D.1.3 · Mutations and gene editing

International Baccalaureate · IB Diploma · Biology · SL

Scope and prerequisites

This handout covers selected SL topics for the course first assessed in 2025; it is not the whole syllabus.

Prerequisites: DNA structure from A.1.2 and protein synthesis from D.1.2.

The examples are original and fictional: they are not real measurements or real student investigations.

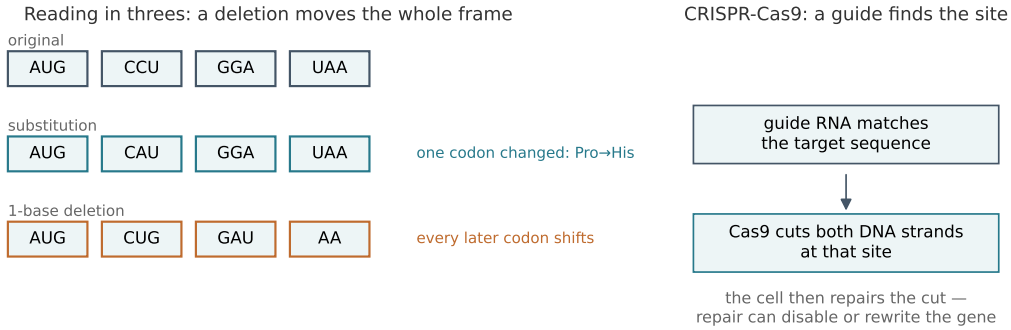
Mutations and gene editing

■ What would explain this observation?

- One changed base in the haemoglobin gene causes sickle cell disease, while a one-base deletion in another gene destroys the entire protein. Why does one small edit matter so much more than the other?

■ Build the model

- A **mutation** is a change in the base sequence of DNA. A **substitution** swaps one base; if the new codon still codes for the same amino acid (a silent change) the protein is unchanged, but a change to a different amino acid can alter the protein's shape, as in sickle cell disease.
- An insertion or deletion within a coding region shifts the **reading frame** when the number of bases added or lost is not a multiple of three. This can change the amino-acid sequence after the edit or introduce an early stop codon, affecting protein function. **Gene editing** with CRISPR-Cas9 uses a guide RNA to find one chosen sequence and the Cas9 enzyme to cut both strands there; the cell's repair then disables or rewrites the gene.
- mutation: A change in the base sequence of DNA; substitution: A mutation swapping one base for another; reading frame: The grouping of bases into codons from a fixed start; gene editing: Deliberately changing a chosen DNA sequence, for example with CRISPR-Cas9.



A coding substitution affects one codon; the one-base deletion shown shifts the frame downstream. Cas9 cuts where the guide RNA matches

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■ Choose evidence that can test it

- Compare the DNA sequence with the protein produced: a coding substitution may change one amino acid, create a stop codon or leave the amino acid unchanged. A one-base coding deletion shifts the downstream reading frame. Sequence data show which changes actually occur.
- In gene editing, sequencing the target site after repair shows whether the cut disabled the gene or inserted the intended change; an off-target check sequences similar sites elsewhere.

■ Work from known quantities

- Known: the coding sequence AUG CCU GGA UAA reads in codons of three. Delete the fourth base (the first C). The sequence becomes AUG CUG GAU AA: the first codon is intact, but every later codon is shifted, and the stop codon is destroyed – the ribosome reads on into wrong codons. A substitution of the fifth base (C to A) instead gives AUG CAU GGA UAA: only the second codon changes.

Example:

A 30-base coding segment loses its fourth base. Is the first codon changed, and what happens after it? (Answer: the first three bases still form the original first codon. The remaining bases are grouped in a new frame. The final peptide length depends on where a stop codon is encountered.)

Check the conclusion and its limits

- This simple model covers the coding region; mutations in control regions can change how MUCH protein is made without changing its sequence. Gene editing's safety depends on off-target rates measured experimentally, not on the guide's design alone.

Warn:

A mutation is always harmful and always changes the protein. This claim is false: silent substitutions change the DNA but not the amino acid, many mutations fall outside coding regions, and a few mutations are beneficial – variation from mutation is the raw material of evolution.

Key:

Mutations and gene editing: a coding substitution affects one codon, with effects that depend on the new sequence. A coding insertion or deletion shifts the frame only when its length is not a multiple of three; protein function may change. CRISPR-Cas9 edits by guide-RNA targeting plus a two-strand cut, with results checked by sequencing.

Write from a 3-base deletion

- One complete codon is deleted from a coding region, without crossing a codon boundary.
- Say what happens to the protein: one amino acid is lost and the frame survives – 3 is a whole codon.

Write from a substitution to a stop codon

- One base changes so a codon becomes a stop codon, 40 codons into the sequence.
- Say what happens: translation ends early – a shortened protein, not a shifted one.

Write from a silent change

- A substitution in a non-coding region appears in a population.
- Does the protein change? No – and even coding substitutions can be silent under degeneracy.