

Mutations

Biology

The types of mutation, and their effect on the protein.

By mechanism and consequence.

Type	Change	Effect
Silent	Base changed, same amino acid	Usually none
Missense	Different amino acid	Variable
Nonsense	Creates a stop codon	Truncated protein
Insertion / deletion	Bases added or removed	Frameshift, unless a multiple of 3
Duplication	Region copied	Dosage effects
Inversion	Segment reversed	Disrupts genes at breakpoints

Sickle cell is a single missense mutation: one base change, one amino acid, and a substantially altered haemoglobin.