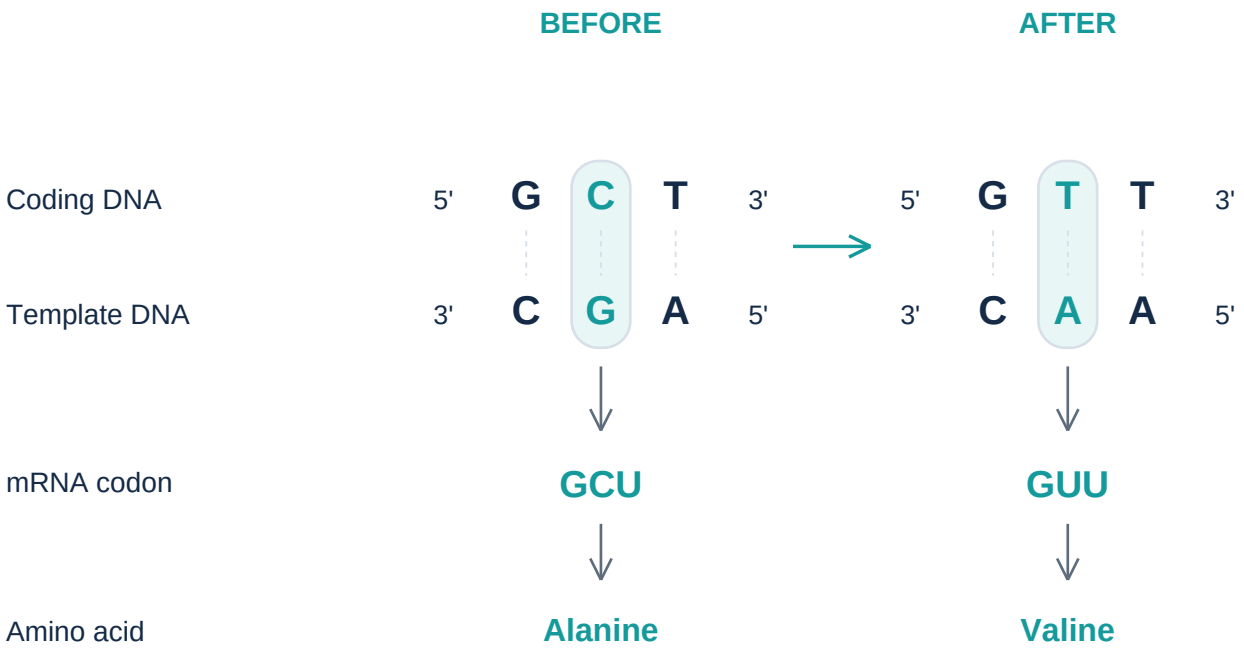
A change in the DNA base sequence

A gene mutation is a change in the sequence of base pairs in DNA that may result in an altered polypeptide. The DNA change is the mutation; a protein change is a possible consequence.

1. FOLLOW ONE BASE-PAIR CHANGE



Highlighted pair: C-G becomes T-A. RNA codons are shown 5' to 3'. This is one internal triplet in an illustrative coding region, not a complete gene.

2. CONNECT SEQUENCE WITH A POSSIBLE EFFECT

Here, the changed DNA triplet produces a different mRNA codon, so valine replaces alanine at this position. A different amino acid may change interactions within the polypeptide, its folding and the function of the resulting protein.

The example establishes an amino-acid change, not a guaranteed loss of function. A mutation can arise through a DNA-copying error or DNA damage. The detailed effects of substitutions, insertions and deletions follow in the next lesson.

DNA changes and their origins: NHGRI, Mutation.
Amino-acid changes and variable effects: NHGRI, Missense Mutation.

Scope: Cambridge Biology 9700 syllabus, section 6.2, outcome 6, p. 25.

Why the definition says "may"

A DNA sequence change does not guarantee a different polypeptide, and a different polypeptide does not guarantee a non-functional protein. Check the consequence at each level.

1. THE AMINO ACID CAN STAY THE SAME

Example: coding DNA GCT becomes GCC. The mRNA codon changes from GCU to GCC, but both specify alanine in the standard code. This is a synonymous (often called silent) change: the amino-acid sequence is unchanged.

2. THE AMINO ACID CAN CHANGE

Example: coding DNA GCT becomes GTT, giving RNA GCU then GUU. Alanine becomes valine, as shown on page 1. This is a missense change. Its effect depends on the position and chemical properties of the amino acid; protein function may or may not change.

3. TRANSLATION CAN STOP EARLIER

Example: an internal coding-DNA triplet TAT becomes TAA. The RNA codon changes from UAU (tyrosine) to UAA (stop). This nonsense change can produce a shorter polypeptide. A stop codon does not add an amino acid.

4. DNA CHANGE AND PROTEIN EFFECT ARE DIFFERENT CLAIMS

A synonymous change is still a DNA mutation even if the amino-acid sequence is unchanged. Conversely, a protein changing shape because of temperature or pH is not itself a gene mutation: that requires a change in DNA sequence.

All example DNA triplets are coding-strand sequences written 5' to 3'.

Codon meanings and degeneracy: OpenStax Biology 2e, section 15.1.
Premature stop signals: NHGRI, Nonsense Mutation.

Scope: Cambridge Biology 9700 syllabus, section 6.2, outcome 6, p. 25.