D.1.3 · Mutations and gene editing · supported focus

International Baccalaureate · IB Diploma · Biology · SL

Scope and prerequisites

Supported SL core focus. First assessment 2025; current subject brief acquired; full Biology guide not acquired. Remaining guide, assessment and practical requirements retain their recorded holds.

Prerequisites: DNA structure from A.1.2 and protein synthesis from D.1.2. Each lesson develops its own method before independent transfer.

These are original or explicitly fictional teaching examples, not actual measurements or completed assessed learner investigations.

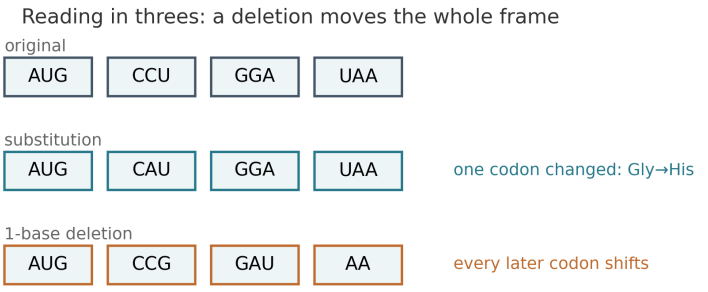
Mutations and gene editing

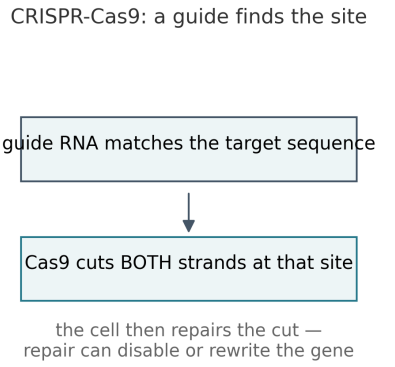
■ What would explain this observation?

- 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?
- Start with a prediction. State the quantities or features you would compare, then decide what evidence could distinguish two explanations.

■ 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 shifts the **reading frame**: every codon after the change is misread, so the protein is usually destroyed. **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.





■ Choose evidence that can test it

- Compare the DNA sequence with the protein produced: a substitution predicts one changed amino acid at one position, while a deletion predicts every amino acid changed after it. Sequence data decide between the two.
- 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

- State the known values and their units. Choose the relation because its assumptions fit this case, then rearrange before substitution.
- Known: the coding sequence AUG CCU GGA UAA reads in codons of three. Delete the fourth base (the first C). The sequence becomes AUG CCG 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 that same C to A instead gives AUG CAU GGA UAA: only the second codon changes.

Example:

A coding sequence of 30 bases loses one base near the start. How many codons are potentially misread, using the same sequence: known quantities → model → relation → substitution → unit and interpretation? (Answer: $30/3 = 10$ codons; losing one base near the start shifts all 10, while a substitution would have changed at most one.)

■ 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.
- Return to the original observation. Explain what the result supports, which conditions it assumes, and one way to test a competing explanation.

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: substitutions change one codon; insertions and deletions shift the reading frame and usually destroy the protein. CRISPR-Cas9 edits by guide-RNA targeting plus a two-strand cut, with results checked by sequencing.