutral in their effects.
  - (c) deleterious as they result in the offspring having a disadvantage in competition with others of the same species.
  - (d) useful, especially in giving future generations an advantage over competitors.
6. The main aim of the Human Genome Project was to:
  - (a) determine the locus of each gene in the human population.
  - (b) determine the locus of each chromosome in the human karyotype.
  - (c) determine the locus of each gene on the karyotypes of a small sample of people.
  - (d) locate where every deleterious gene was located on a human chromosome.

7. The survival rates of people who have the sickle-cell gene differ from those who do not in parts of the world where malaria is prevalent. People who are homozygous normal (SS) are less likely to survive than those who have the sickle-cell trait (Ss). People who have sickle-cell anaemia (ss) are less likely to survive than either of these two groups as they need frequent blood transfusions, which are not available to most people in developing countries.

The description above can best be described as:

- (a) an adaptation.
  - (b) differential survival rates.
  - (c) genetic competition.
  - (d) hybrid vigour.
8. Cystic fibrosis is a recessive, human genetic disease that most commonly results from the loss of three base pairs on chromosome 7. The loss of these and the amino acid that they code for, changes a protein on the cell membrane. As a consequence, the mucus coating lining the lung accumulates, becoming stickier and thicker than normal and it becomes harder to breathe. Currently, it is treated with antibiotics and daily physiotherapy. If two normal parents, heterozygous for this trait had a child, what would be the chance of their child having cystic fibrosis?
- (a) 0%.
  - (b) 25%.
  - (c) 75%.
  - (d) 100%.
9. The triplet code for the amino acid tryptophan can be written on the genetic code as UGG. This code represents what appears on:
- (a) DNA.
  - (b) tRNA.
  - (c) mRNA.
  - (d) none of the above.
10. Huntington's Chorea, a form of dementia, is caused by multiple repeats of a three base sequence on chromosome 4. Everyone has repeats of this sequence, CAG, usually 10 to 15. However, if someone has more than 36 repeats of this sequence, Huntington's will develop.
- This sequence of three bases is called:
- (a) an amino acid.
  - (b) an anti-codon.
  - (c) a codon.
  - (d) a triplet.

## SECTION 2 – SHORT ANSWER (50 MARKS)

1. Write the appropriate term or words for each of the phrases below:

(i) An organism which has a mutation.

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(ii) A record of an individual's ancestral history showing inheritance patterns for a particular trait.

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(iii) Any type of condition present at birth either inherited or caused by environmental factors.

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(iv) An alternative form of a gene.

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(v) Body cell (not a gamete).

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(vi) Having a harmful effect.

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(vii) The expression of a gene.

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(viii) A chromosome which is not a sex chromosome (X or Y).

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(ix) Cell division that produces gametes.

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(x) Uncontrolled cell growth.

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[10 marks]

2. Genetic disorders such as Fragile X Syndrome, sickle-cell anaemia, cystic fibrosis, and Huntington's Chorea, are all caused by mutations.

(a) What are mutations?

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[2 marks]

(b) List three types of mutagens and give an example of each.

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[6 marks]

(c) Explain in general terms, how these might result in mutations.

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[3 marks]

3. Distinguish between somatic mutations and germ-line (gametic) mutations. Which ones have the potential to be passed on? Why?

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[6 marks]

4. Explain what is meant by each of the following statements.

(a) 'Mutations arise spontaneously, they are undirected by the environment.'

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[2 marks]

(b) 'Mutations are relatively persistent'.

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[2 marks]

- (c) 'The vast majority of mutations confer no advantages on the organisms that inherit them.'

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[2 marks]

5. (a) What kind of mutations are likely to be most important in evolution?

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[2 marks]

- (b) What happens to unfavourable mutations?

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[2 marks]

6. (a) If a person had a karyotype that included the sex chromosomes XXY, what syndrome would they have?

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[1 mark]

- (b) Would this person be a male or a female?

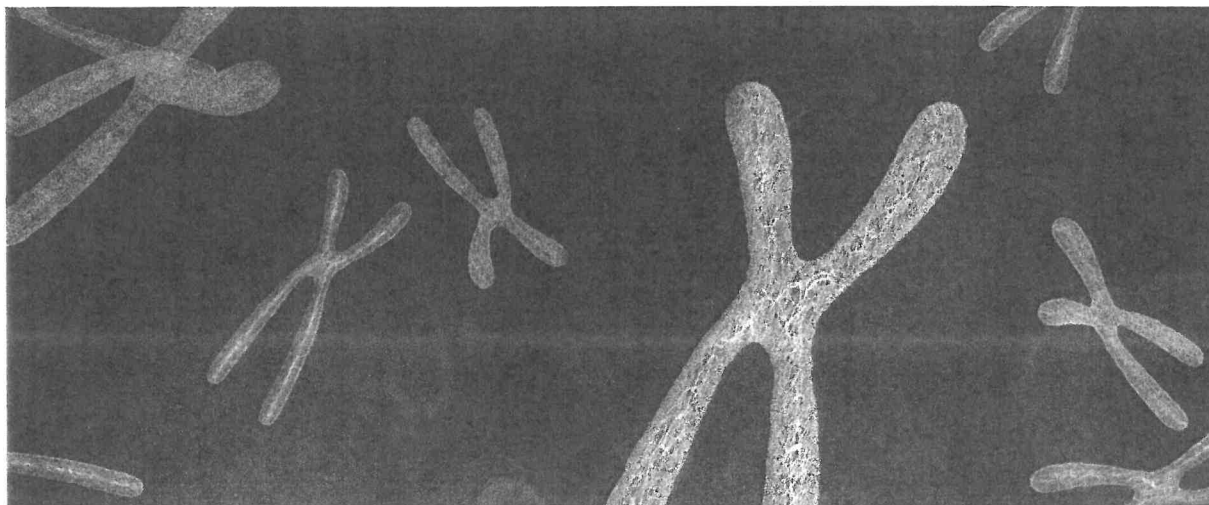
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[1 mark]

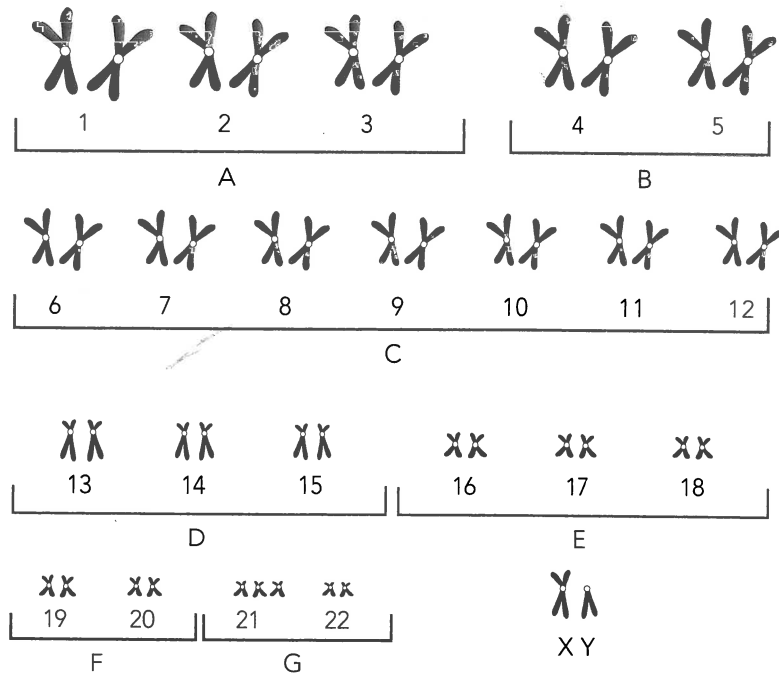
- (c) How many chromosomes would this person have in a somatic (body) cell?

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[1 mark]



7. Study this karyotype.



(a) What is the sex of the person having this karyotype?

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[1 mark]

(b) Is there any abnormality in the karyotype?

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[1 mark]

(c) What condition would this person have?

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[1 mark]

(d) How might this condition come about?

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[2 marks]

8. Explain how degeneracy of the genetic code reduces the chances of mutation.

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### SECTION 3 – EXTENDED ANSWER (20 MARKS)

- (a) A woman recently had her genome mapped. In it she discovered that she has a rare mutation for breast cancer and she chose to have a double mastectomy to prevent the disease. Discuss how genetic diseases may arise in a population and what may be done to minimise them.

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[10 marks]

- (b) How do genetic counsellors use pedigrees and genetic testing to provide advice to couples who are planning to have children?

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[10 marks]

END OF TEST (100 MARKS)