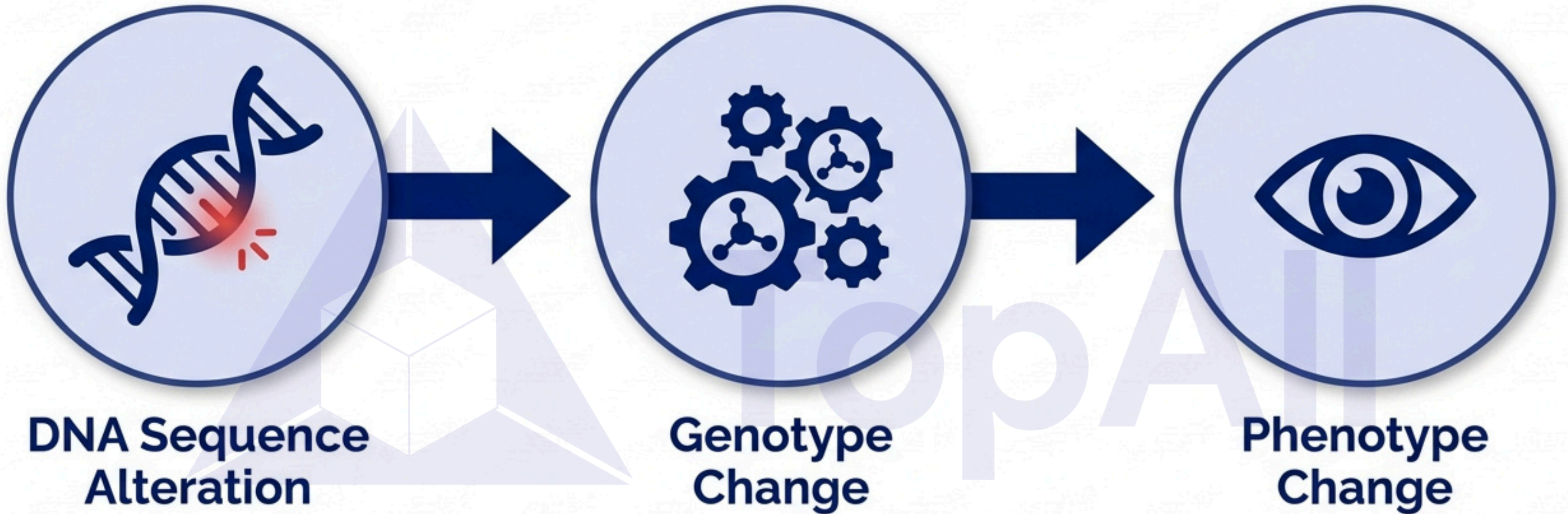


Mutation

Editing the Molecular Blueprint

An overview of DNA sequence changes, mechanisms, and variations.



In addition to recombination, mutation is the primary phenomenon that leads to variation in DNA.

Genetic Mutations

Chromosomal Aberrations

- Loss (Deletion) or Gain (Insertion/Duplication) of a macro-DNA segment.



Commonly observed in cancer cells.

Gene-Level Mutations

- Change in a single base pair or localized sequence.

**Point
Mutation**

**Frameshift
Mutation**

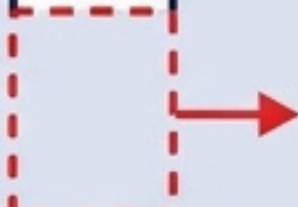
Deletion

Normal

[A]
[B]
[C]
[D]
[E]

Mutated

[A]
[B]
[D]
[E]



Insertion

Normal

[A]
[B]
[C]
[D]
[E]

Mutated

[A]
[B]
[X]
[C]
[D]
[E]

Duplication

Normal

[A]
[B]
[C]
[D]
[E]

Mutated

[A]
[B]
[B]
[C]
[D]
[E]



You've Learned the Theory. Let's Test Them Now.

$$E=mc^2$$



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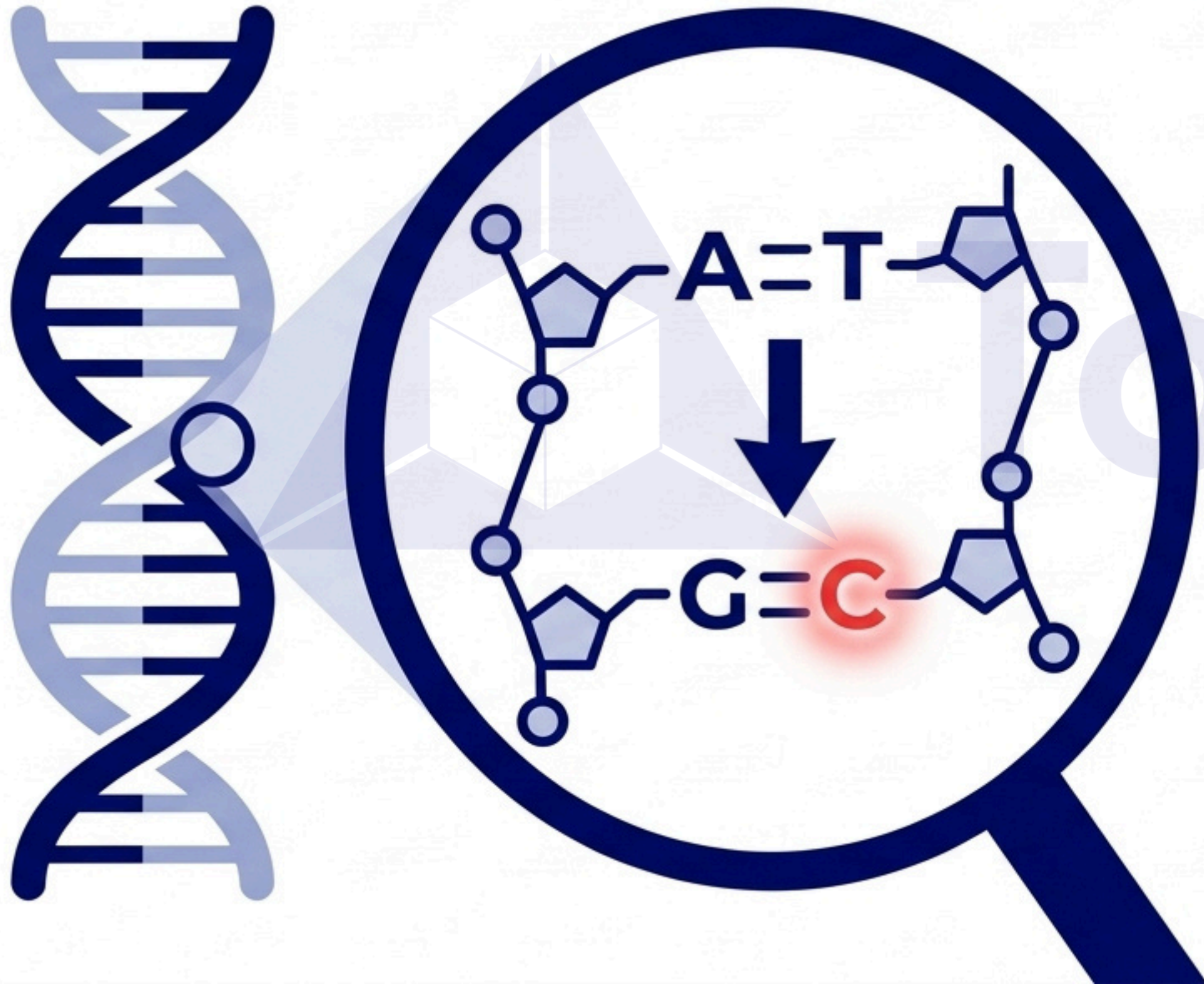
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- » Free Study Material, Videos & Infographics
- » Push Your Limits with Level-Based Challenges
- » Climb the Leaderboard Among Fellow Aspirants
- » Practice Unlimited MCQs for NEET & JEE
- » Get Clear Explanations, Not Just Marks
- » Track Your Performance Smarter



The Base-Pair Zoom



Point Mutation

A change arising from the alteration of a single base pair of DNA.

Classical Example: Sickle Cell Anemia (A single base substitution causes profound physiological changes).

Normal Reading Frame



Frameshift Mutation



Takeaway:

Deletions or insertions of single base pairs shift the entire reading frame, altering every subsequent codon.

Mechanism Diagnostic Matrix

Point Mutation

Mechanism: Base Substitution
(1:1 swap).

Scope of Impact: Usually affects only one amino acid (localized).

Example: Sickle Cell Anemia.

Frameshift Mutation

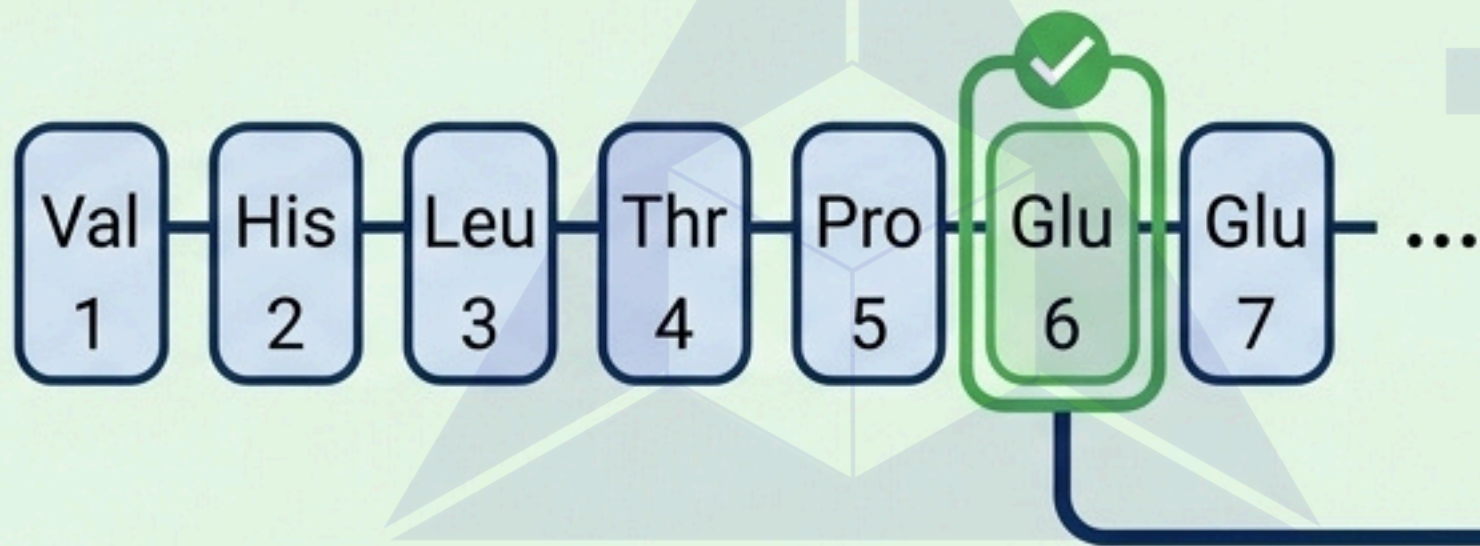
Mechanism: Base Insertion or Deletion.

Scope of Impact: Shifts the entire sequence downstream; completely changes the resulting protein.

Example: Often results in early stop codons or non-functional proteins.

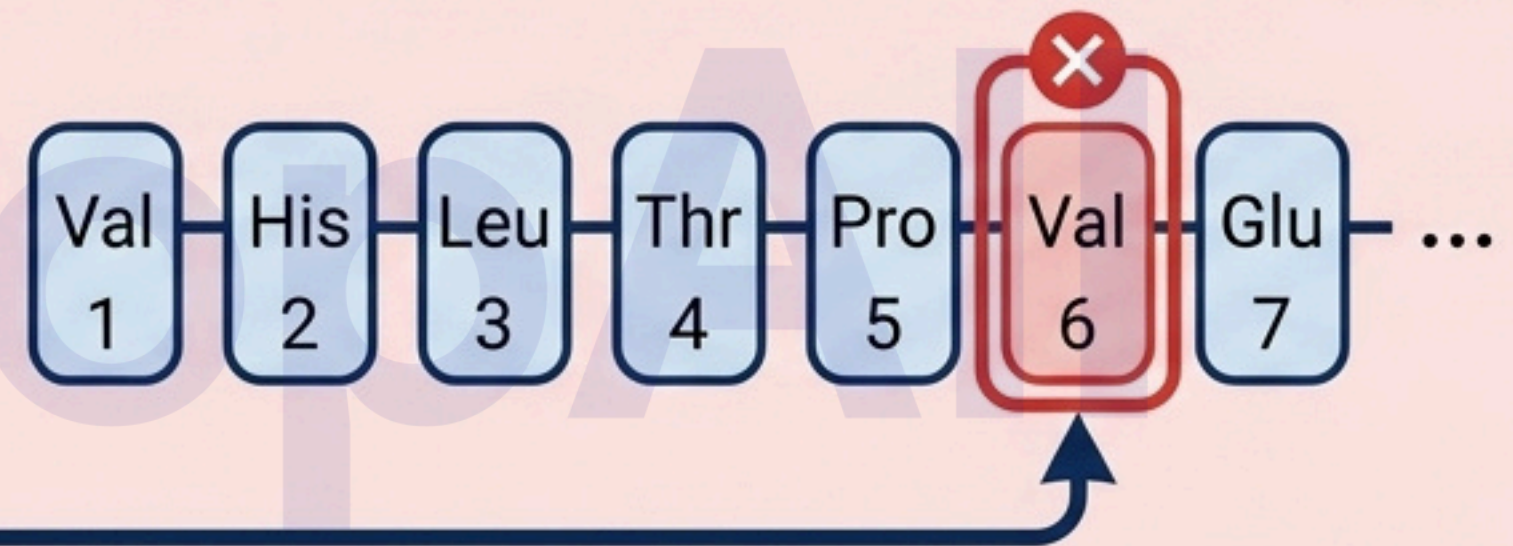
The Molecular Typo: Sickle Cell Anemia

Normal Individual (HbA)



Codon **GAG** codes for Glutamic Acid (Glu).

Sickle-Cell Individual (HbS)



Single base substitution changes codon to **GUG**, coding for Valine (Val) at the 6th position.

Mutant Haemoglobin (HbS) contains Valine instead of Glutamic Acid.



Under low oxygen tension, the mutant haemoglobin molecules undergo physical polymerization.



Cell Shape Change

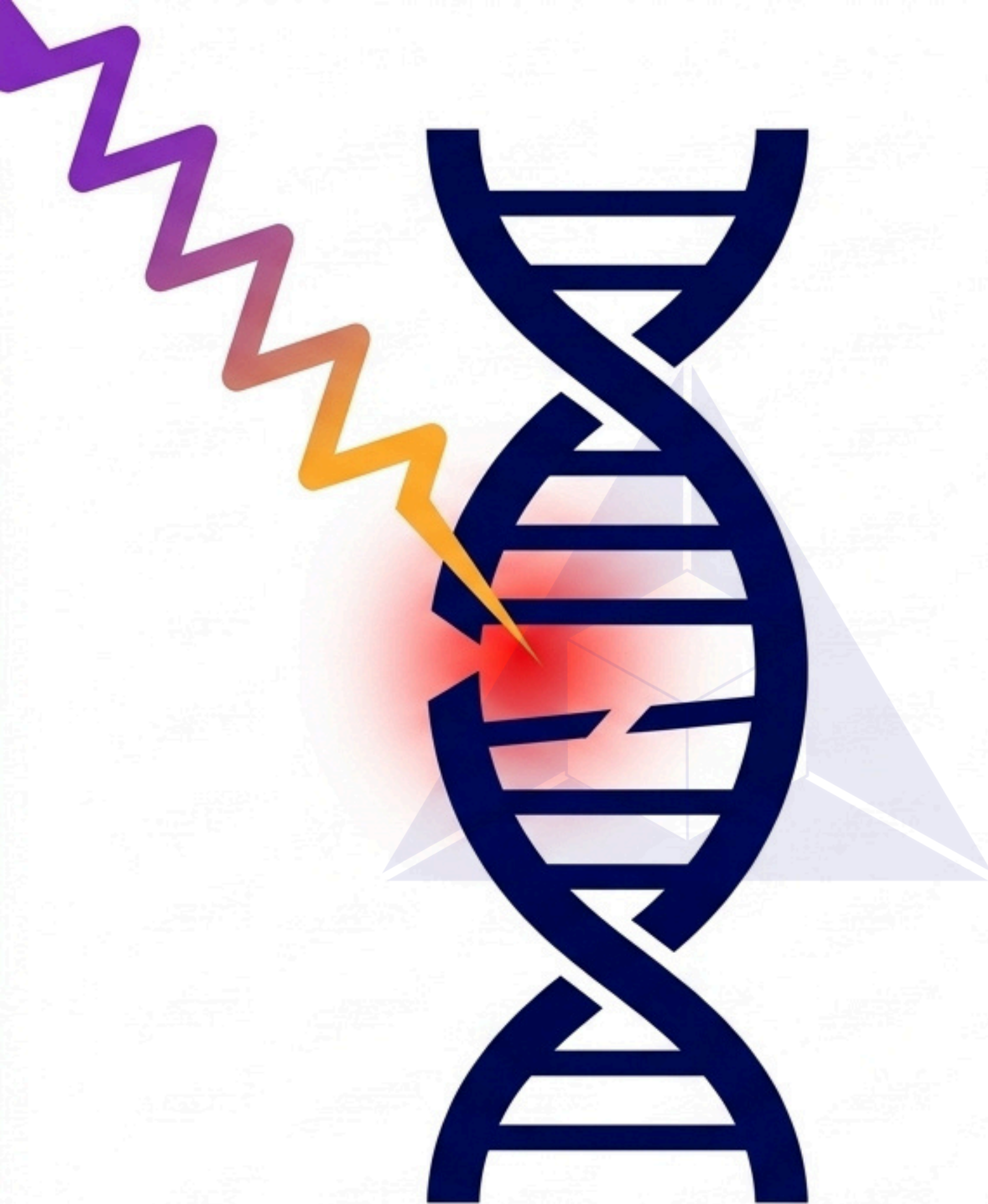


Normal
biconcave disc



Elongated sickle-like
structure

Note: Heterozygous individuals (HbA/HbS) are carriers and exhibit the sickle-cell trait but are generally unaffected.



The Catalysts: **Mutagens**

Chemical or physical factors that induce alterations in the DNA sequence.

Specific Callout: UV radiation acts as a powerful physical mutagen, physically damaging the structural integrity of the DNA helix.



Mutation creates genetic variation



While mutations can result in disorders, they are simultaneously the essential engine providing the genetic variation necessary for evolution.



You've Learned the Theory.

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