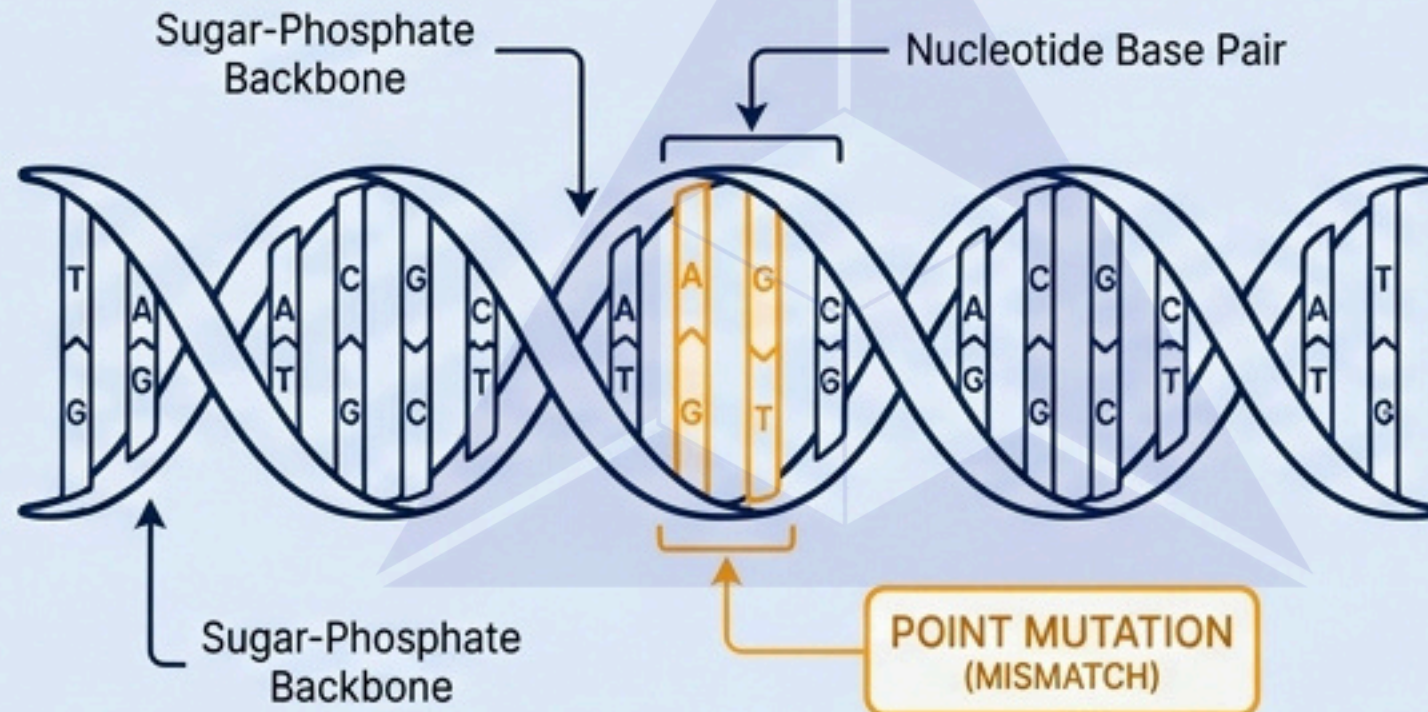


Genetic Disorders: The Clinical Blueprint

Mapping the mechanics of altered genes and chromosomes.

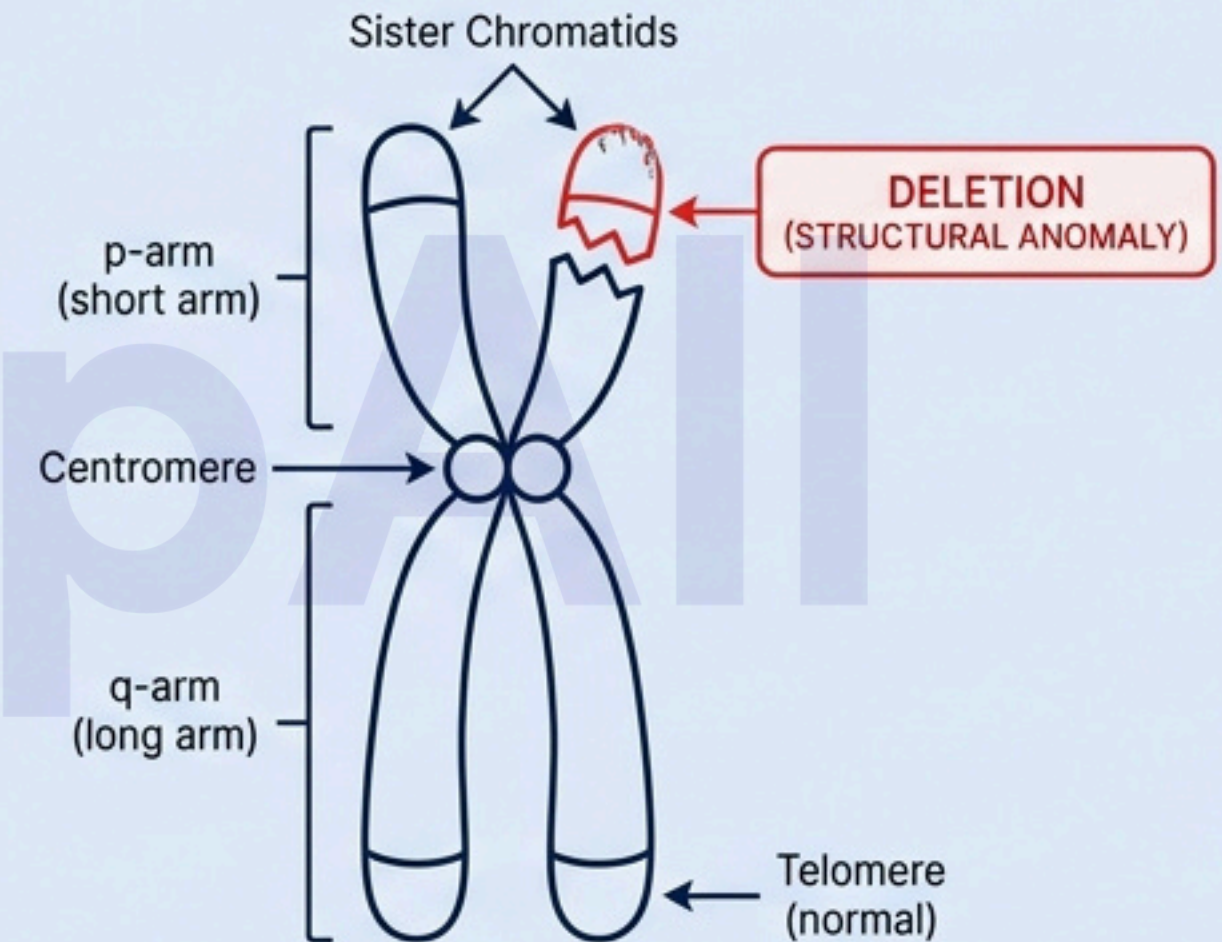
THE MICROSCOPIC ERROR: ALTERED GENE



A single base pair substitution disrupts the genetic code, potentially leading to an incorrect protein product or no protein at all.

THE MICROSCOPIC ERROR: ALTERED GENE

THE MACROSCOPIC ERROR: ALTERED CHROMOSOME



A loss of a chromosome segment deletes multiple genes, causing significant developmental and functional impairments.

THE MACROSCOPIC ERROR: ALTERED CHROMOSOME

The Root Cause: Mutation

The Pathway of Expression



Altered DNA Sequence

Loss, gain, or substitution of base pairs.



Altered Genotype

The genetic code is permanently modified.



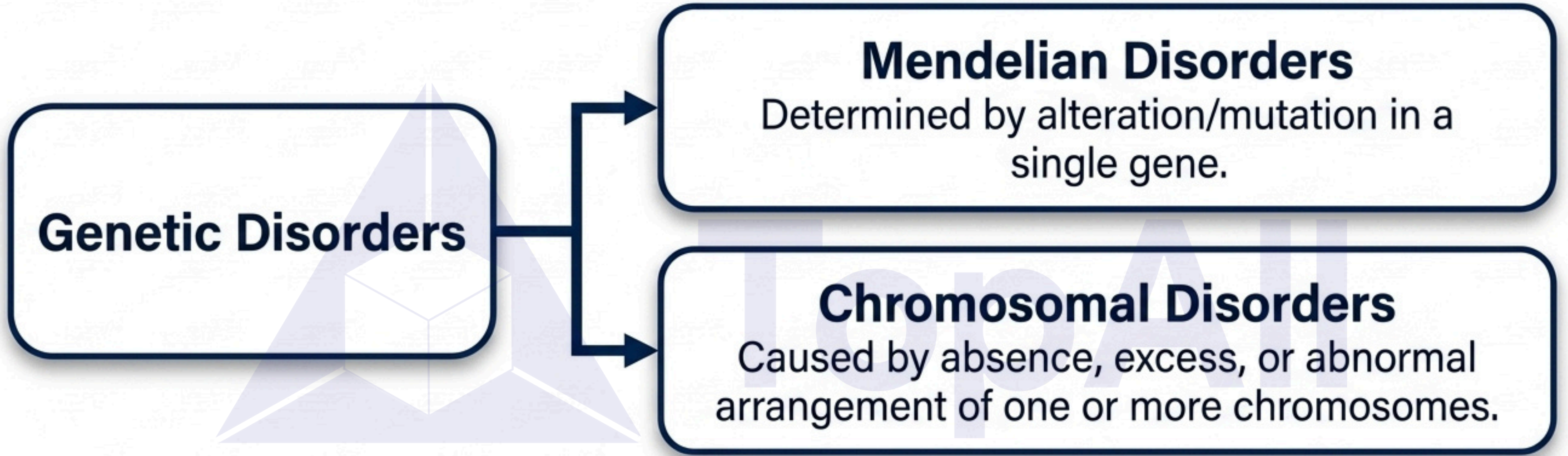
Altered Phenotype

Abnormal physical expression or metabolic function.

Quick-Tip Exam Note

Mutagens: Physical (UV radiation) or chemical factors can actively induce these DNA alterations.

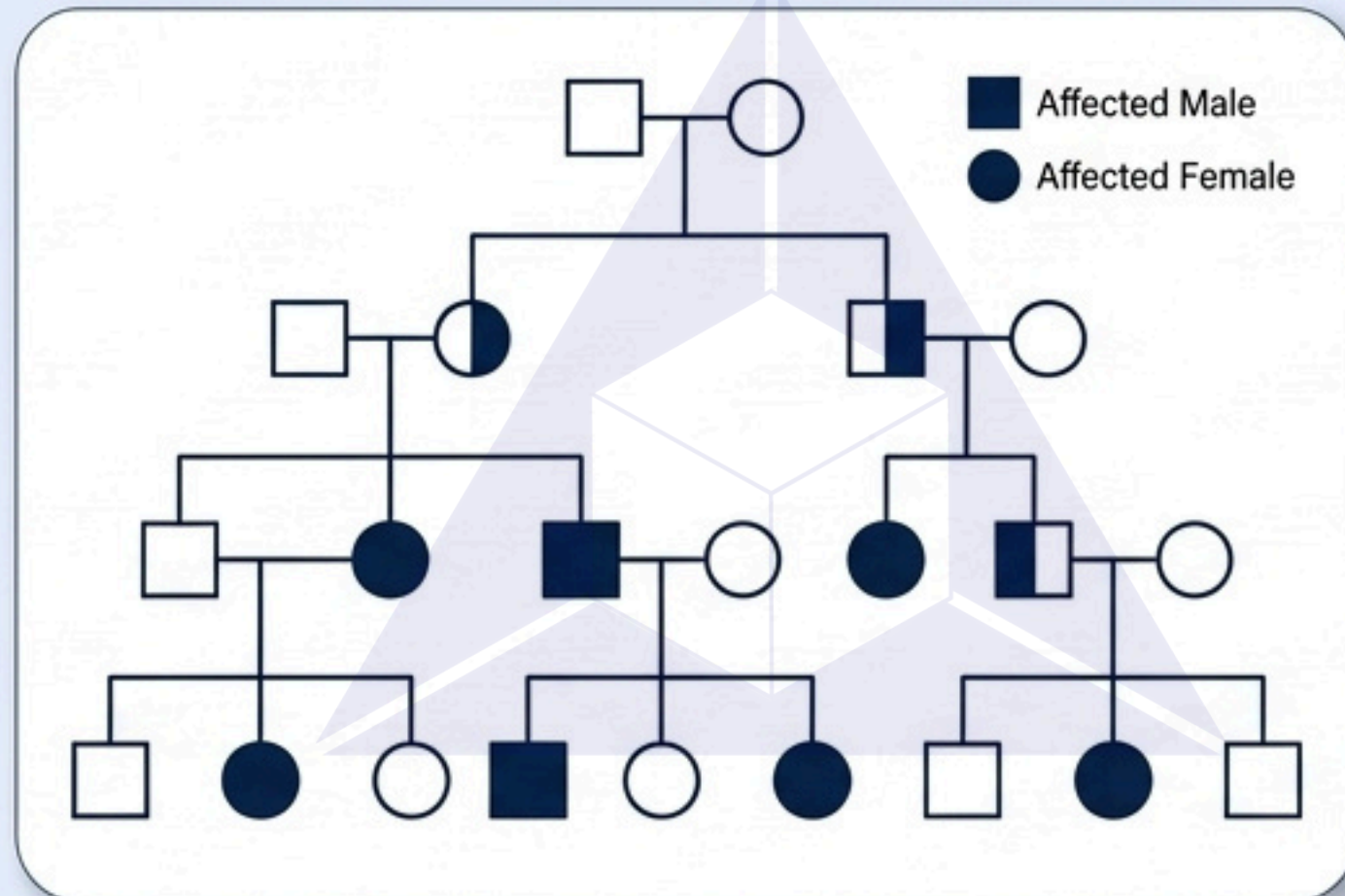
The Genetic Disorder Taxonomy



**KEY TAKEAWAY: Single gene error = Mendelian.
Chromosome number/structure error = Chromosomal.**

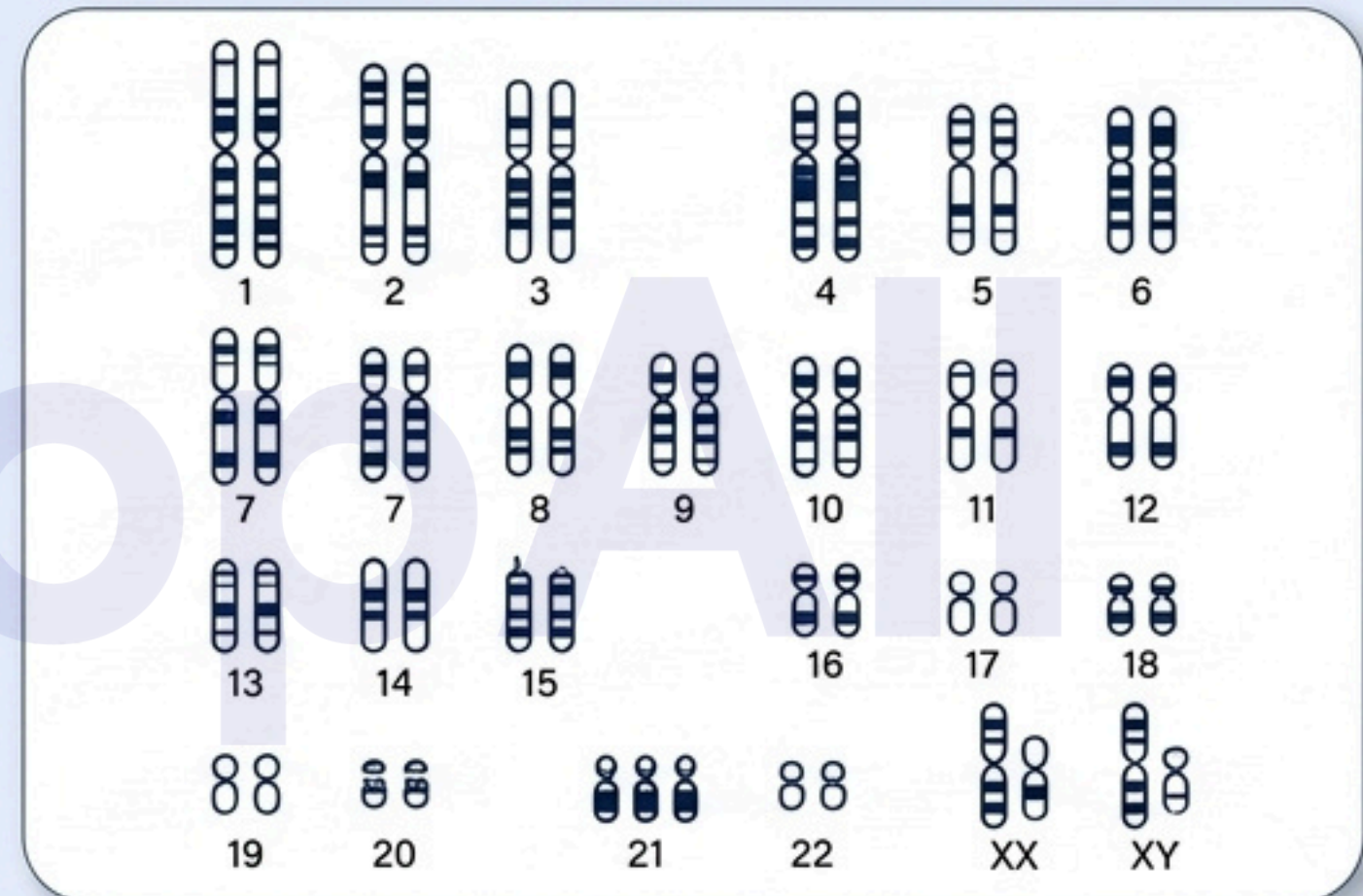
Tracing the Defect: Clinical Detection

Mendelian Detection: Pedigree Analysis



Traces the inheritance pattern of a single-gene trait over generations. Essential because controlled crosses aren't possible in humans.

Chromosomal Detection: Karyotype Analysis



Visual map of an individual's chromosomes. Directly reveals aneuploidy (gain/loss) or structural anomalies at a cellular level.

Mendelian Mechanics: The Single Gene Error

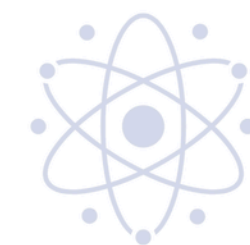
Transmitted to offspring along standard Mendelian inheritance principles.

	Dominant (Expressed even in heterozygotes)	Recessive (Expressed only in homozygotes; heterozygotes are asymptomatic 'carriers')
Autosomal (Linked to chromosome pairs 1-22)	(Example: Myotonic dystrophy)	- Sickle-cell anaemia - Phenylketonuria - Thalassemia
Sex-Linked (Linked to X/Y chromosomes)		- Haemophilia - Colour blindness



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

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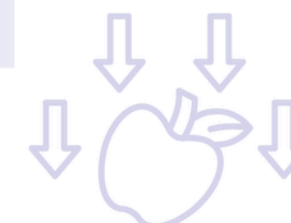
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- » Track Your Performance Smarter



Group 1: Mendelian Blood Disorders (Sickle-Cell)

The Gene

Autosomal Recessive. HbA (normal) vs HbS (mutant) alleles.

The Point Mutation

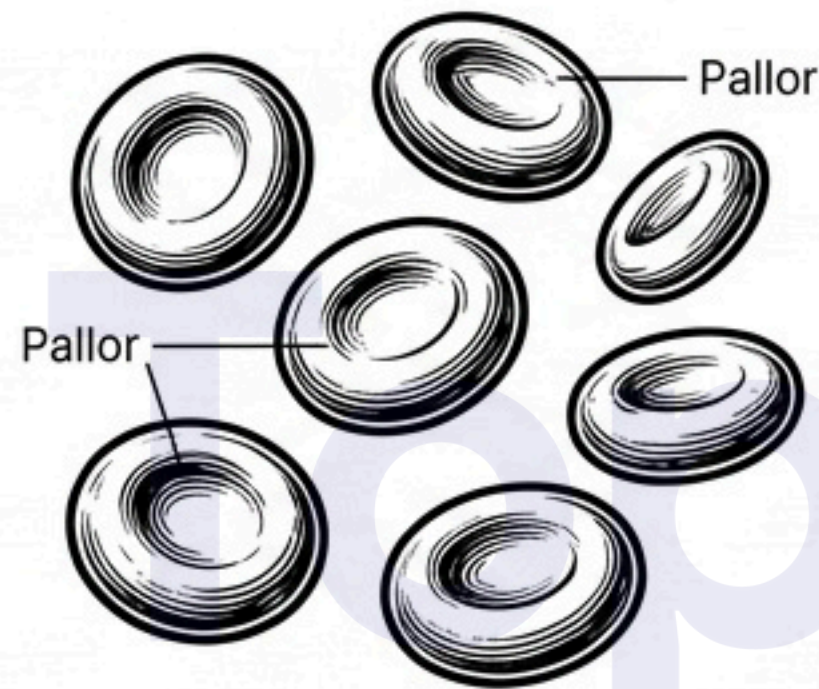
A single base substitution at the 6th codon of the beta-globin gene: GAG -> GUG.

The Protein

Substitution of Glutamic acid (Glu) by Valine (Val).

The Cell

Under low oxygen, mutant haemoglobin polymerises.



Normal Biconcave RBC

Flexible, donut-shaped cells for efficient oxygen transport.



Sickle RBC

Rigid, crescent-shaped cells that block blood flow and reduce oxygen delivery.

Thalassemia vs. Sickle-Cell Anaemia

⚠ Mistake Card

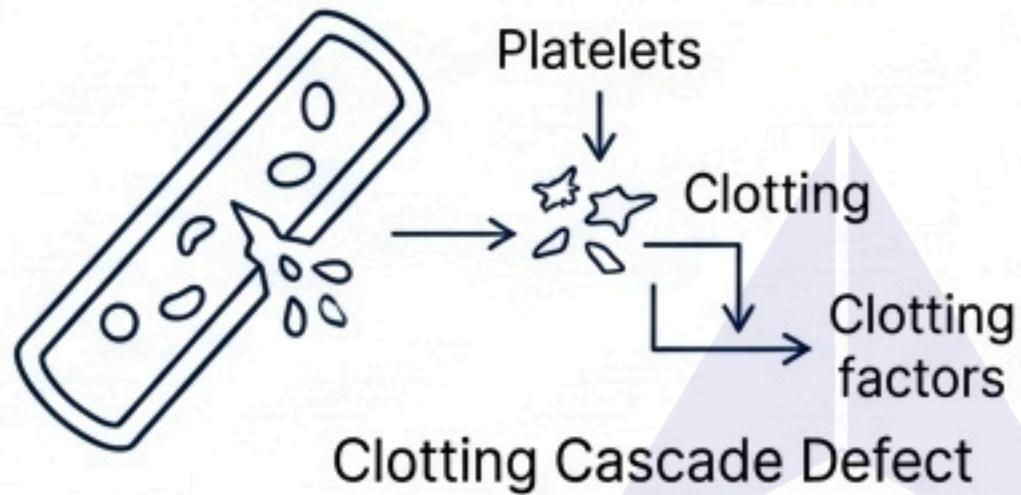
Don't confuse the two! Sickle-cell is a QUALITATIVE problem (incorrectly functioning globin). Thalassemia is a QUANTITATIVE problem (too few globin molecules synthesized).

Thalassemia Breakdown

Alpha Thalassemia	Beta Thalassemia
• Affected alpha globin chain. • Controlled by two linked genes: HBA1 and HBA2. • Located on Chromosome 16.	• Affected beta globin chain. • Controlled by a single gene: HBB. • Located on Chromosome 11.

Commonalities: Both are Autosomal Recessive. Both result in abnormal haemoglobin and anaemia.

Group 2: Sex-Linked Mendelian Disorders

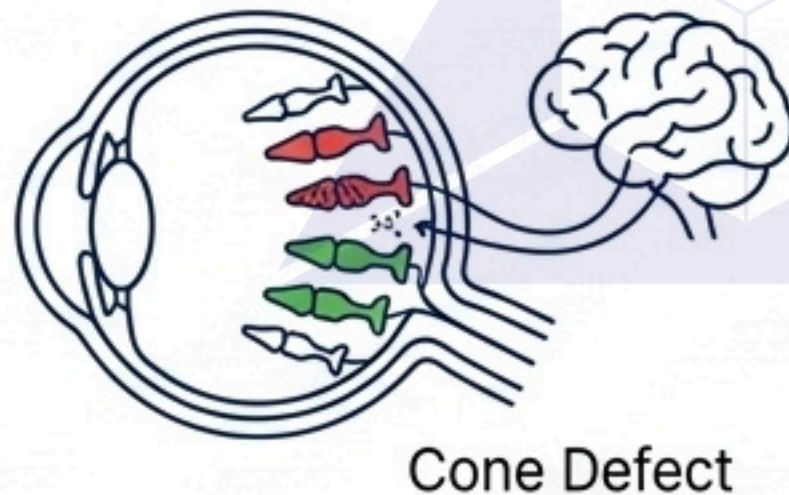


Haemophilia

Mechanism: Defect in the blood clotting protein cascade. A simple cut causes non-stop bleeding.

Pedigree Note

Famous example is Queen Victoria's family tree (she was an unaffected carrier transmitting to male progeny).



Colour Blindness

Mechanism: Defect in red/green eye cones preventing colour discrimination.

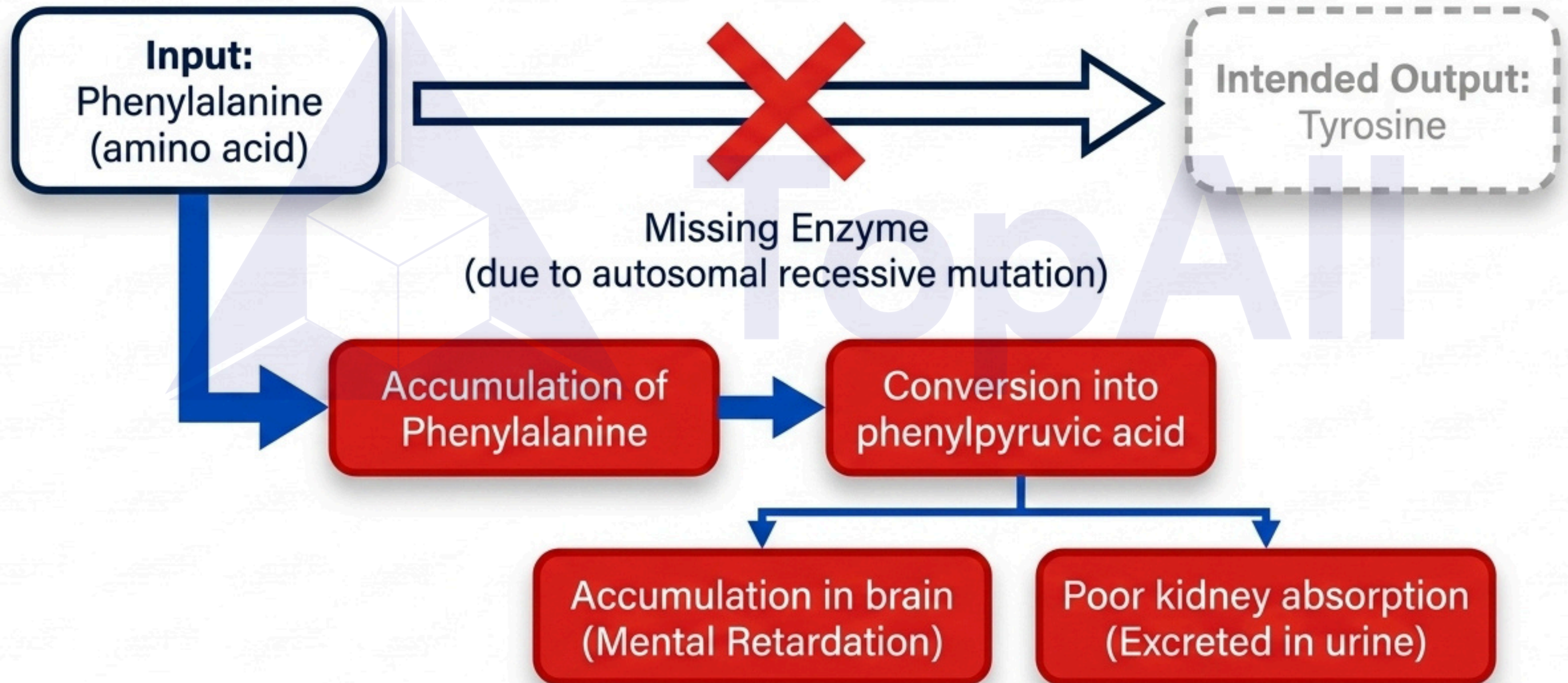


Quick-Tip: Why does Colour Blindness affect 8% of males but only 0.4% of females?

It is an X-linked recessive trait. Males only have one X chromosome—a single defective gene causes the disease. Females have two X chromosomes, requiring two defective genes to express the trait.

Group 3: Mendelian Metabolic Disorders

Disease Focus: Phenylketonuria (PKU)



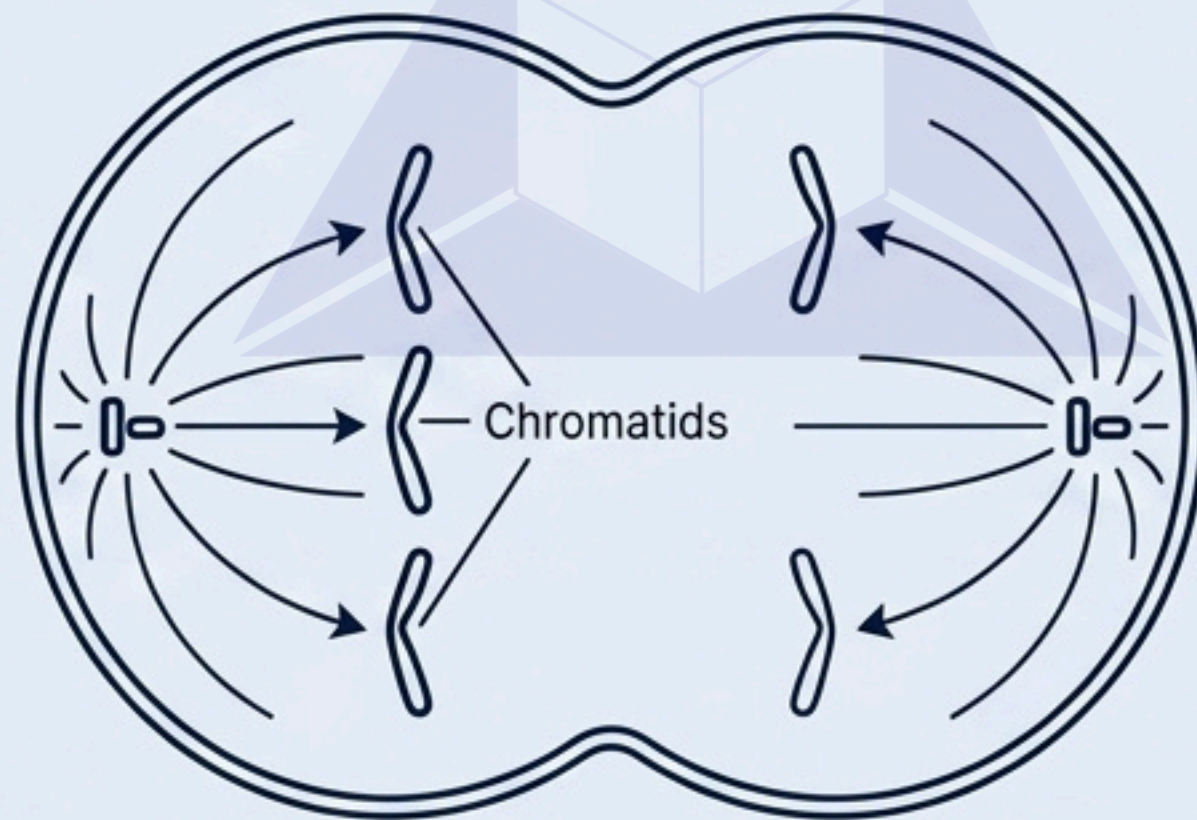
Chromosomal Mechanics: The Structural Error

Caused by the absence, excess, or abnormal arrangement of whole chromosomes.

Aneuploidy

Definition: Gain or loss of individual chromosomes.

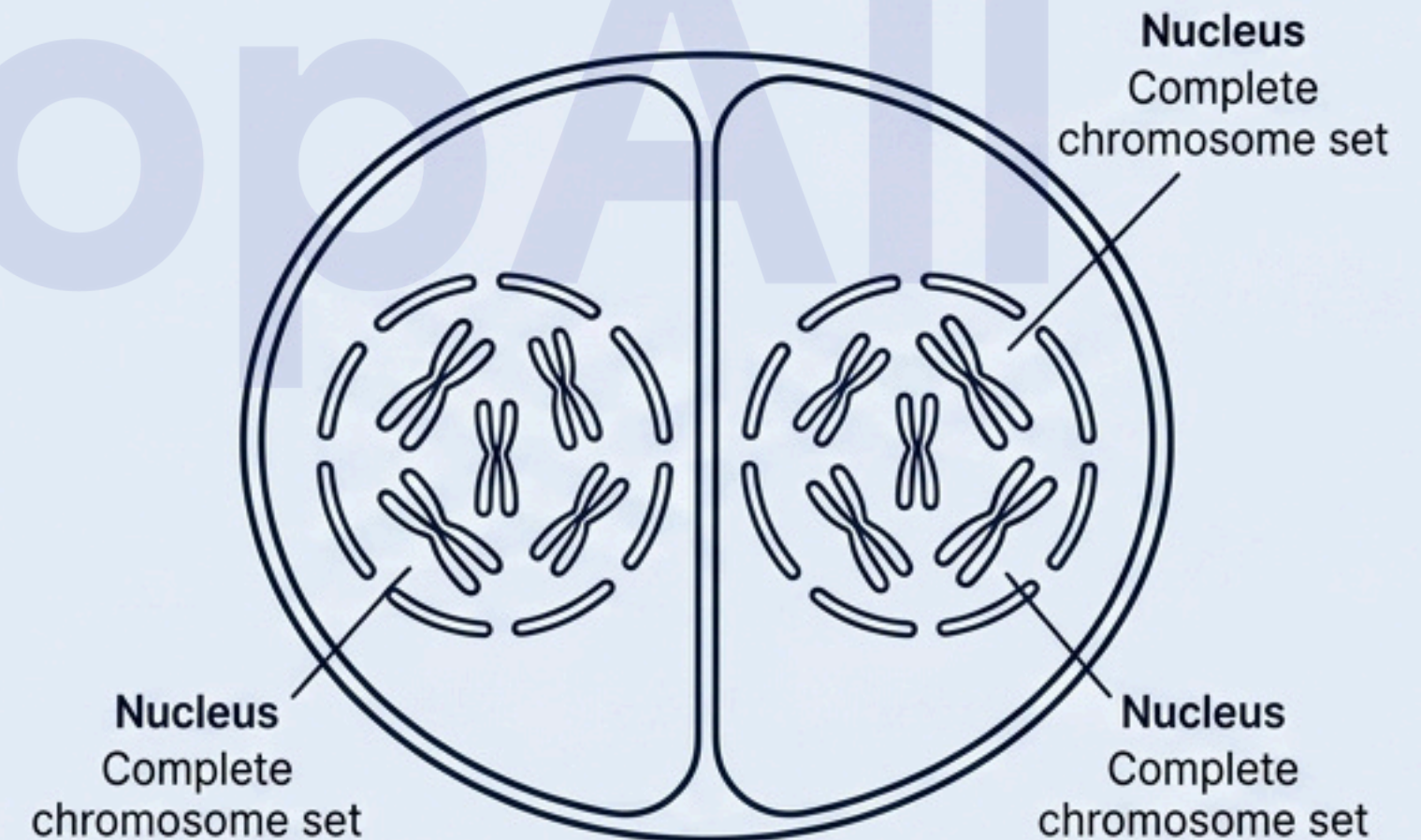
Mechanism: Failure of segregation of chromatids during the cell division cycle.



Polyploidy

Definition: Increase in a whole set of chromosomes (common in plants).

Mechanism: Failure of cytokinesis after the telophase stage.



Autosomal Aneuploidy: Down's Syndrome

Patient Profile

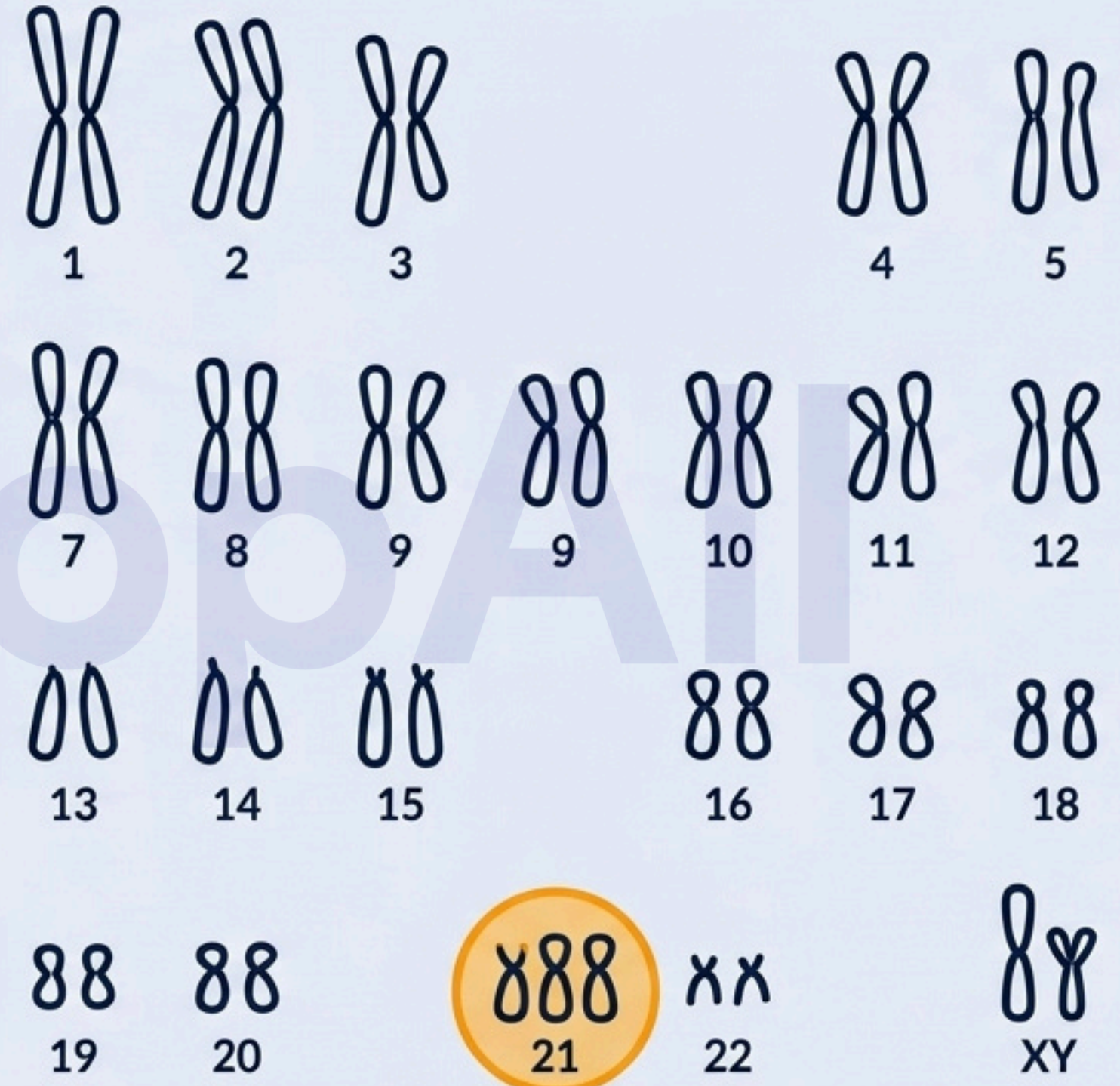
The Chromosomal Math

Formula: 47 Chromosomes (Trisomy 21)

Defect: Presence of an additional copy of chromosome 21. First described by Langdon Down (1866).

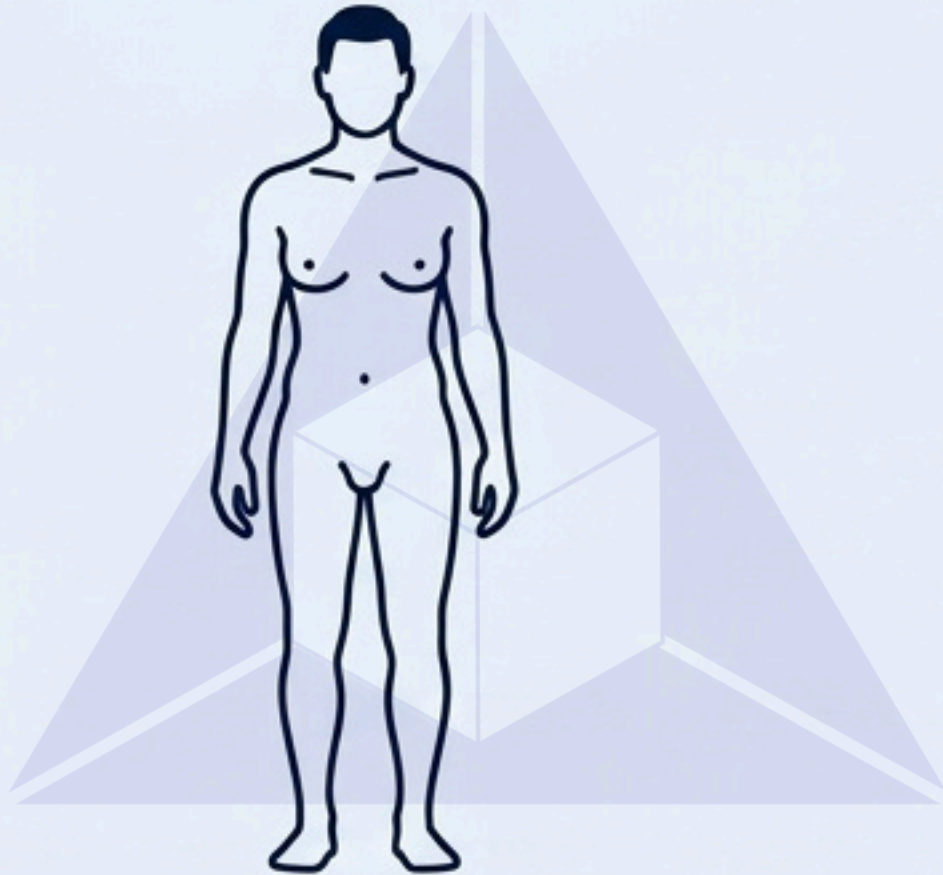
Clinical Features

-  Short stature with small round head.
-  Furrowed, big, wrinkled tongue & partially open mouth.
-  Broad palm with characteristic palm crease.
-  Retarded physical, psychomotor, and mental development.
-  Congenital heart disease.



Sex-Chromosome Aneuploidies

Klinefelter's Syndrome



- **Karyotype:** 47, XXY (Gain of an X chromosome in a male).
- **Phenotype:** Overall masculine development, but expresses feminine characteristics. Tall stature.
- **Status:** Sterile.

Turner's Syndrome



- **Karyotype:** 45, XO (Absence of one X chromosome in a female).
- **Phenotype:** Rudimentary ovaries, lack of secondary sexual characteristics. Short stature.
- **Status:** Sterile.



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The Clinical Blueprint: Master Synthesis

	Mendelian Disorders	Chromosomal Disorders
The Core Defect	Alteration/mutation in a single gene.	Absence, excess, or abnormal arrangement of whole chromosome(s).
Inheritance Predictability	Highly predictable via Mendelian laws and Punnett squares.	Random errors during cell division (segregation/cytokinesis failure).
Primary Diagnostic Tool	Pedigree Analysis.	Karyotype Analysis.
Key Examples	Haemophilia, Sickle-cell, PKU, Thalassemia.	Down's (Trisomy 21), Klinefelter's (XXY), Turner's (X0).

The Golden Rule of Genetic Disorders

Single gene = Mendelian; chromosome number/structure = chromosomal.



Memorize this framework to instantly categorize any genetic anomaly.