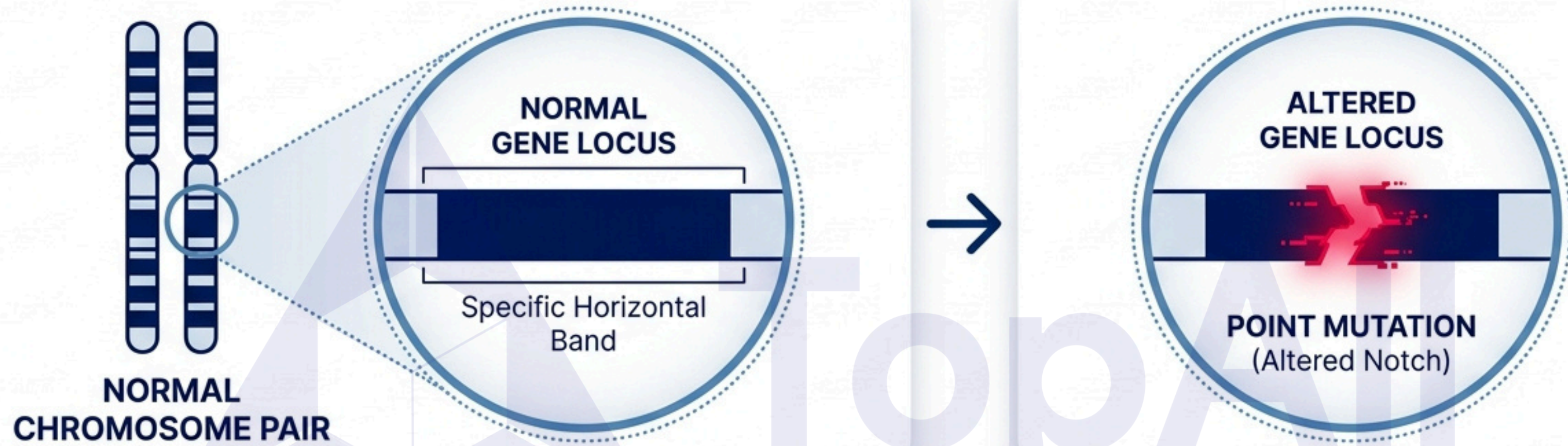


Mendelian Disorders

Decoding the inheritance patterns of single-gene mutations.

The Clinical Study Board: Mendelian Disorders



GENETIC ALTERATION OVERVIEW

Mendelian Disorders are genetic diseases determined primarily by an alteration or mutation in a single gene. They are transmitted to offspring following Mendel's principles of inheritance.

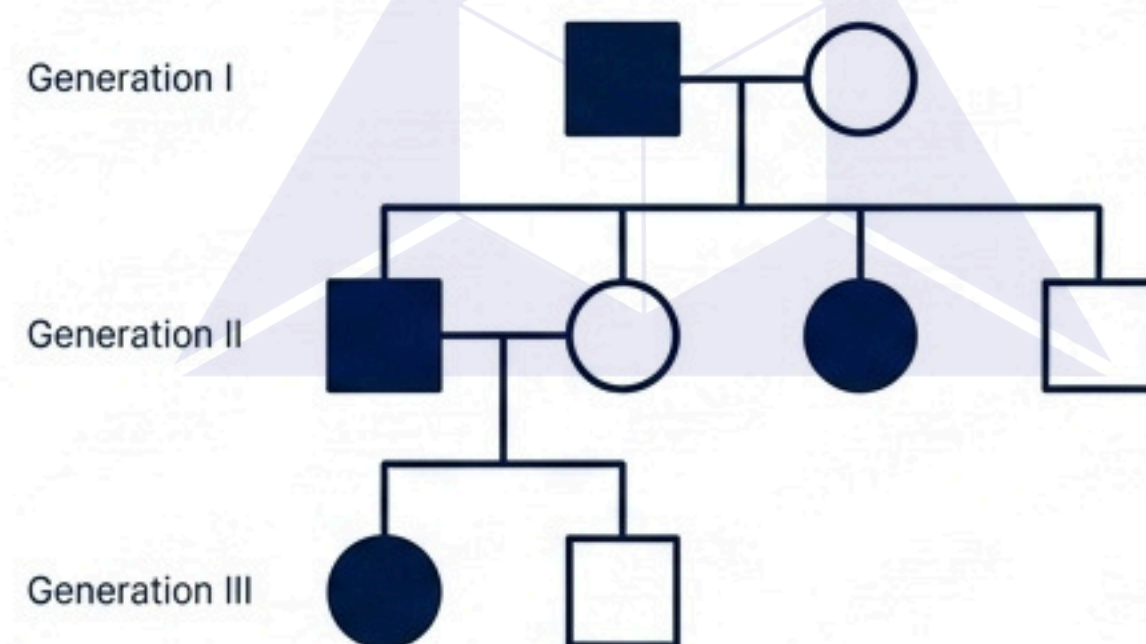


Unlike chromosomal disorders (like Down's syndrome) which involve whole extra or missing chromosomes, **Mendelian disorders** are **pinpoint errors** in the DNA sequence.

Expression Rules: Zygosity

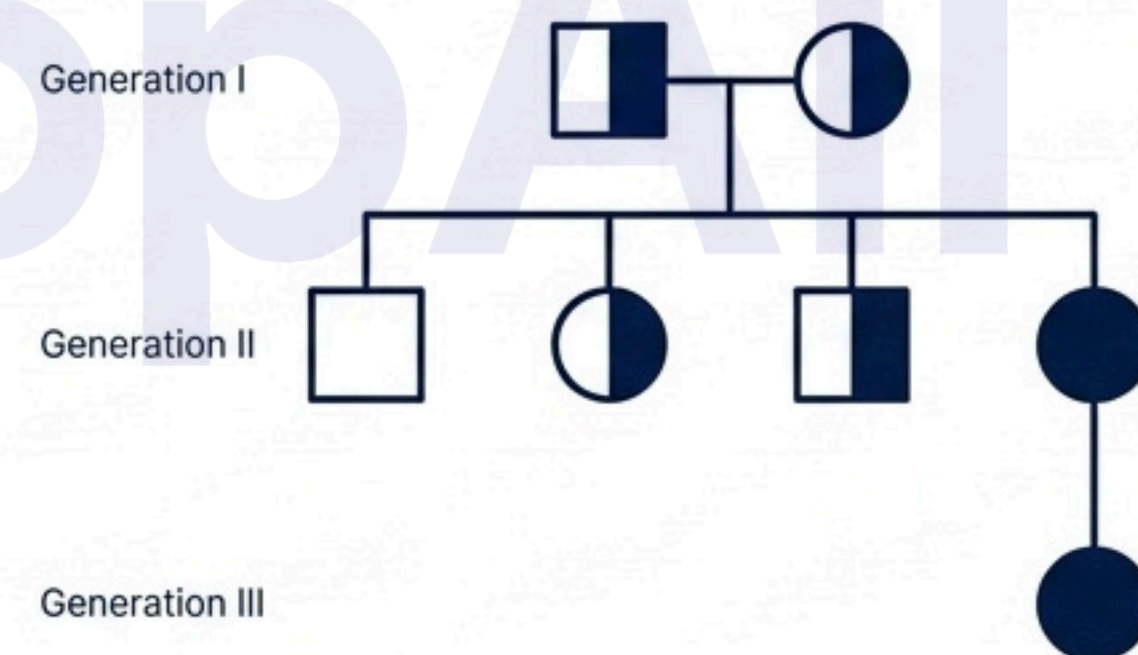
Dominant Inheritance

Requires only one mutant allele to express the trait.



Recessive Inheritance

Requires two mutant alleles to express the trait.



Pedigree analysis is a crucial clinical tool used to trace these inheritance patterns over generations.



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

$$E=mc^2$$



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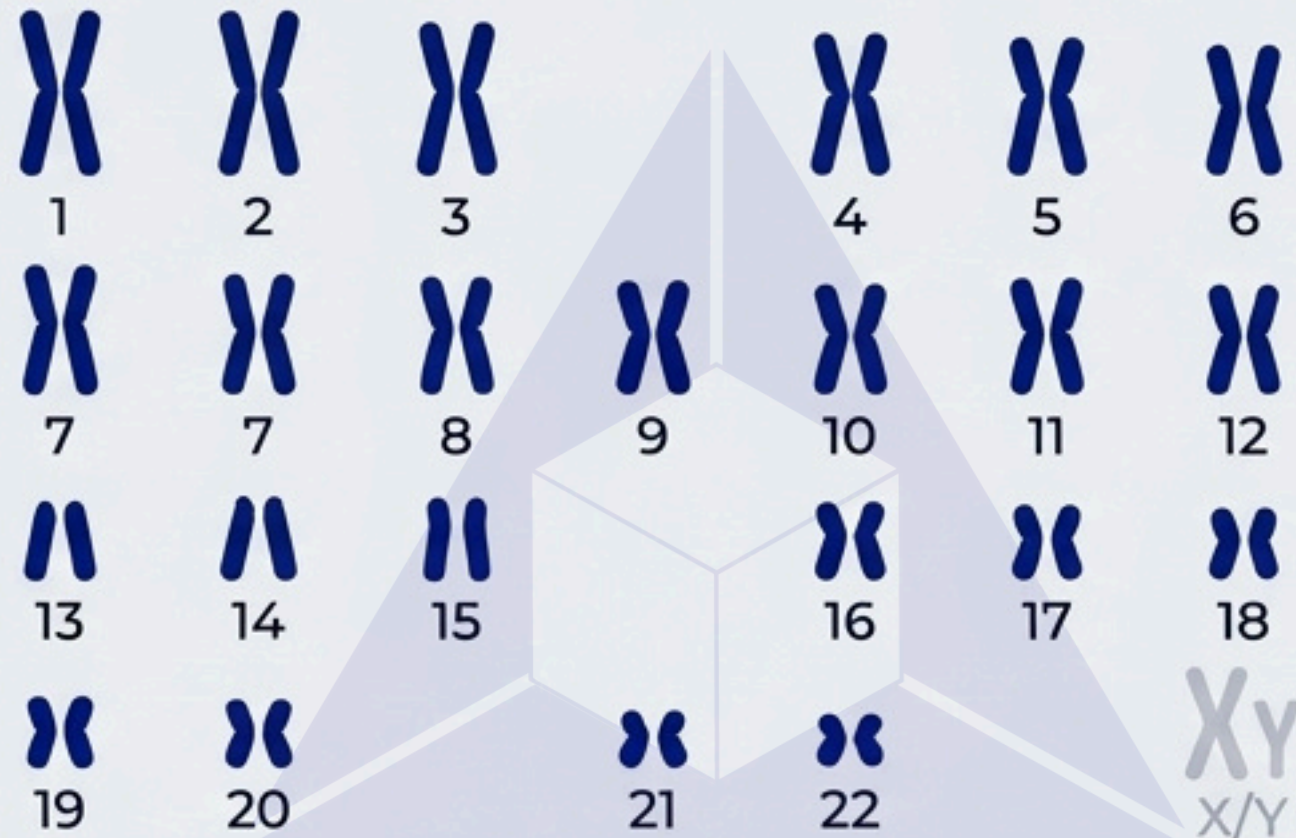
Key Highlights

- » 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



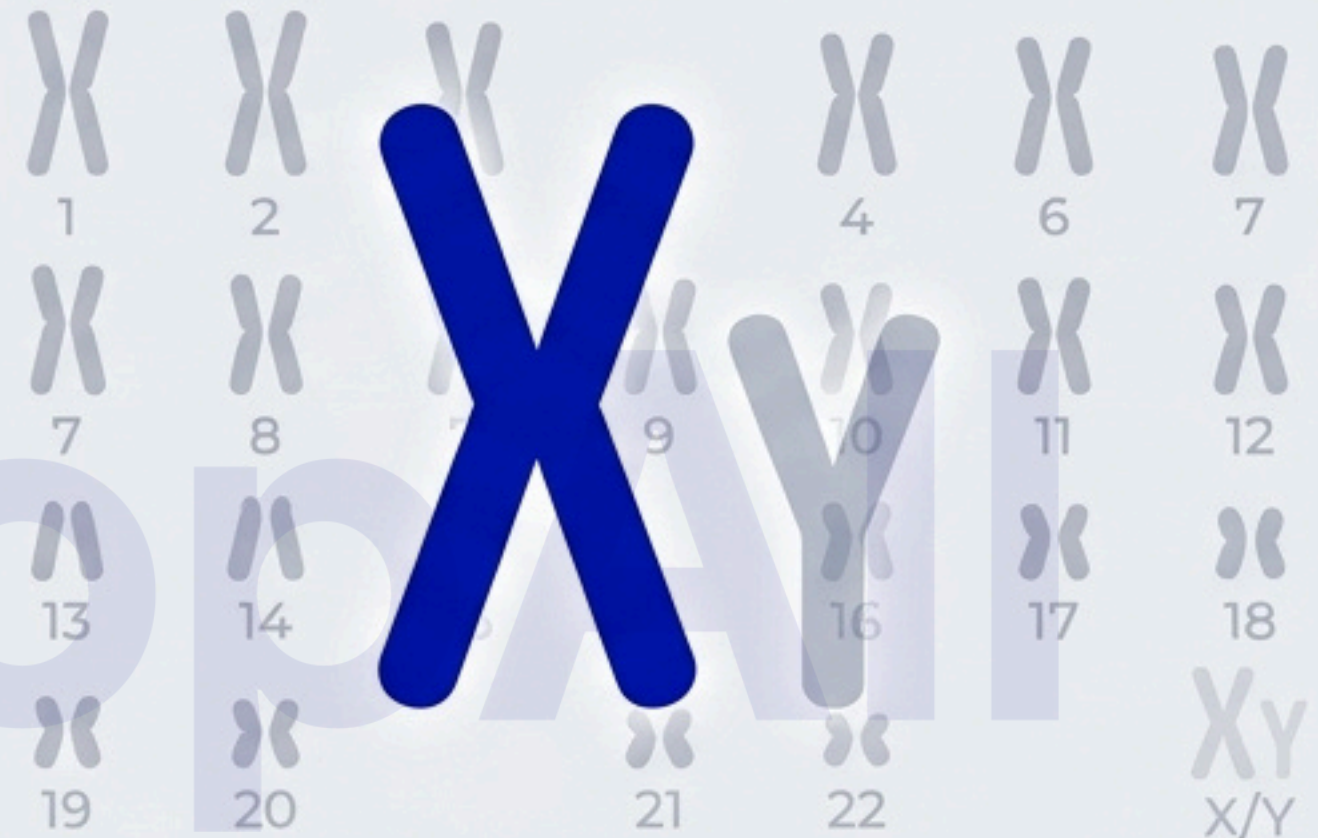
Locus Rules: Chromosomal Location

Autosomal Locus



Affects males and females with equal probability.

Sex-Linked Locus



Expression probability is heavily dependent on the biological sex of the individual.



Common Misconception: Assuming sex-linked means it only affects one sex. Females can have X-linked recessive disorders, but it is much rarer. A female needs two mutant copies (from a carrier mother and affected father), whereas a male only needs one.

Classic Examples in Human Genetics

Sex-Linked Recessive

Haemophilia



A defect in the cascade of proteins involved in blood clotting, causing non-stop bleeding from simple cuts.

Queen Victoria was a famous carrier, transmitting the disease to royal descendants.

Sex-Linked Recessive

Colour Blindness

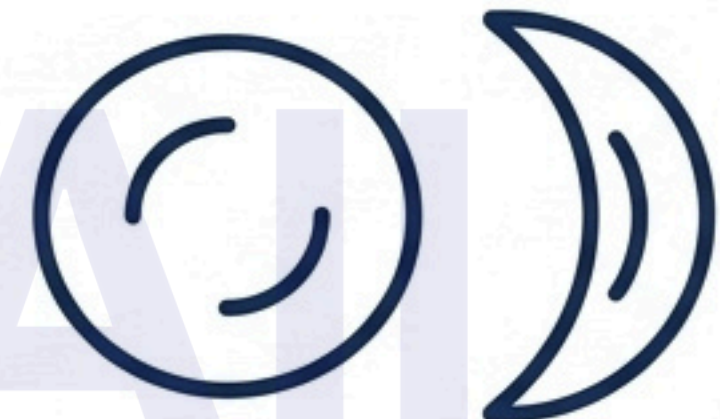


A defect in the red or green cones of the eye, located on the X chromosome.

Occurs in ~8% of males, but only ~0.4% of females.

Autosomal Recessive

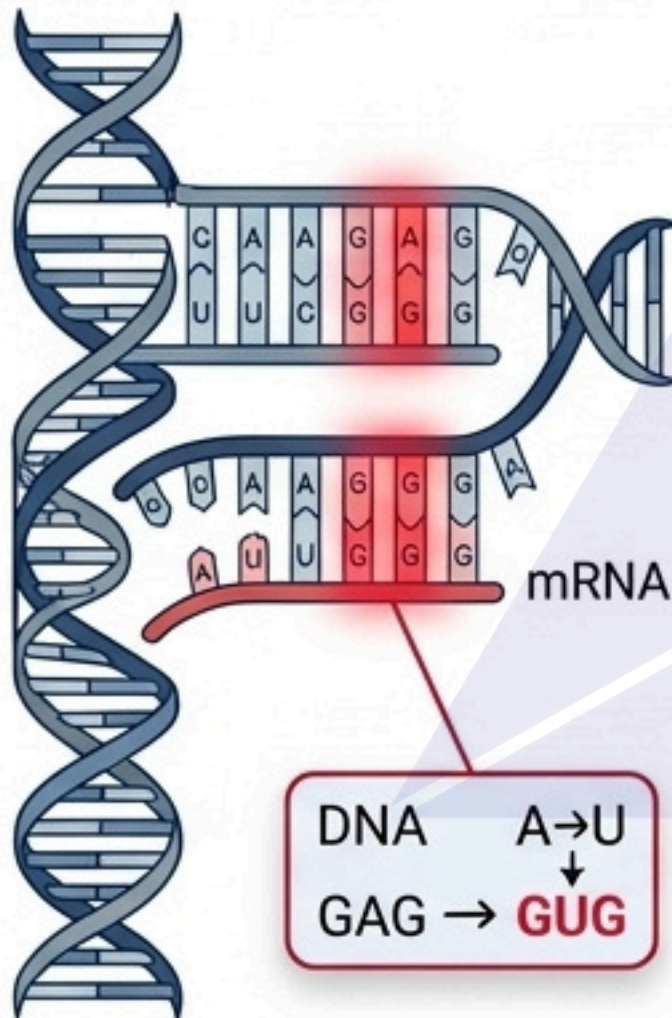
Sickle-Cell Anaemia



Controlled by alleles HbA and HbS. Only homozygous (HbS HbS) individuals show the diseased phenotype.

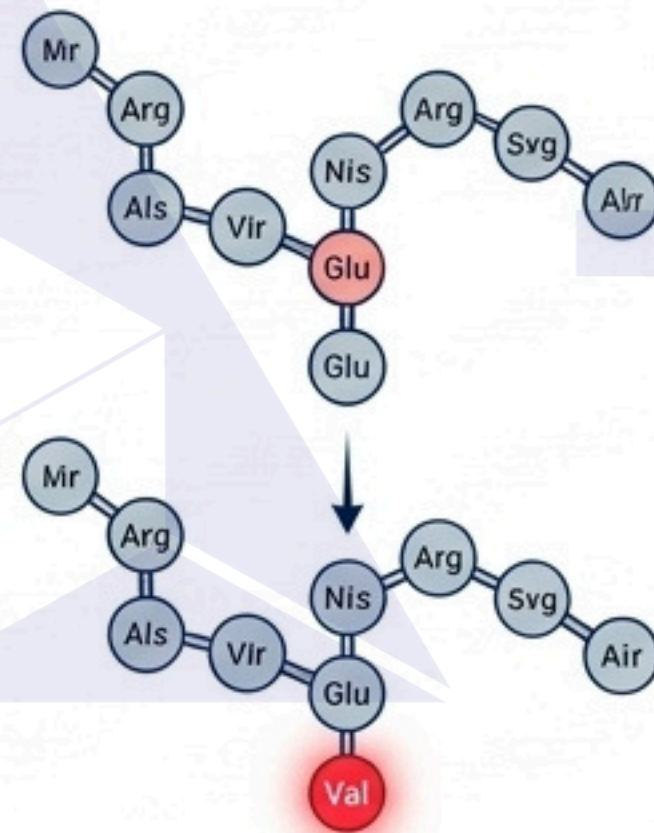
Molecular Deep-Dive: Sickle-Cell Anaemia

1. The Genetic Code



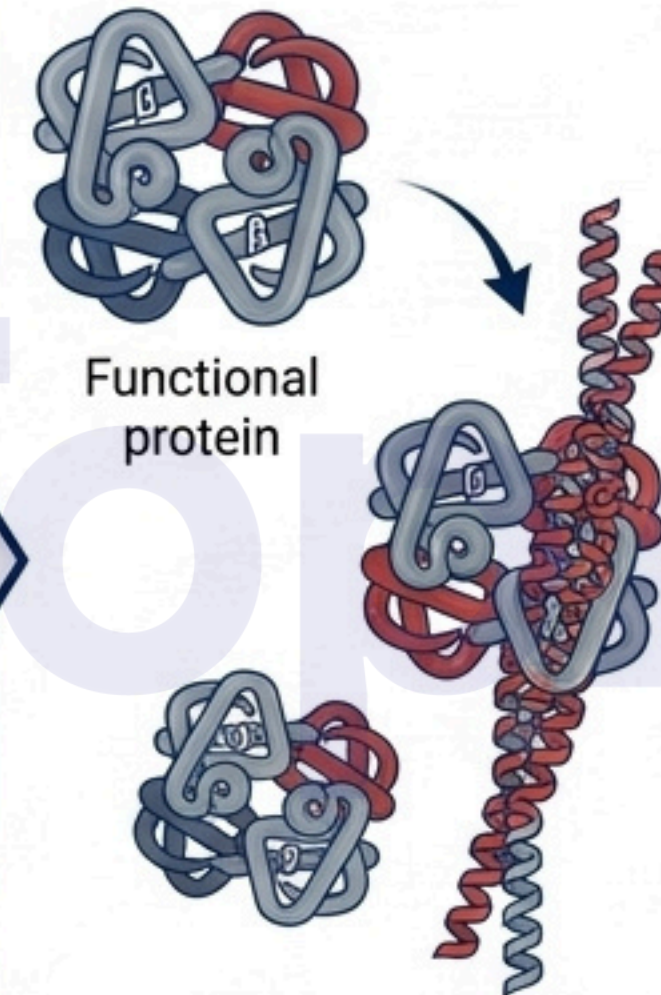
The 6th codon of the beta-globin gene mutates:
GAG → **GUG**.

2. Amino Acid Substitution



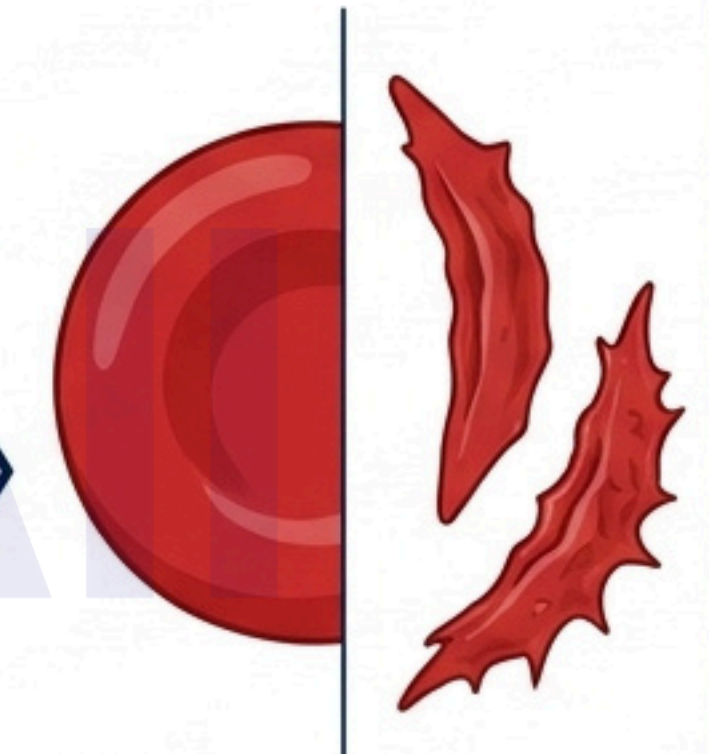
Glutamic acid (Glu) is replaced by Valine (Val).

3. Protein Alteration



Mutant haemoglobin undergoes polymerisation under low oxygen tension.

4. Cellular Phenotype

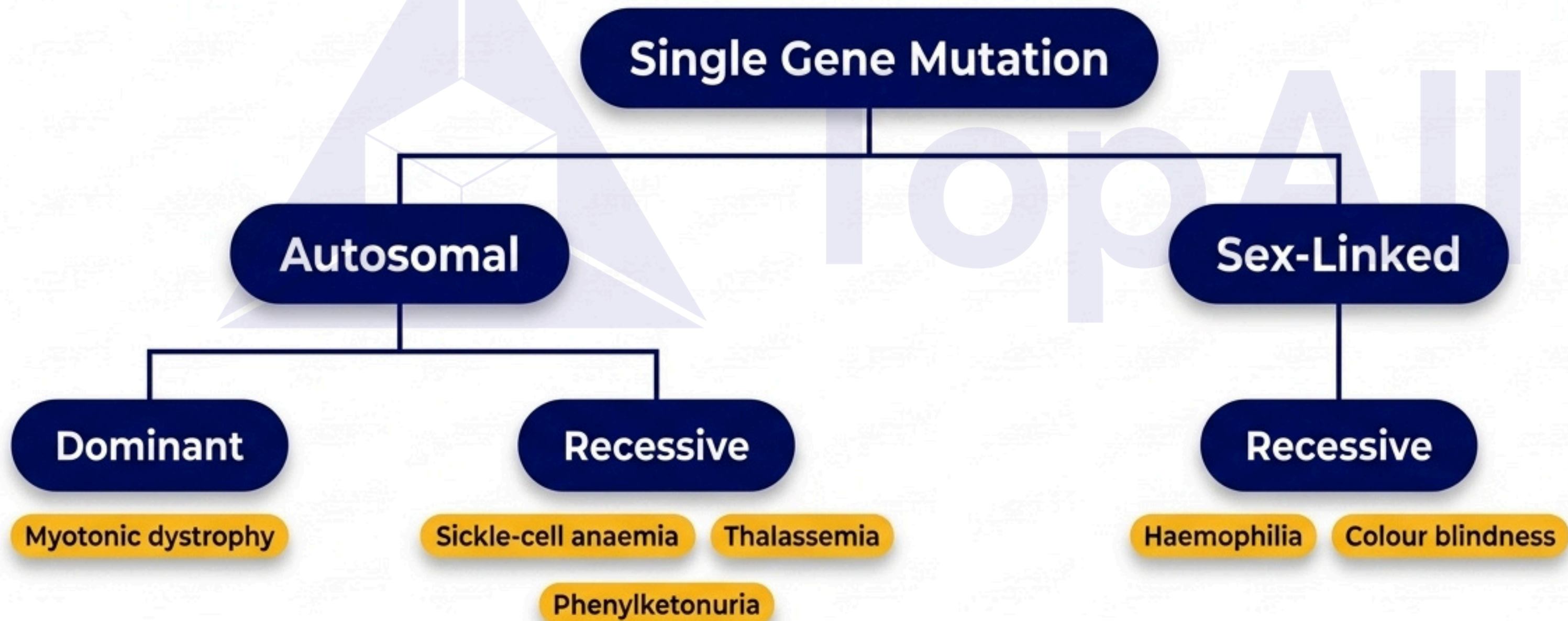


Normal RBC

Sickle RBC

RBC shape distorts, leading to the clinical symptoms of anaemia.

Mendelian disorders follow inheritance patterns.





You've Learned the Theory.

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