

6.7 Mutations

Name: _____ Class: _____ Date: _____ **Total: 10 marks****KEY WORDS** mutation 突变 • frameshift 移码 • point mutations 点突变 • codons 密码子

Objective

Build the skills to answer exam questions on **mutations**.**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

1 Worked examples

Study these first. Each one shows the method for a question type used later — follow the steps and you can do the Practice and Exam-style questions yourself.

■ 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.

■ 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.

■ 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.

■ Consequences

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

2 Practice

Now apply the methods above.

2.1 What is a point mutation? [1]**2.2** What kind of mutation shifts the reading frame? [1]**2.3** What is a silent mutation? [1]

3 Exam-style questions

3.1 A substitution that creates an early stop codon is a [1]

- **A** silent mutation
- **B** missense mutation
- **C** nonsense mutation
- **D** frameshift

3.2 A single base is deleted near the start of a gene.

(a) Name the type of mutation and its effect on the reading frame. [2]

(b) Explain why this is usually more damaging than a single substitution. [2]

3.3 Explain how mutations can be both harmful and a source of useful variation. [2]

Solutions

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

2.2 An insertion or deletion (frameshift).

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

3.1 C — a nonsense mutation.

3.2 (a) A frameshift (deletion); it shifts the reading frame so all following codons change.

(b) A substitution changes at most one amino acid, but a frameshift changes every downstream codon, usually ruining the whole rest of the protein.

3.3 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.