

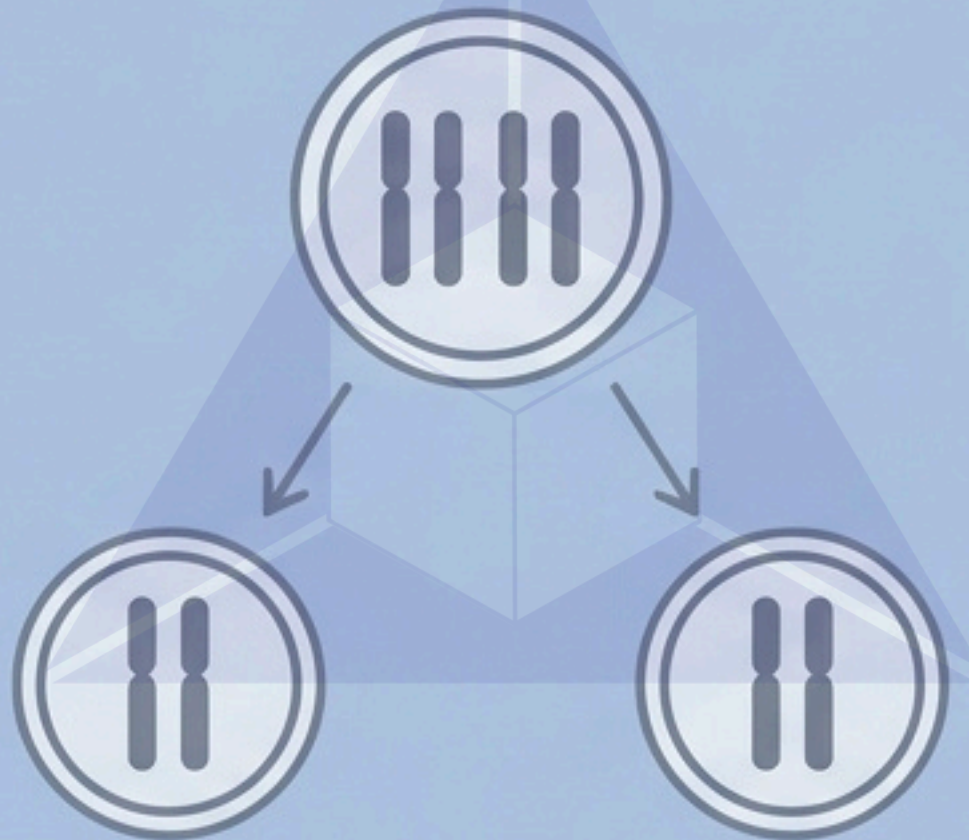
Chromosomal Disorders

The Mechanical Failures of the Human Genome

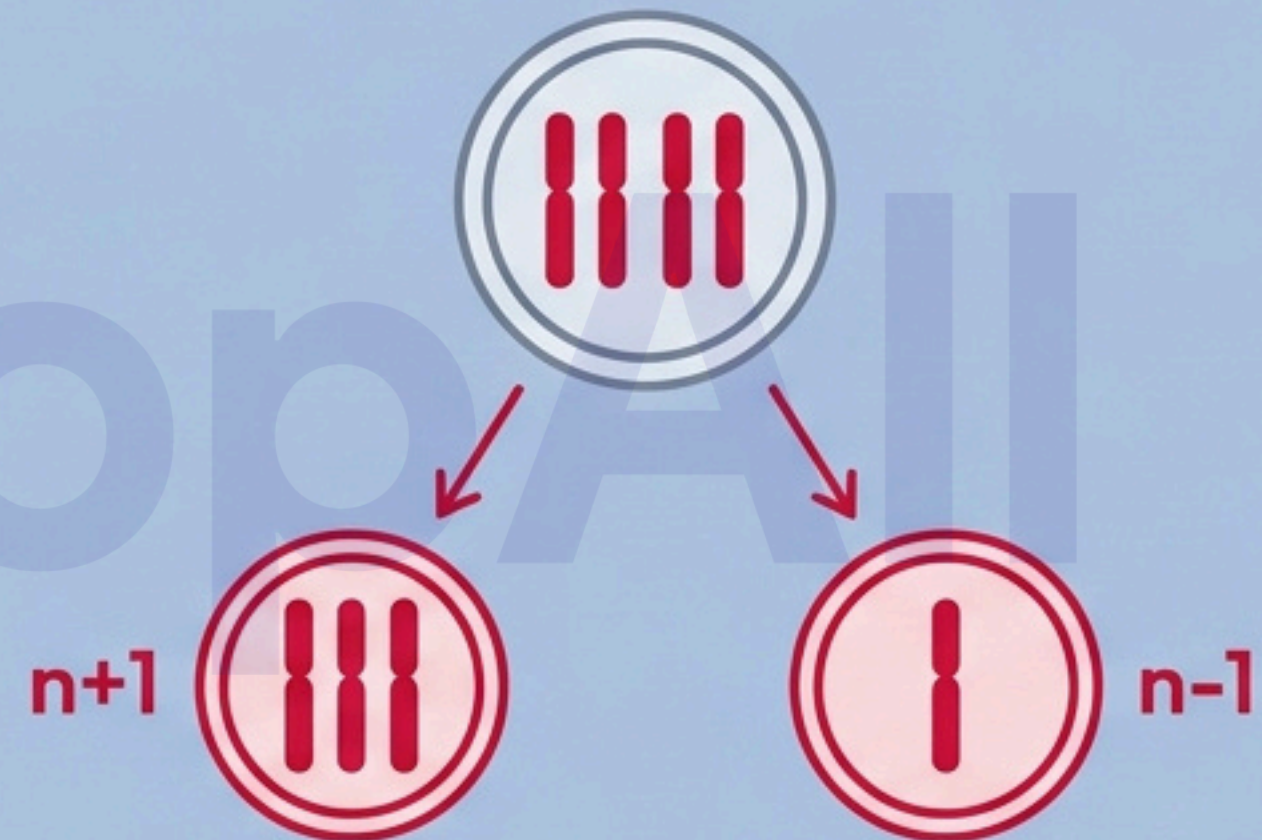
Key Takeaway: Extra or missing chromosomes cause chromosomal disorders.

The Root Cause: Division Failure

Normal Segregation



Nondisjunction: Failure of chromatids to segregate.



Chromosomal disorders occur when the physical machinery of cell division fails, resulting in an abnormal chromosome count.

The Taxonomy of Error: Aneuploidy vs. Polyploidy

Aneuploidy (Gain/Loss of Individual Chromosomes)

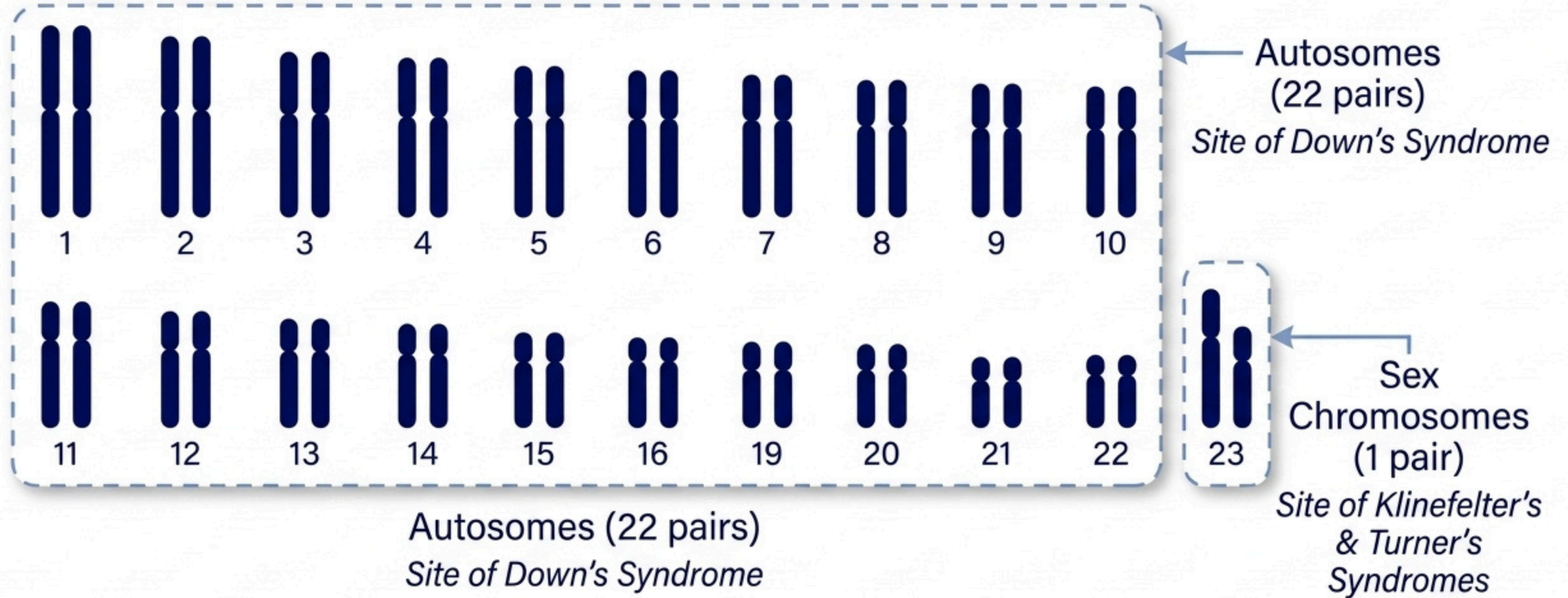
- **Mechanism:** Failure of chromatid segregation during the cell division cycle.
- **Result:** Trisomy ($2n+1$) or Monosomy ($2n-1$).
- **Occurrence:** Common cause of genetic disorders in humans.

Polyploidy (Gain of Entire Sets)

- **Mechanism:** Failure of cytokinesis after the telophase stage.
- **Result:** An increase in a whole set of chromosomes (e.g., $3n$, $4n$).
- **Occurrence:** Often seen in plants; lethal in humans.

❗ **Quick-Tip:** Do not confuse Trisomy (**one extra chromosome = 47 total**) with Triploidy (an entire extra set = **69 total**).

Two Zones of Aneuploidy



A normal human cell contains exactly 46 chromosomes. Aneuploidy can introduce an extra copy (trisomy) or lack a copy (monosomy) in either the autosomes or the sex chromosomes.



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Clinical Dossier: Down's Syndrome

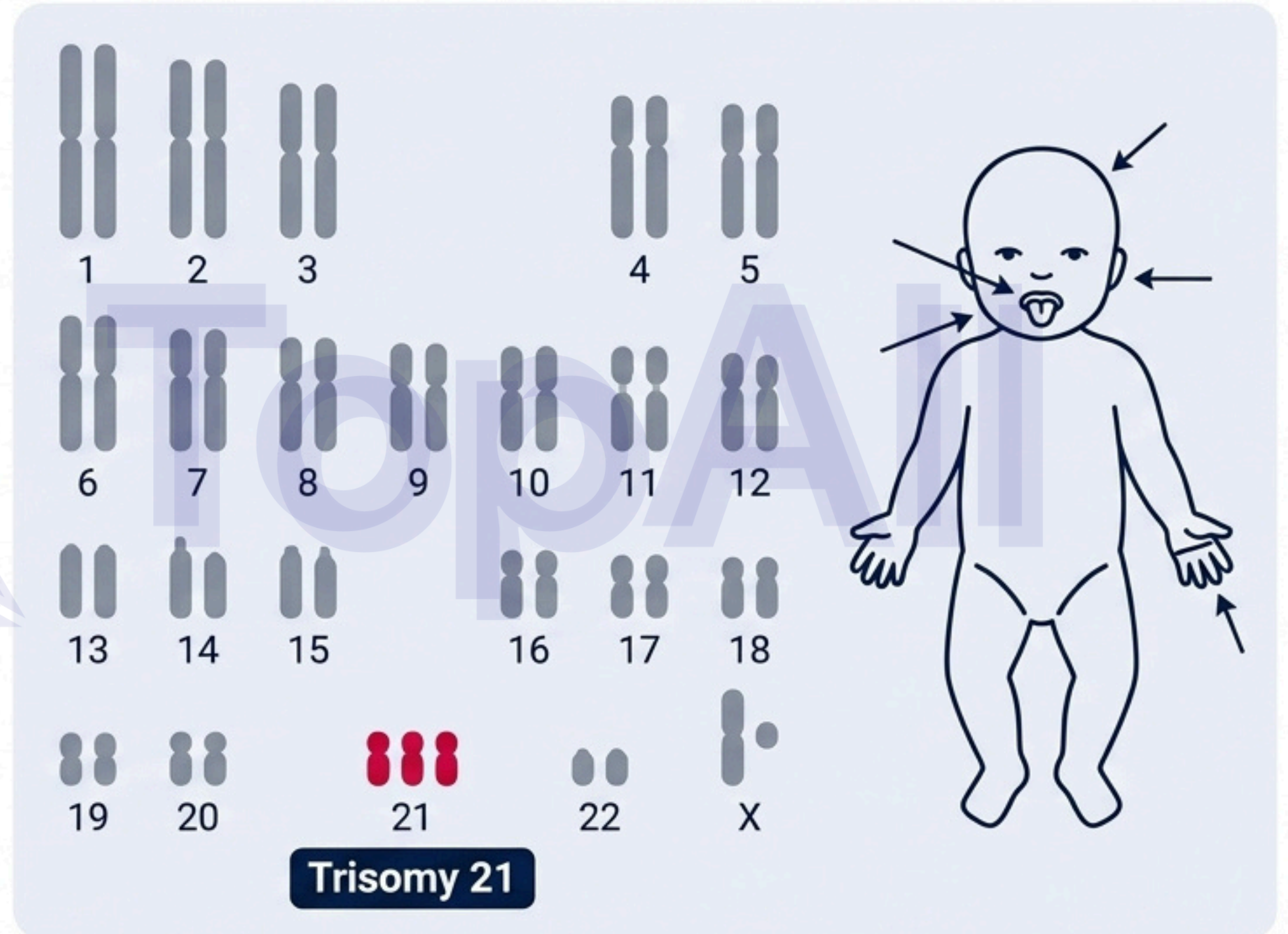
Karyotype Formula: 47, XX,+21 or 47, XY,+21 •

Total Chromosomes: 47 •

Discovery: First described by Langdon Down (1866).

Phenotypic Traits

- ▶ Short stature and small round head
- ▶ Furrowed, big, wrinkled tongue
- ▶ Partially open mouth
- ▶ Broad flat face
- ▶ Broad palm with characteristic palm crease
- ▶ Retarded physical, psychomotor, and mental development



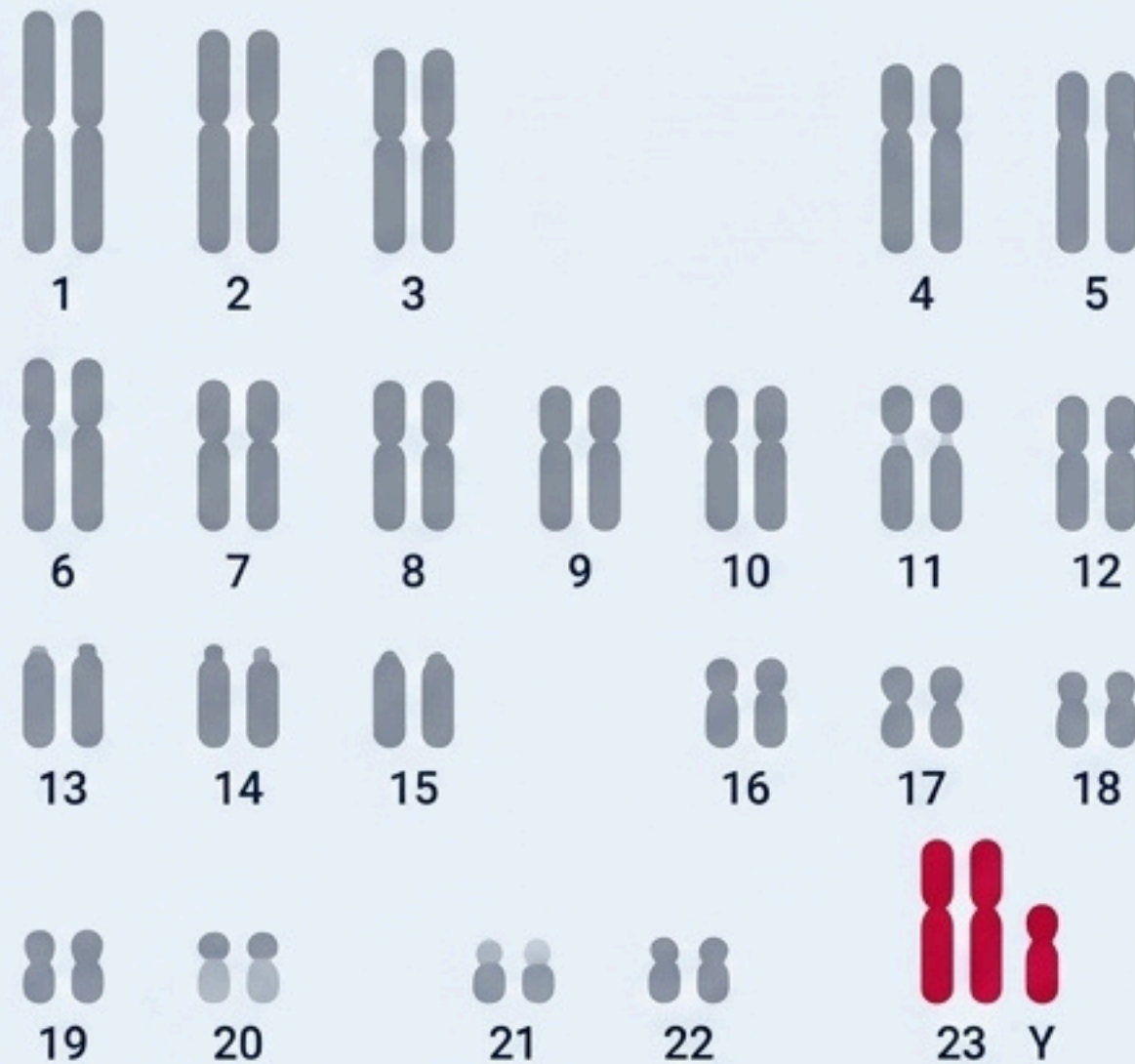
Clinical Dossier: Klinefelter's Syndrome

Clinical Specifications

- 🔬 Karyotype Formula: 47, XXY
- 🔬 Total Chromosomes: 47
- 🔬 Mutation Type: Sex-Linked Trisomy

Phenotypic Traits

- 🔬 Overall masculine development
- 🔬 Expression of feminine development (gynaecomastia / breast development)
- 🔬 Sterile individuals
- 🔬 Tall stature with feminised character



Additional X Chromosome

Clinical Dossier: Turner's Syndrome

Clinical Specifications

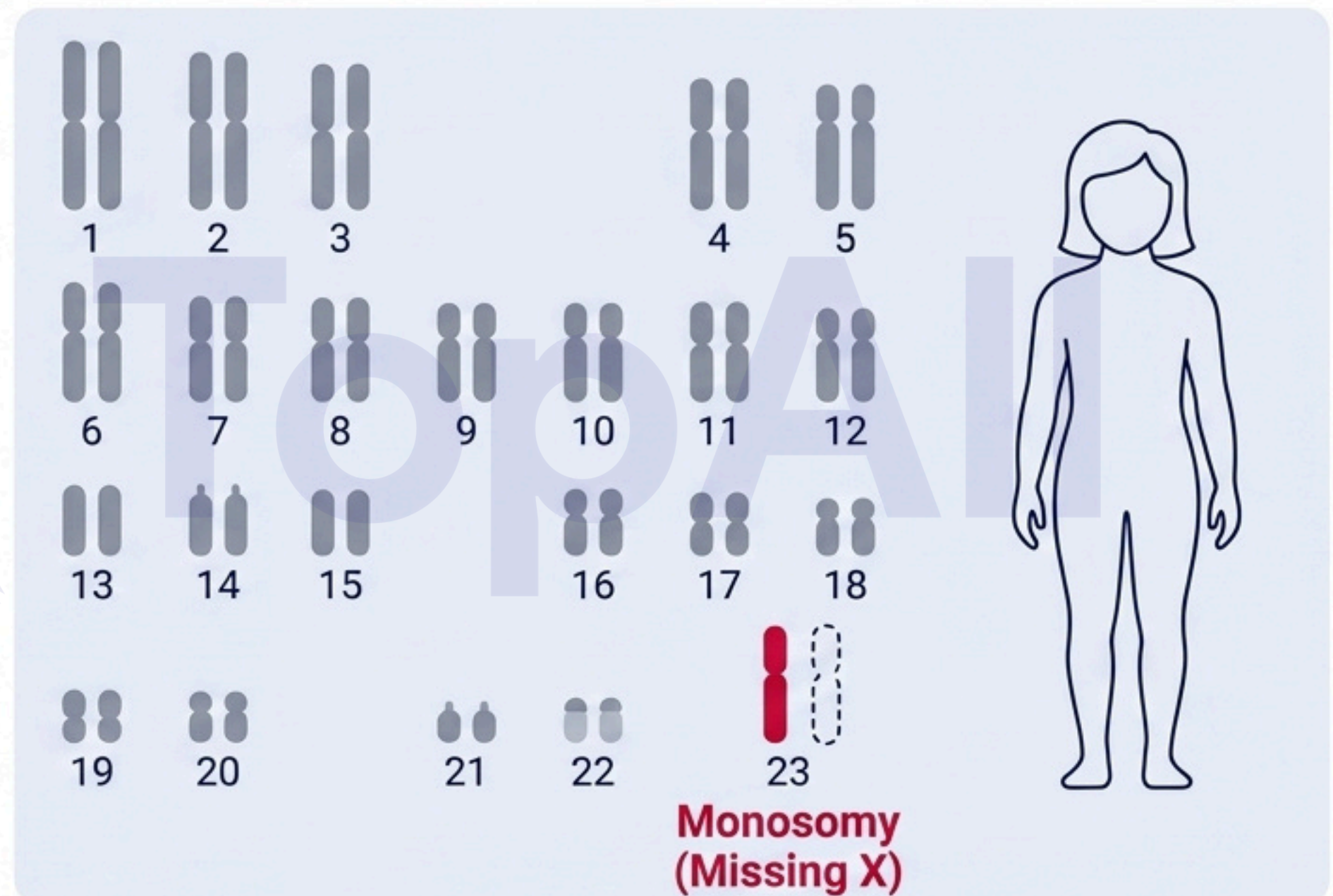
Karyotype Formula: 45, XO

Total Chromosomes: 45

Mutation Type: Sex-Linked Monosomy

Phenotypic Traits

- Sterile females
- Rudimentary ovaries
- Lack of secondary sexual characters
- Short stature



Master Diagnostic Matrix



Syndrome	Mutation Type	Formula	Total Count	Fertility	Key Identifier
Down's	Autosomal Trisomy	47 (+21)	47	(Usually Unaffected)*	Palm crease, furrowed tongue
Klinefelter's	Sex Chromosome Trisomy	47, XXY	47	Sterile (Male)	Gynaecomastia, tall stature
Turner's	Sex Chromosome Monosomy	45, XO	45	Sterile (Female)	Rudimentary ovaries, short stature

 Red text indicates the specific chromosomal deviation from the normal 46 baseline.



The Biological Rule

Whether through trisomy or monosomy, any deviation from the baseline 46 chromosomes causes profound systemic disorders.

Aneuploidy fundamentally alters human development by providing too much or too little genetic instruction.



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