CHROMOSOMAL ABERRATIONS AND TYPES OF CHROMOSOMAL DISEASES

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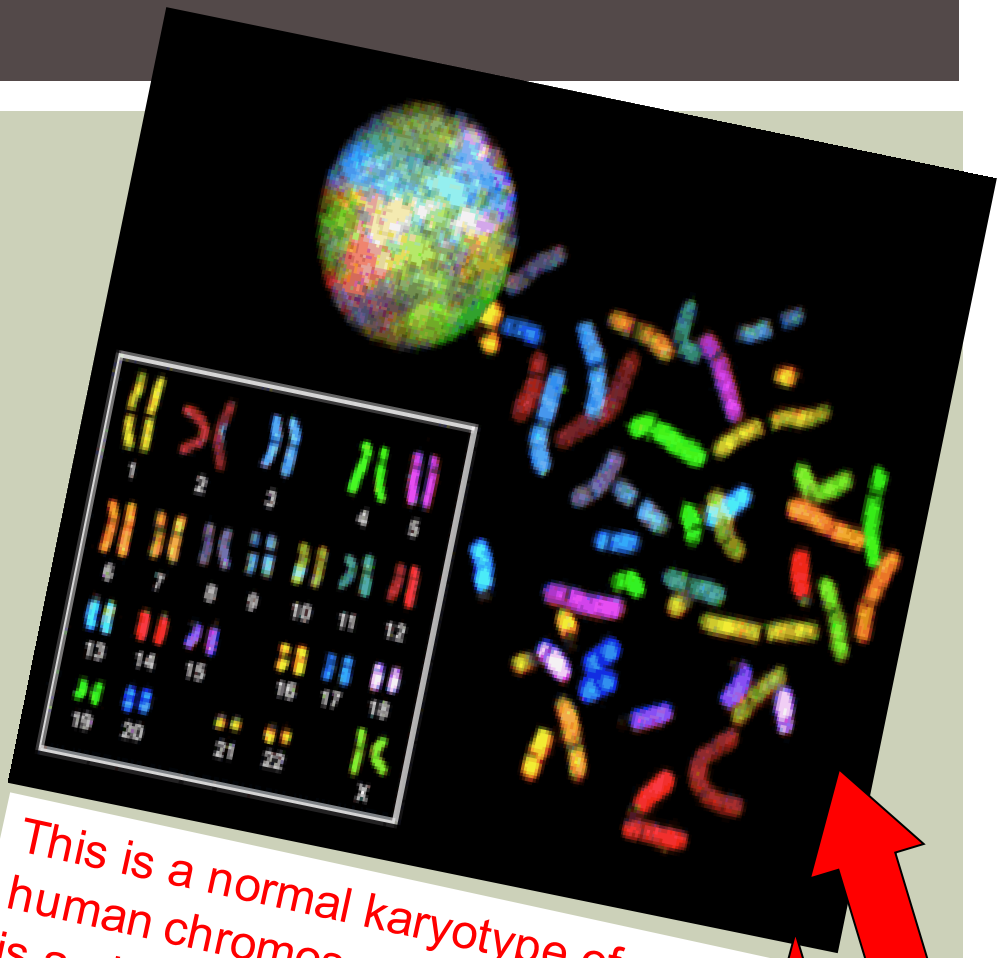
2 MAIN TYPES OF MUTATIONS

1.) Chromosomal Mutations

2.) Gene Mutations

WHAT ARE CHROMOSOMES?

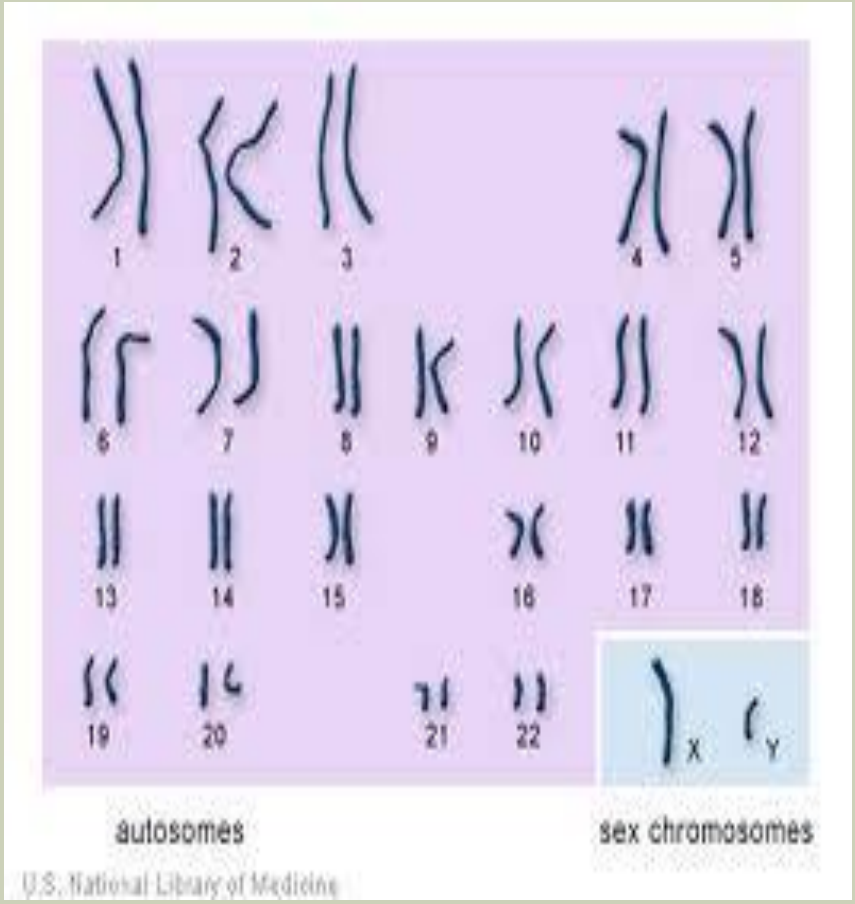
- Humans have 23 pairs of chromosomes, with one chromosome from each parent.
- The chromosomes are coiled up DNA.
- Under normal conditions all of the chromosomes are inherited in tact.



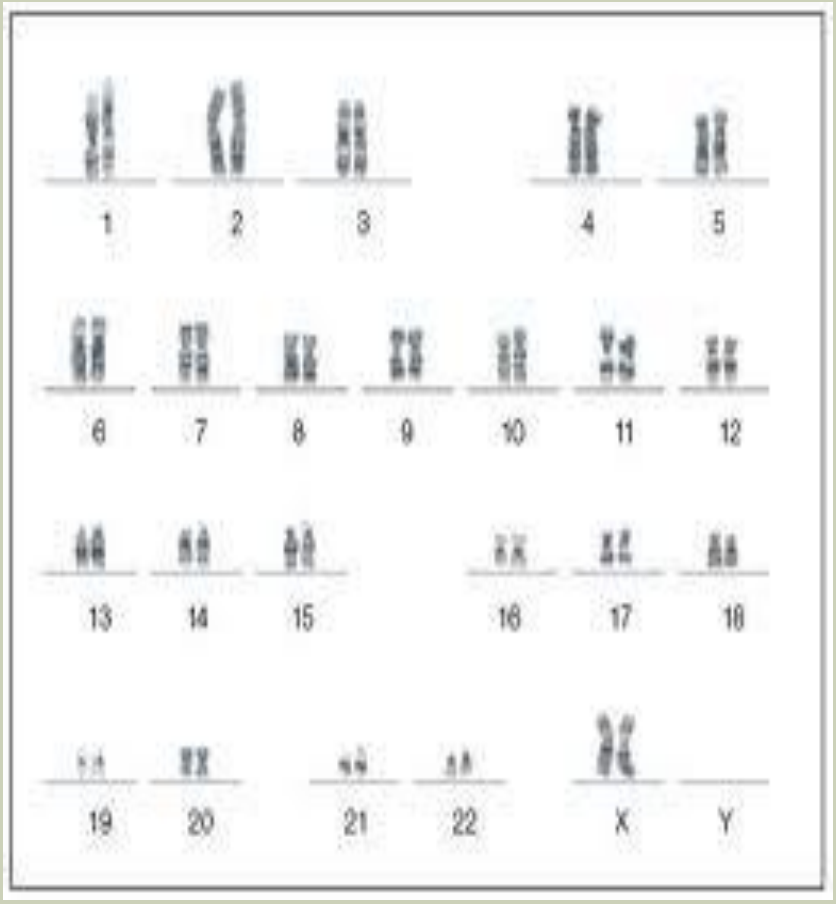
This is a normal karyotype of human chromosomes. A karyotype is a picture of chromosomes lined up to look at and compare.

KARYOTYPES (PICTURES OF CHROMOSOME PAIRS)

- Normal male



- Normal female



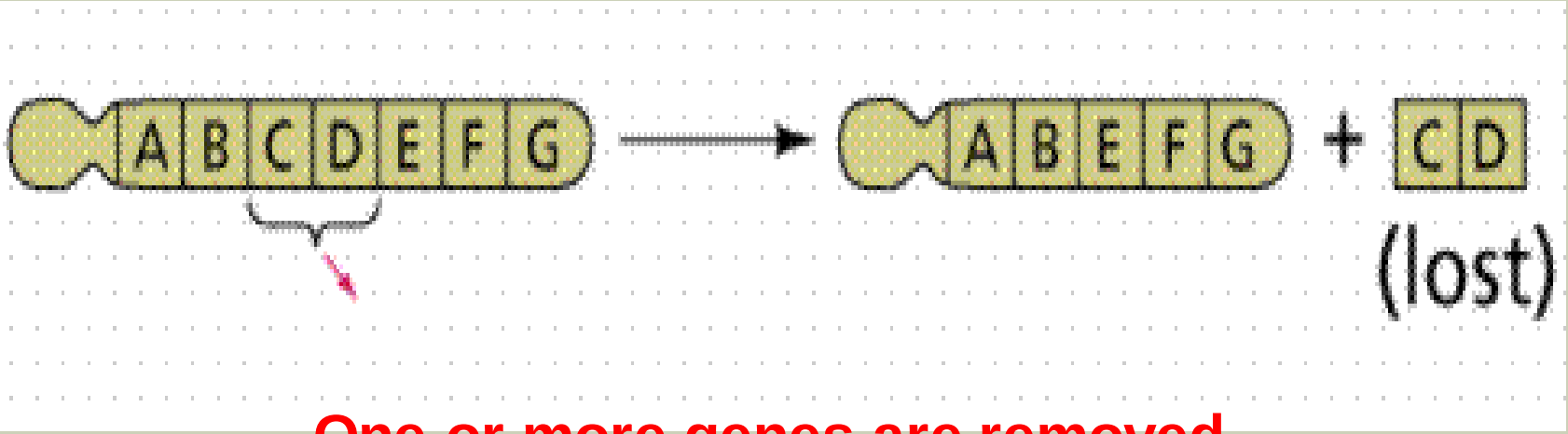
CHROMOSOMAL MUTATIONS

- Any change in the **structure** or **number** of chromosomes
- Large scale: Affect *many* genes

5 TYPES

1. Deletion
2. Duplication
3. Inversion
4. Translocation
5. NonDisjunction

CHROMOSOMAL DELETION



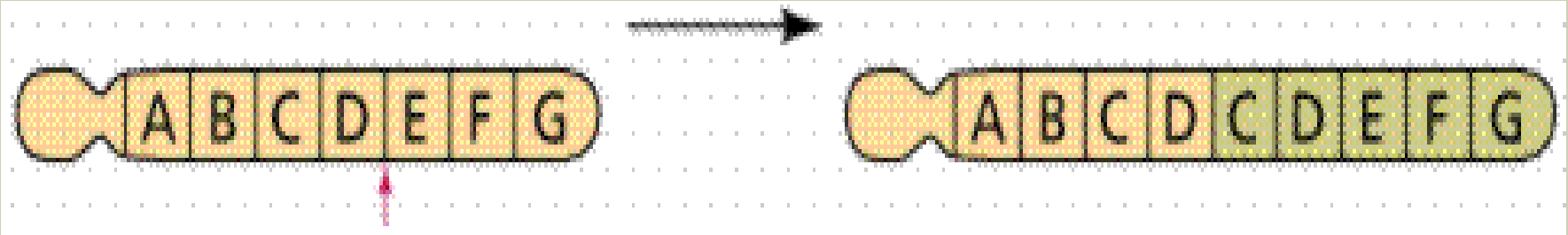
One or more genes are removed

Causes:

Wolf-Hirschhorn syndrome (severe mental retardation)

cri du chat syndrome (mewing sounds, mental retardation)

CHROMOSOMAL DUPLICATION



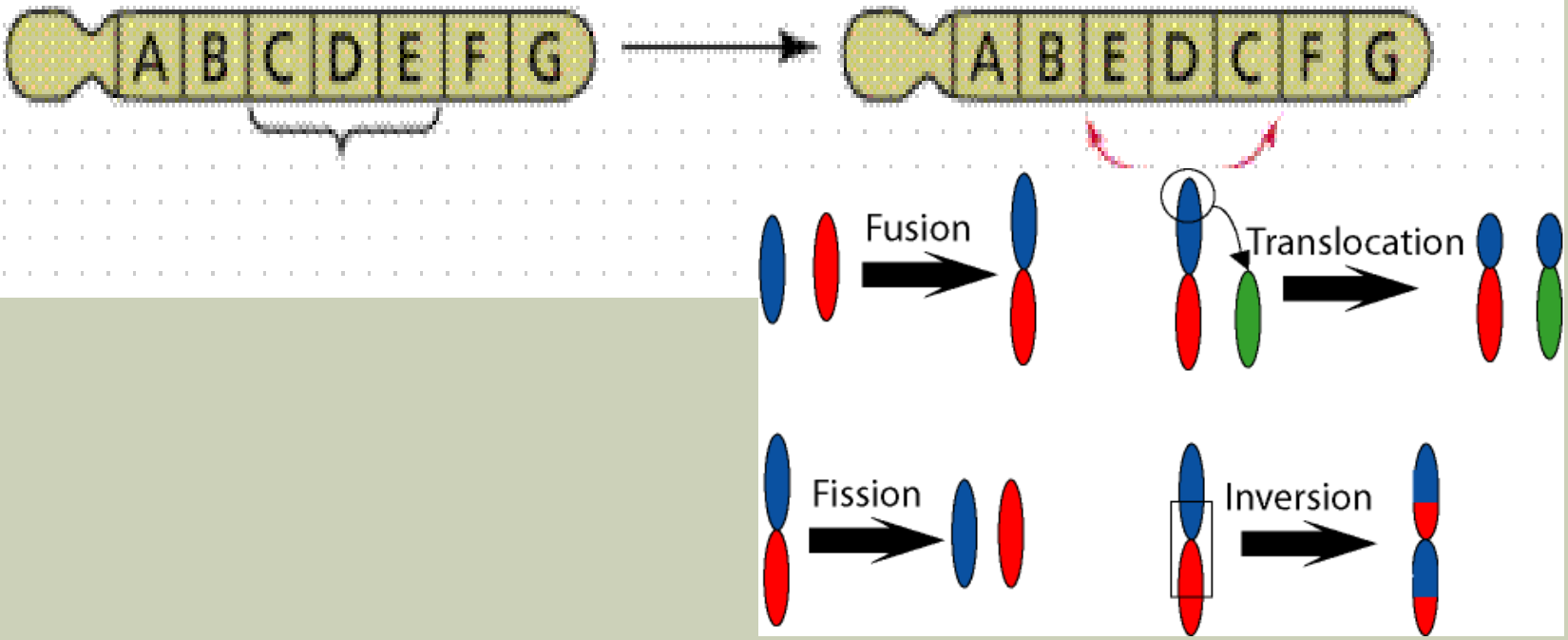
A segment of genes is copied twice and added to the chromosome

Causes:

Charcot–Marie–Tooth disease

(high arched foot, claw feet, confined to a wheelchair)

CHROMOSOMAL INVERSION



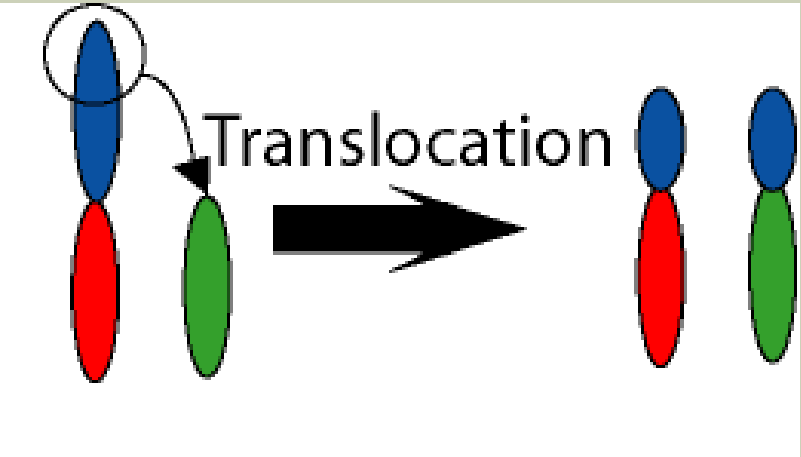
a segment of genes flip end-to-end on the chromosome

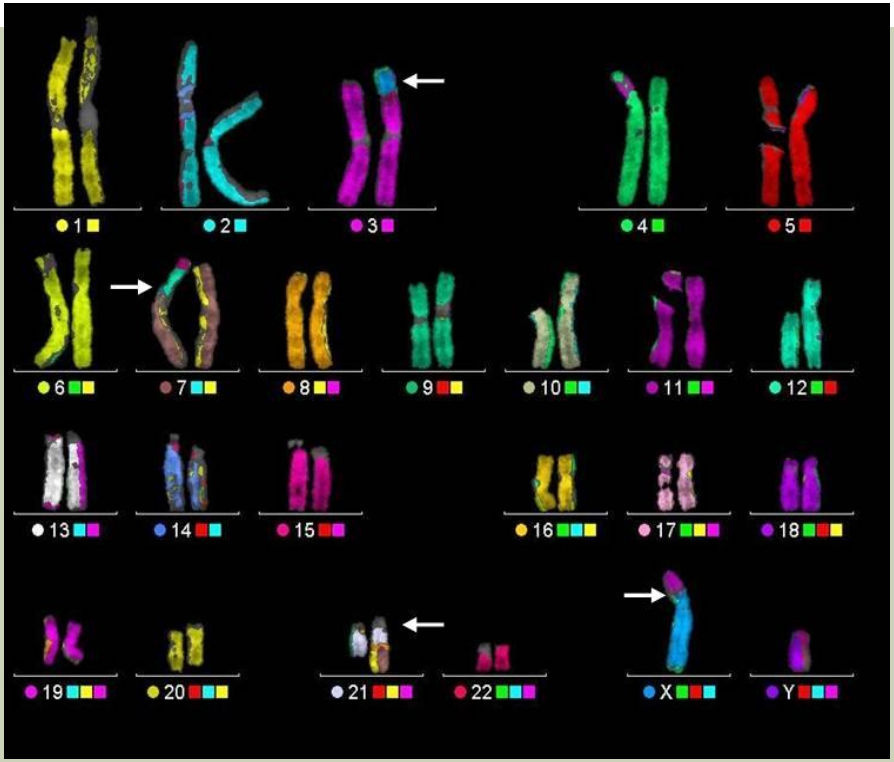
Causes:

Four-Ring Syndrome

(cleft pallate, club feet, testes don't descend)

CHROMOSOMAL TRANSLOCATION





Material is swapped with another chromosome

Causes:

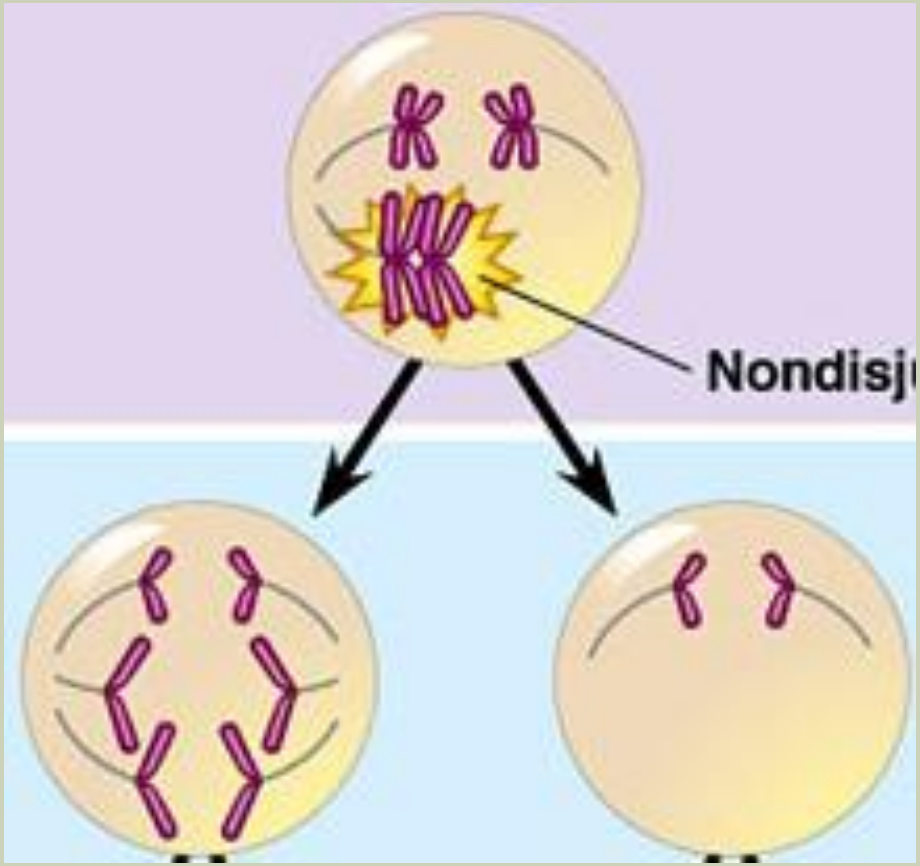
Burkitt's Lymphoma

(cancer of the lymph nodes, in children)

NONDISJUNCTION

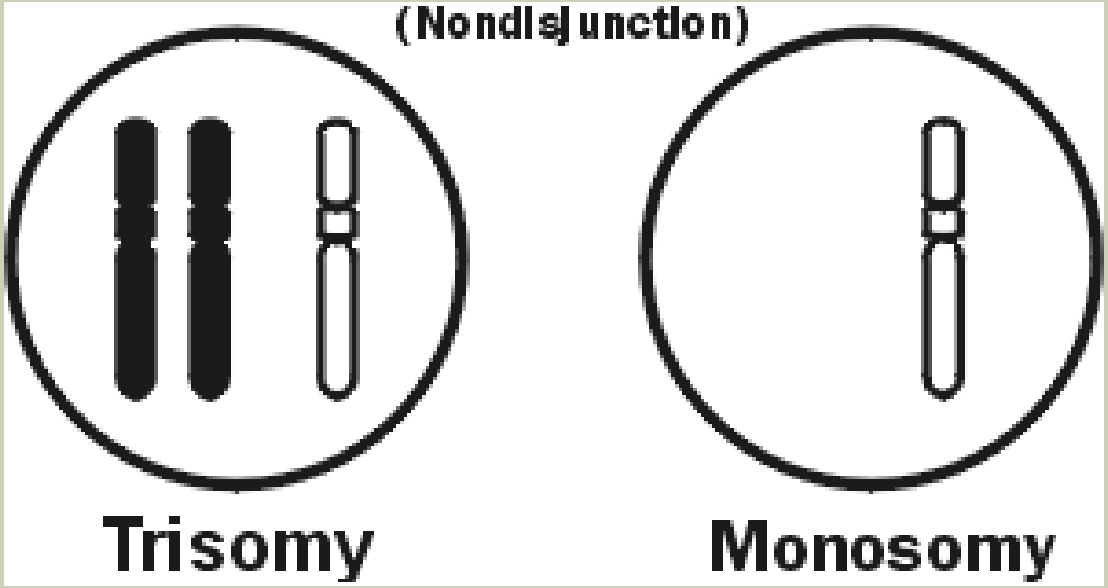
- Chromosomes **FAIL TO SEPARATE** during **meiosis**

  - Meiosis I Nondisjunction
  - Meiosis II Nondisjunction

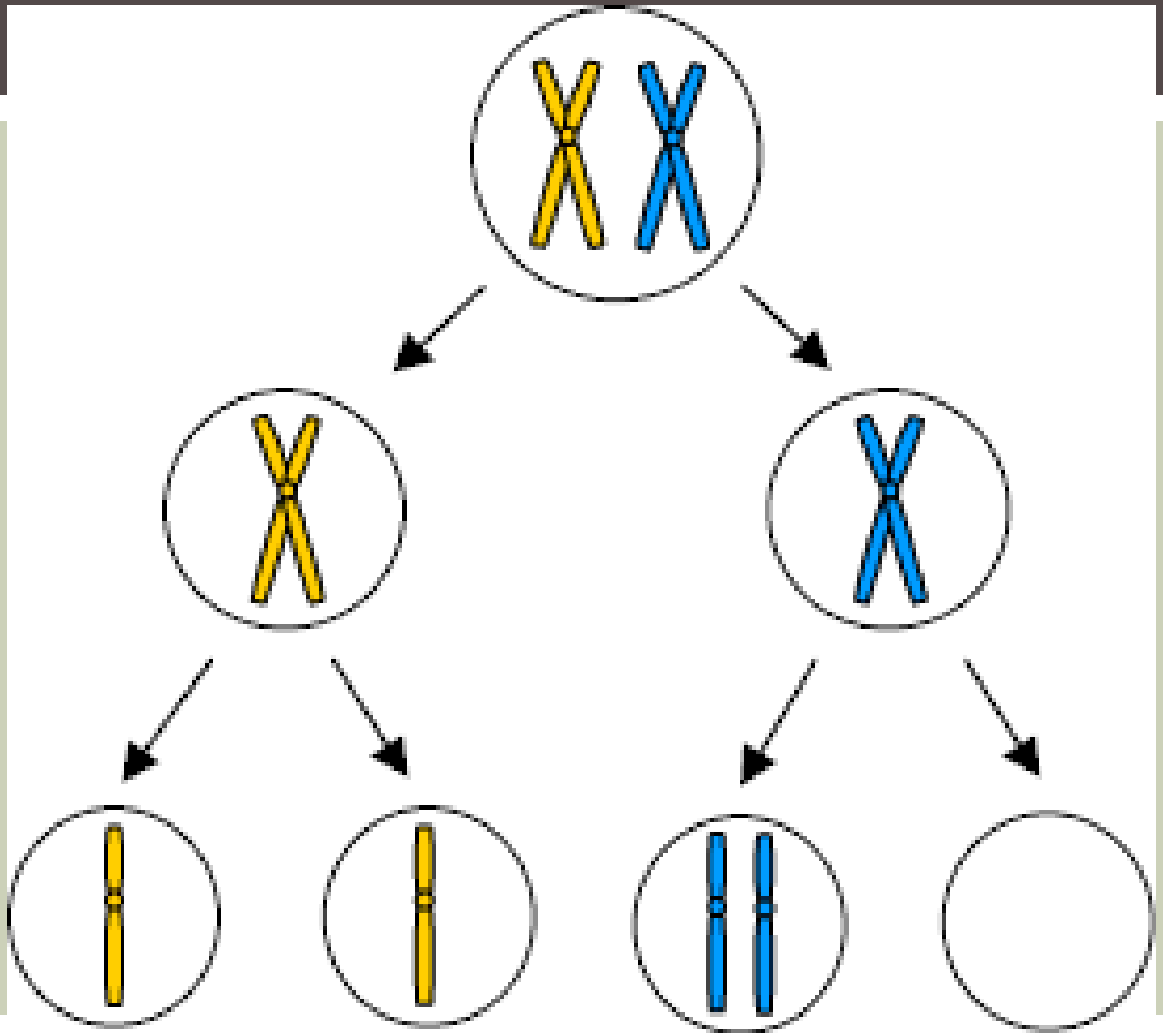


NONDISJUNCTION

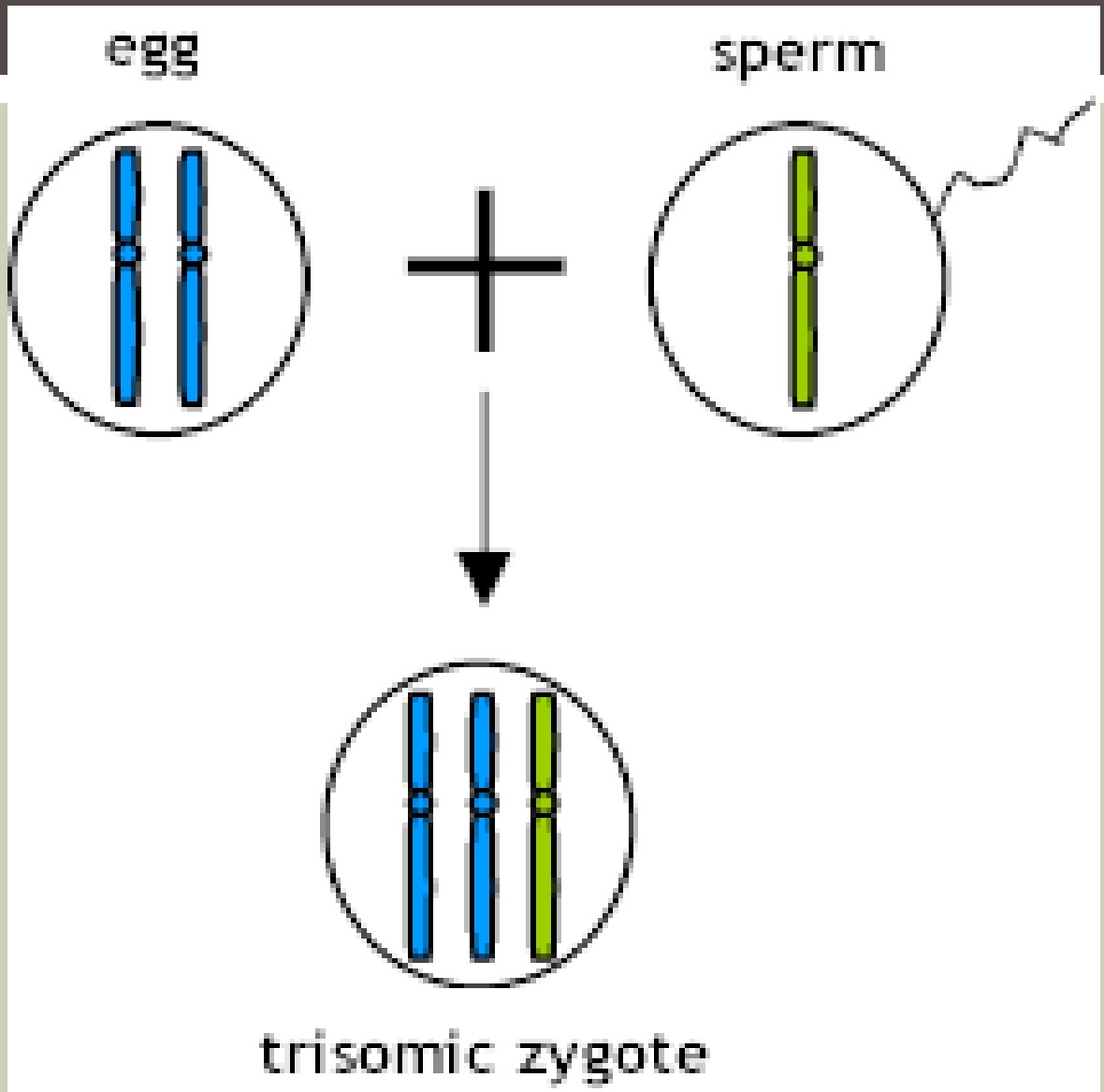
- Produces gametes (and therefore a baby) with one missing chromosome **or** one extra chromosome



NONDISJUNCTION (IN MEIOSIS II)

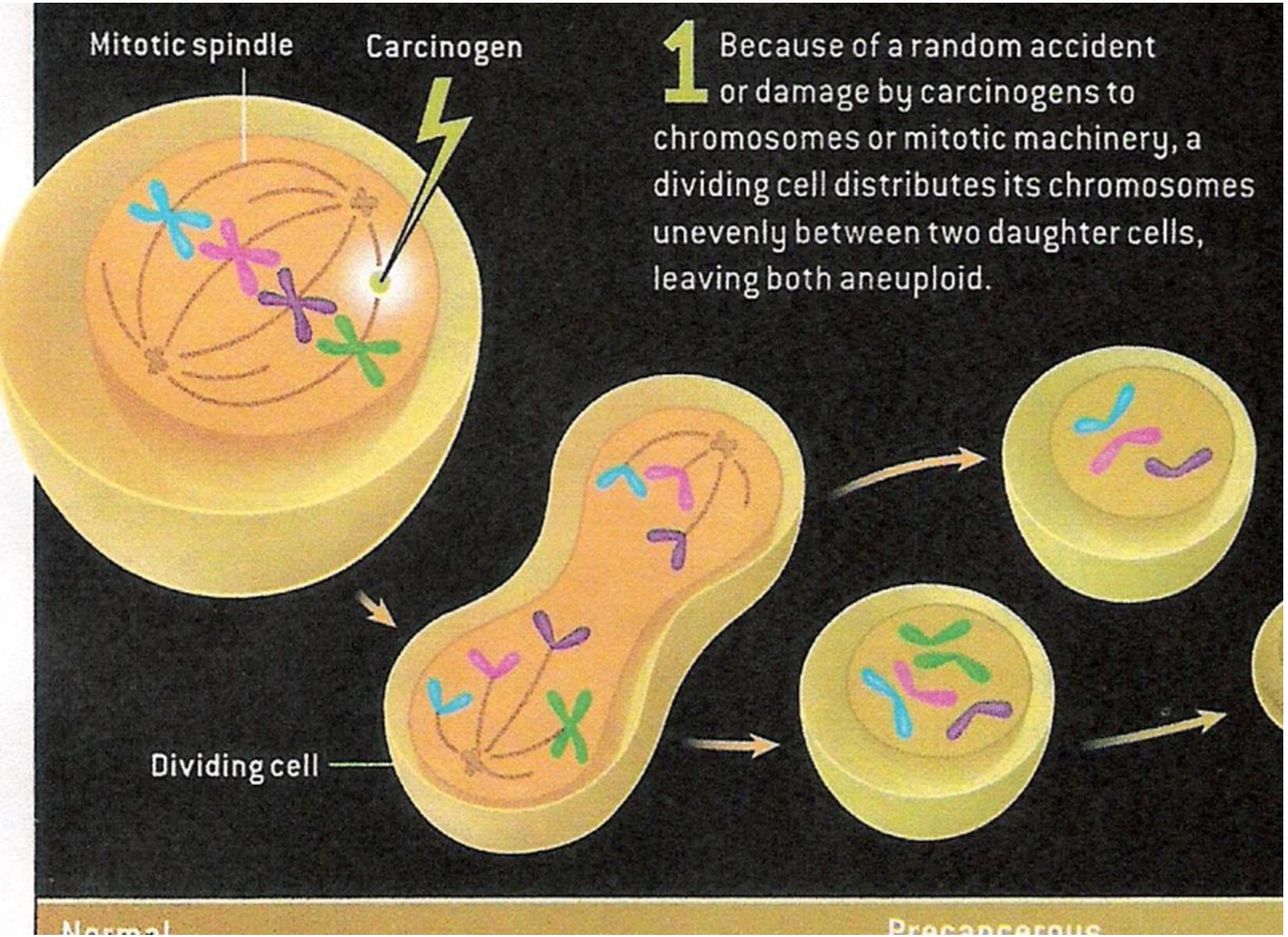


FERTILIZATION



NONDISJUNCTION

- Every cell in that baby's body will have 3 copies of this chromosome instead of 2.
- This condition is called **TRI**SOMY
- **Trisomy 21** = Individual has **3** copies of chromosome # **21**.


Mitotic spindle
Carcinogen
1 Because of a random accident or damage by carcinogens to chromosomes or mitotic machinery, a dividing cell distributes its chromosomes unevenly between two daughter cells, leaving both aneuploid.
Dividing cell

CHROMOSOMAL MUTATIONS

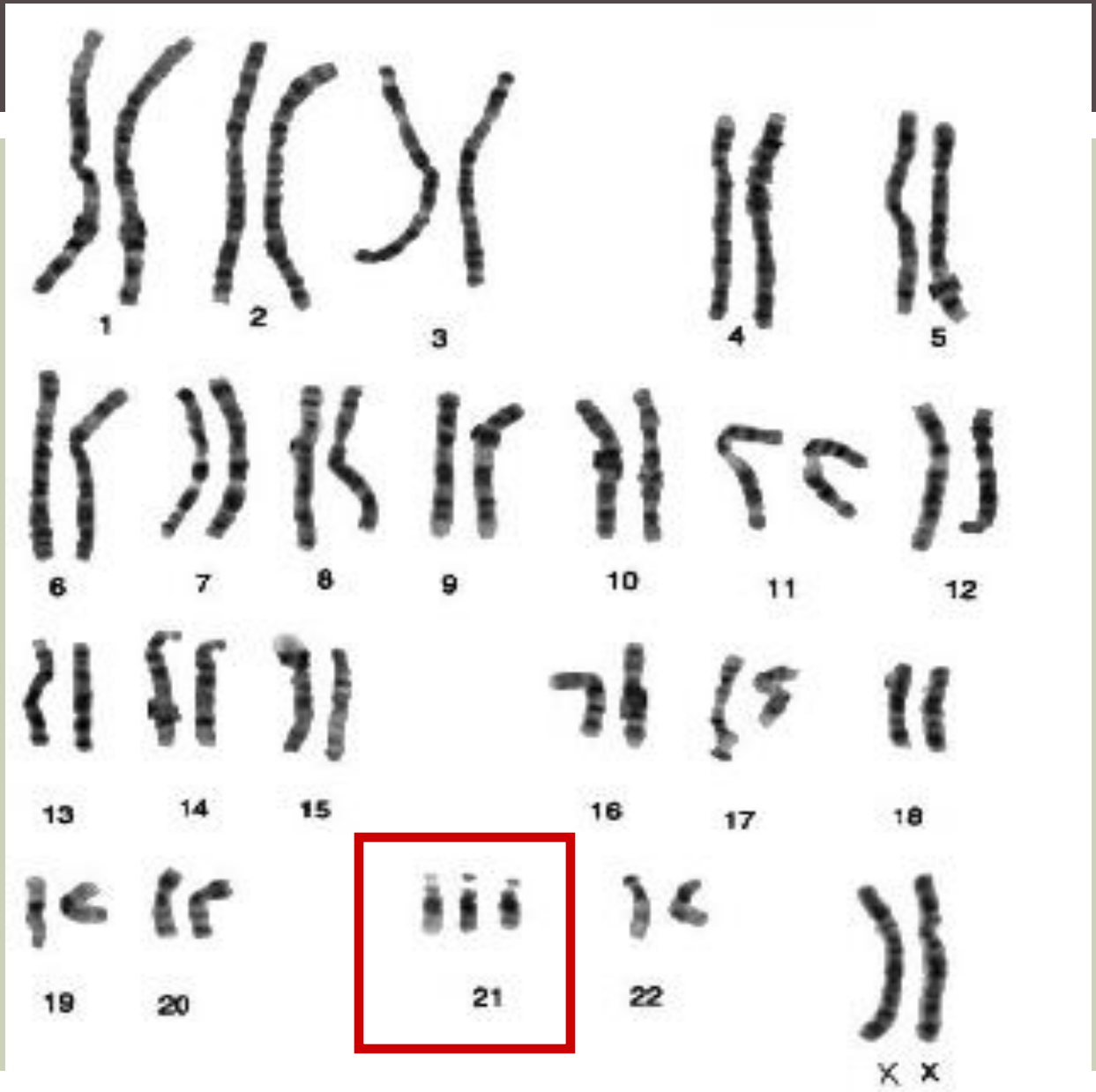
- Most chromosomal mutations are *lethal*
- *If* the fetus survives: Tend to cause wide-spread abnormalities
- *Example:* Down Syndrome

DOWN SYNDROME

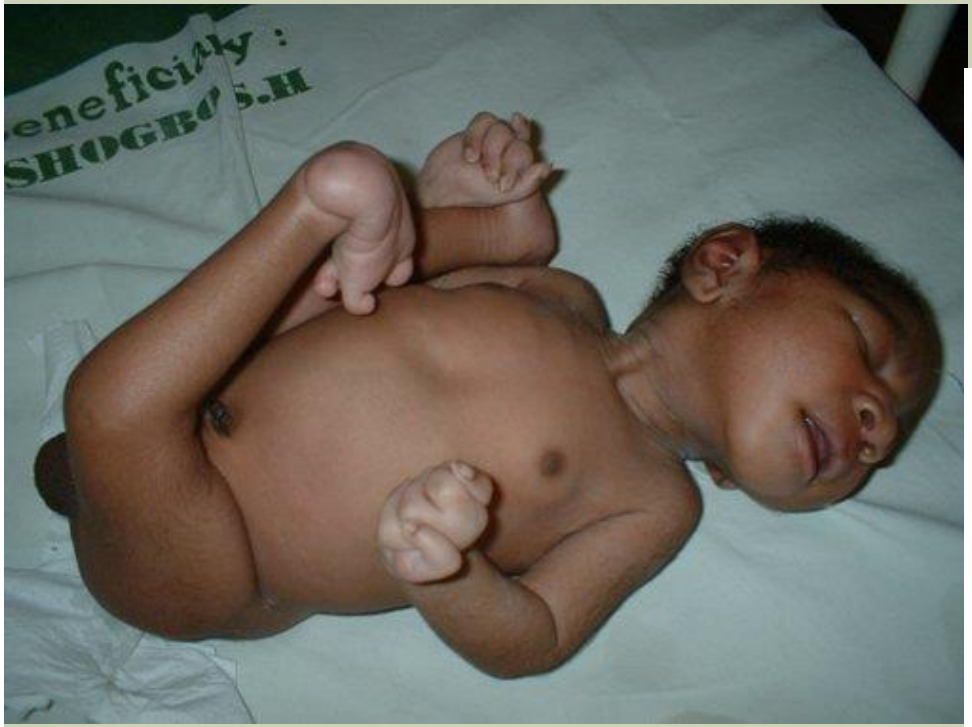
- **Cause:**
  Nondisjunction of chromosome 2_1

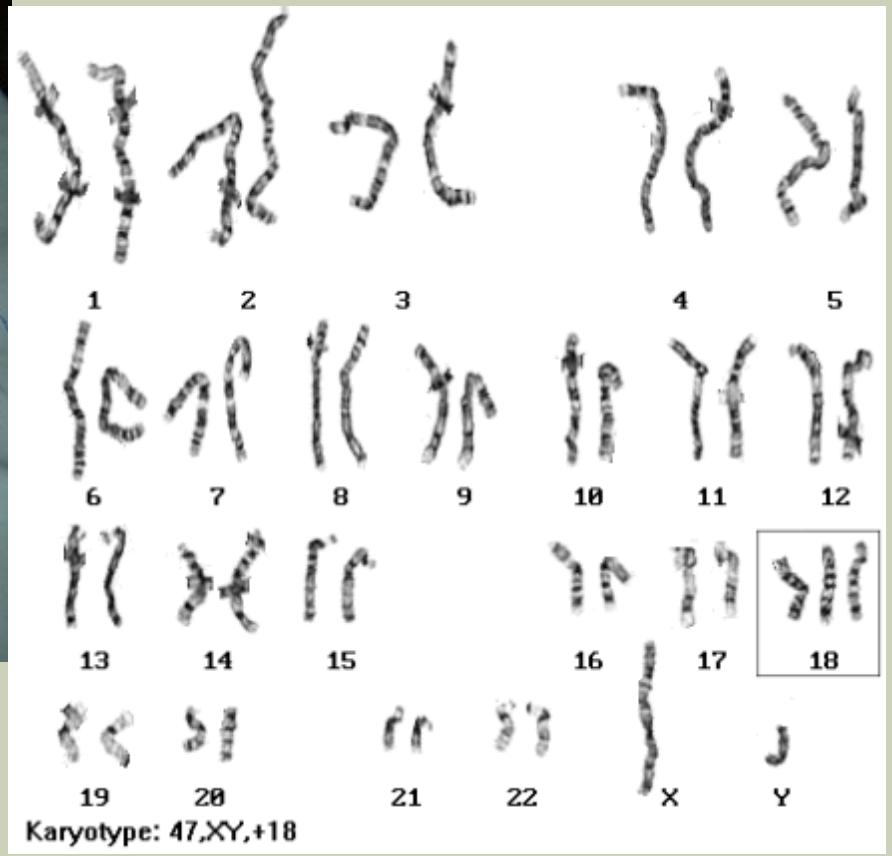
- **Three** copies of chromosome **21** = "**TRISOMY 21**"

TRISOMY 21 - DOWN SYNDROME



EXAMPLES: PATAU'S SYNDROME





GENETIC SCREENING - AMNIOCENTESIS