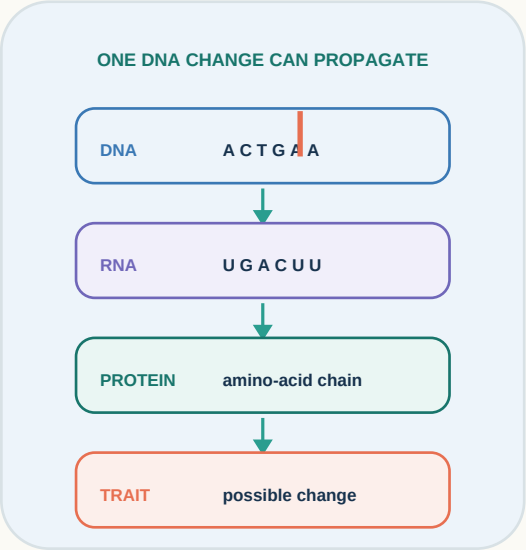
AP BIOLOGY | UNIT 6

TOPIC 6.7

Mutations

A mutation changes genetic information. Its effect depends on where the change occurs, how it alters a gene product, and the environment in which the phenotype is expressed.



THE CORE CAUSAL CHAIN

DNA CHANGE
sequence or structure

→

RNA / CODONS
message may change

→

PROTEIN
shape or amount

→

PHENOTYPE
variation in context

BY THE END, YOU SHOULD BE ABLE TO...

1. Distinguish substitution, insertion, and deletion mutations.
2. Predict silent, missense, nonsense, and frameshift effects.
3. Explain why mutation outcomes depend on biological context.

CENTRAL IDEA

Mutations create new genetic variation. They may be neutral, harmful, or beneficial rather than automatically changing phenotype in one predictable way.

1 Sequence mutations can change protein instructions

A substitution affects one nucleotide; insertions and deletions may reorganize every codon that follows.

START WITH AN ORIGINAL MESSAGE



ONE SUBSTITUTION: THREE POSSIBLE CODING RESULTS

SILENT

GAA -> GAG

Codon changes, but the amino acid stays the same.

MISSENSE

GAA -> GUA

Codon specifies a different amino acid.

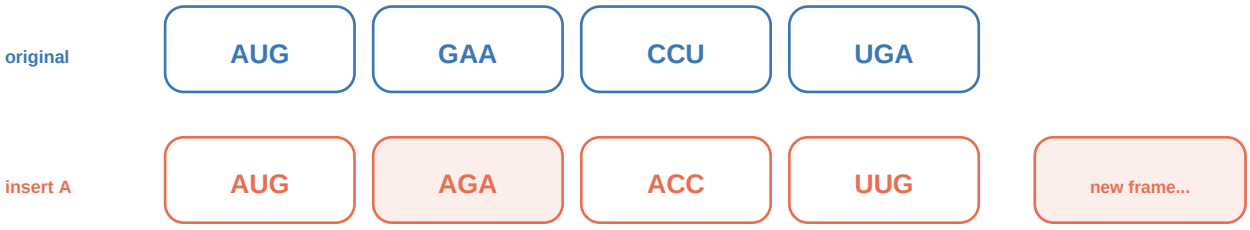
NONSENSE

GAA -> UAA

Codon becomes an early stop signal.

INSERTION OR DELETION

INSERTIONS AND DELETIONS CAN SHIFT THE READING FRAME



A shift changes every downstream codon; an insertion or deletion of 3 bases preserves the frame.

PREDICT THE EFFECT

Ask whether the reading frame changes, whether a stop codon appears, and whether the altered amino acid matters to protein shape or function.

2

Causes and consequences depend on context

Mutations may arise spontaneously or from mutagens, and their effects range from undetectable to major changes in phenotype.

WHERE MUTATIONS COME FROM

SPONTANEOUS ERRORS

DNA replication or repair can occasionally introduce an incorrect nucleotide or fail to correct damage.

MUTAGENS

Radiation and reactive chemicals can damage DNA and increase mutation rate.

MORE THAN SINGLE-BASE CHANGES

CHROMOSOME STRUCTURE OR NUMBER CAN CHANGE

Separation errors can also produce cells with abnormal chromosome numbers.

DELETION
segment lost

DUPLICATION
segment copied

INVERSION
segment reversed

TRANSLOCATION
segment moved

FROM GENOTYPE TO PHENOTYPE

GENE → RNA → PROTEIN → CELL FUNCTION → PHENOTYPE

THE SAME MUTATION CAN HAVE DIFFERENT FITNESS EFFECTS

NEUTRAL

No meaningful change to phenotype or reproductive success.

HARMFUL

Protein or regulation changes in a way that lowers function or fitness.

BENEFICIAL

A phenotype improves survival or reproduction in a particular environment.

FINAL MAP

Mutation creates an allele or chromosome change -> gene product may change -> phenotype may change -> environment determines the fitness effect.