26.1 Mutations and gene editing

Name: _____ Class: _____ Date: _____ **Total: 20 marks**

KEY WORDS mutation • substitution • frameshift • mutagen • somatic • gene editing

Objective

Build the skills to answer exam questions on **mutations and gene editing** : substitutions, insertions and deletions, causes of mutation, cancer, and gene editing.

You must be able to:

- describe base substitutions, insertions and deletions, and their effects on proteins
- state causes of mutation and explain why mutation is random
- distinguish germ-line and somatic mutations, and link mutations to cancer
- explain why mutation is the original source of genetic variation

1 Key knowledge and worked examples

Mutations and gene editing

Gene mutation: a change in the base sequence of a gene.

type	change	effect
substitution	one base replaced	one codon changes: same amino acid, a different amino acid, or a stop codon
insertion	one or more bases added	if not a multiple of 3, a **frameshift**: every later codon changes
deletion	one or more bases lost	frameshift, usually a non-working protein

Causes: mutagens such as **ionizing radiation** (X-rays, gamma rays), **UV light**, and chemicals (e.g. in tobacco smoke), plus copying errors in replication. Mutation is **random**: no mechanism directs a change to a particular gene, although some sites mutate more often.

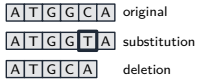
Germ-line mutations (in cells that form gametes) can be **inherited**; **somatic** mutations (body cells) are not passed to offspring but can cause **cancer** if they affect genes that control cell division. Mutation is the original source of all new **alleles**, so it provides the **variation** that natural selection acts on.

Worked example. Coding strand ATG GCA TTT. Deleting the C gives ATG GAT TT…: from the second codon on, every codon changes: a frameshift.

Answer shapes: base change → codon change → amino acid change → shape/function change.

2 Practice

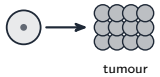
2.1 Distinguish between a substitution and a deletion. [2]



2.2 Explain why a deletion of one base usually has a bigger effect than a substitution. [3]



2.3 Explain why a mutation in a skin cell cannot be inherited, but can still be harmful. [3]



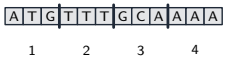
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3 Exam-style questions

3.1 The coding strand of part of a gene is ATG TTT GCA AAA.

codon	AUG	UUU	GCA	AAA	GAA	CAA	UAA
amino acid	Met	Phe	Ala	Lys	Glu	Gln	stop

(a) Give the amino acid sequence coded by this DNA. [1]



(b) The second base of codon 3 (C) is deleted. Write the new codons and amino acids, as far as the table allows. [2]

(c) A substitution changes AAA to UAA in the mRNA. Explain its effect. [2]

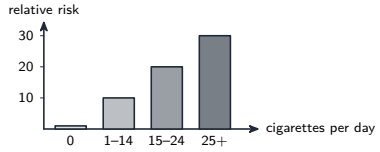
(d) State one mutagen. [1]

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3.2 The graph shows the risk of lung cancer in people who smoke different numbers of cigarettes per day.

(a) Describe the relationship shown. [1]

(b) Explain how smoking can cause lung cancer. [3]



(c) Explain why mutation is important for evolution. [2]

Solutions

Marking: **M1** a correct method, **A1** the correct answer, **B1** an independent mark.

Each answer shows the steps, then the **result**.

2.1

substitution: one base is replaced by another; deletion: a base is removed, shifting the reading frame

2.2

a substitution changes only one codon, and may not change the amino acid (degenerate code); a deletion shifts the reading frame (frameshift); every codon after it changes, so many amino acids change / an early stop codon appears

2.3

it is a somatic mutation, not in a germ-line cell / gamete, so it is not passed to offspring; if it affects a gene that controls cell division; the cell can divide uncontrollably, forming a tumour / cancer

3.1

(a) Met–Phe–Ala–Lys

(b) ATG TTT GAA AA…: codons AUG UUU GAA A…; Met–Phe–Glu: every codon after the deletion changes, so Ala and Lys are lost: a frameshift

(c) UAA is a stop codon; translation stops early, so the protein is shorter and probably does not work

(d) any one: X-rays / gamma rays / UV light / chemicals in tobacco smoke

3.2

(a) the more cigarettes per day, the higher the risk (positive correlation)

(b) tobacco smoke contains mutagens / carcinogens; these cause mutations in the DNA of lung cells; mutations in genes that control cell division make cells divide uncontrollably, forming a tumour

(c) mutation creates new alleles; this gives the genetic variation on which natural selection acts

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