Exercise walk-through

Mutations ▪ 6.7

eXercise

You must be able to

- describe **point mutations** 点突变 (substitution) and **frameshift** 移码 (insertion/deletion)
- explain silent, missense, and nonsense effects
- link mutations to variation and disease

Method — Types of mutation

A **mutation** is a change in the DNA sequence:

- **Substitution** (point) — one base is swapped.
- **Insertion / deletion** — bases are added or removed, causing a **frameshift** that changes every codon after it.

Method — Effects of a substitution

- **Silent** — the codon still codes for the same amino acid (no change).
- **Missense** — a different amino acid is used (protein may work worse).
- **Nonsense** — a stop codon appears early, cutting the protein short.

Method — Frameshift is severe

An insertion or deletion (not a multiple of three) shifts the **reading frame**, so all downstream codons change — usually ruining the protein.

Method — Consequences

Mutations create **variation** (raw material for evolution) but can also cause disease (e.g. sickle-cell from a single substitution).

Practice 2.1 ▪ 1 mark

What is a point mutation?

Solution 2.1

Method: Types of mutation

Worked steps

A change in a single DNA base (substitution) B1.

Practice 2.2 ▪ 1 mark

What kind of mutation shifts the reading frame?

Solution 2.2

Method: Frameshift is severe

Worked steps

An insertion or deletion (frameshift) B1.

Practice 2.3 ▪ 1 mark

What is a silent mutation?

Solution 2.3

Worked steps

A base change that does not change the amino acid coded B1.

Exam-style 3.1 ▪ 1 mark

A substitution that creates an early stop codon is a

A silent mutation

B missense mutation

C nonsense mutation

D frameshift

Solution 3.1

Method: Effects of a substitution

A silent mutation

B missense mutation

C nonsense mutation ✓

D frameshift

Worked steps

C — a nonsense mutation.

Exam-style 3.2 ▪ 2 marks

A single base is deleted near the start of a gene.
(a) Name the type of mutation and its effect on the reading frame.
(b) Explain why this is usually more damaging than a single substitution.

Solution 3.2

Worked steps

(a) A frameshift (deletion); it shifts the reading frame so all following codons change M1 A1. (b) A substitution changes at most one amino acid, but a frameshift changes every downstream codon, usually ruining the whole rest of the protein M1 A1.

Exam-style 3.3 ▪ 2 marks

Explain how mutations can be both harmful and a source of useful variation.

Solution 3.3

Method: Consequences

Worked steps

Harmful mutations can disrupt a protein and cause disease, but mutations also create new alleles (variation) that natural selection can act on, sometimes benefiting the organism M1 A1.