

The background features a dark green field filled with various sizes of semi-transparent green gears. On the left side, there is a vertical strip with a colorful, abstract, and pixelated pattern in shades of red, orange, yellow, and blue. The text is centered in a large, white, sans-serif font.

Mutations & Genetic Disorders

Mutations

Mutation:

- Any **mistake** or **change** in the DNA sequence

Point mutation:

- Change in **one** nitrogen base in DNA
- Ex: albinism

point mutation

WILD-TYPE
DNA

ATGCATGCATGC
TACGTACGTACG

| change in one
| base

MUTANT
DNA

ATGCTTGCATGC
TACGAACGTACG

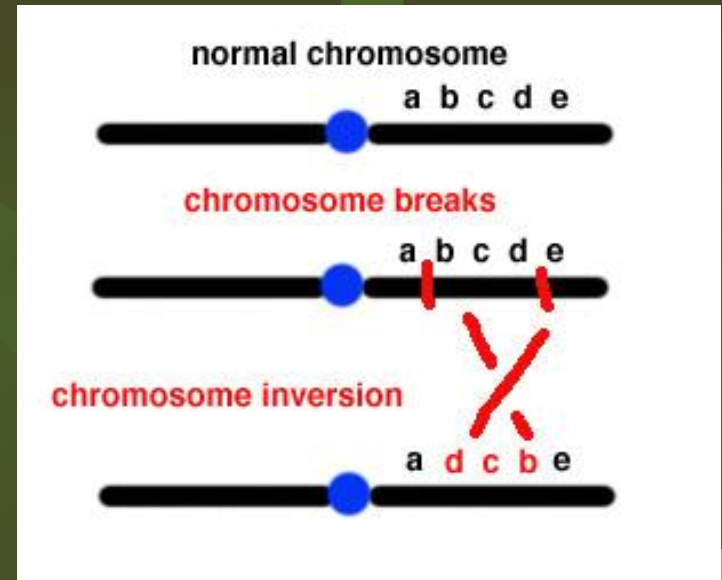


Chromosomal Mutation:

- Changes in chromosome structure

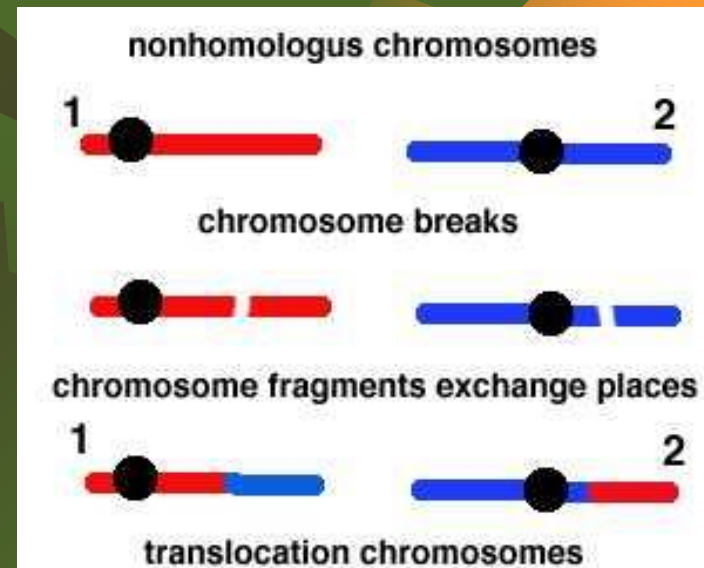
1) INVERSION:

- the order of genes on a chromosome is **inverted**



2) TRANSLOCATION:

- the movement of a chromosome fragment to a **nonhomologous** chromosome



3. DELETION

- ✦ **Loss** of a few bases
- ✦ Loss of large regions of a chromosome

deletion mutation

WILD-TYPE DNA	ATGCATGCATGC TACGTACGTACG
	ATG TAC <i>deleted</i>
MUTANT DNA	ATGCCATGC TACGGTACG

4. DUPLICATION

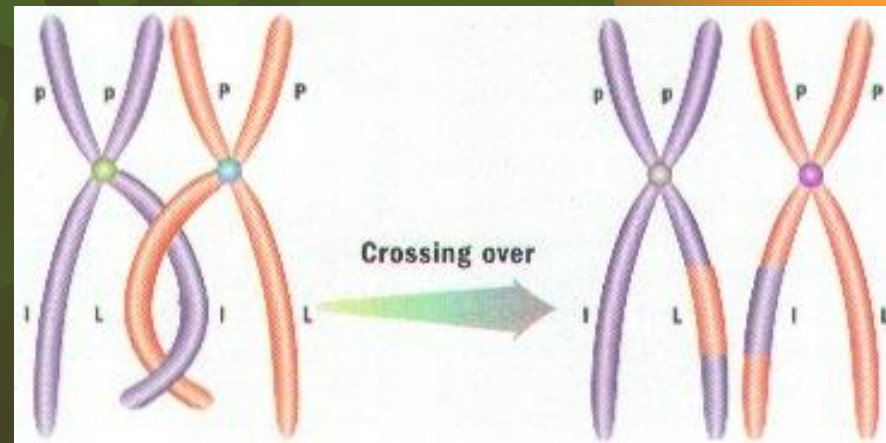
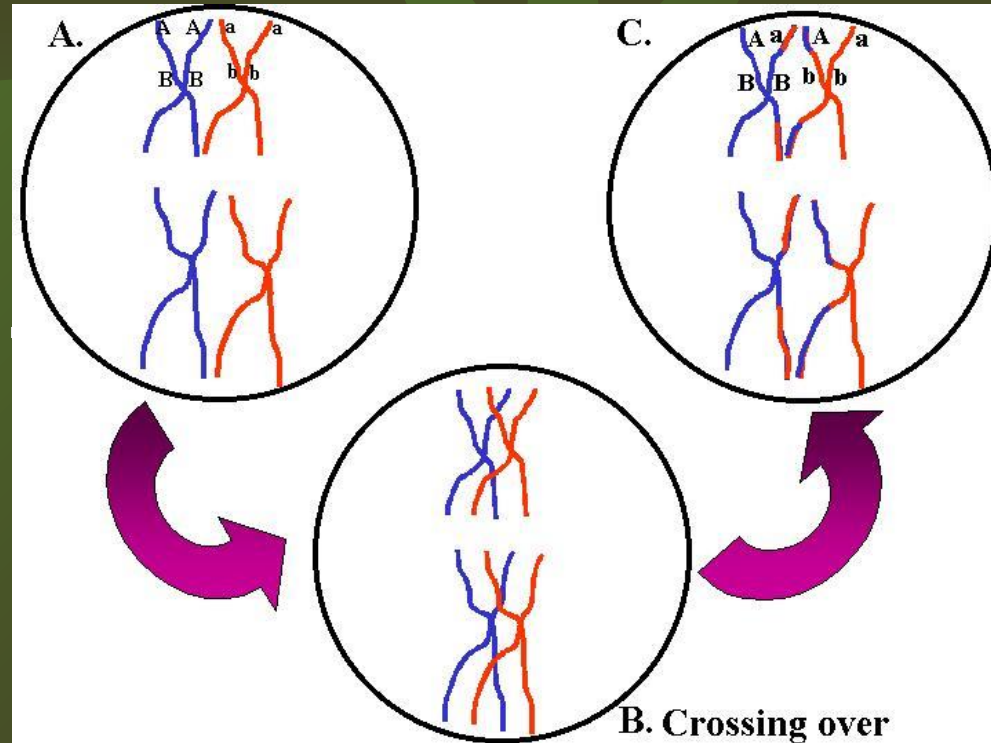
- ✦ Duplication of a few bases
- ✦ Duplication of large regions of a chromosome

DUPLICATION MUTATION

WILD-TYPE DNA	ATGCATGCATGC TACGTACGTACG
	AT TA <i>duplication of AT</i>
MUTANT DNA	ATGCATATGCTACG TACGTATACGATGC

Crossing Over

- Occurs when chromosomes **exchange genes**
- 2 chromosomes overlap.
- Some genes cross over and **switch** places



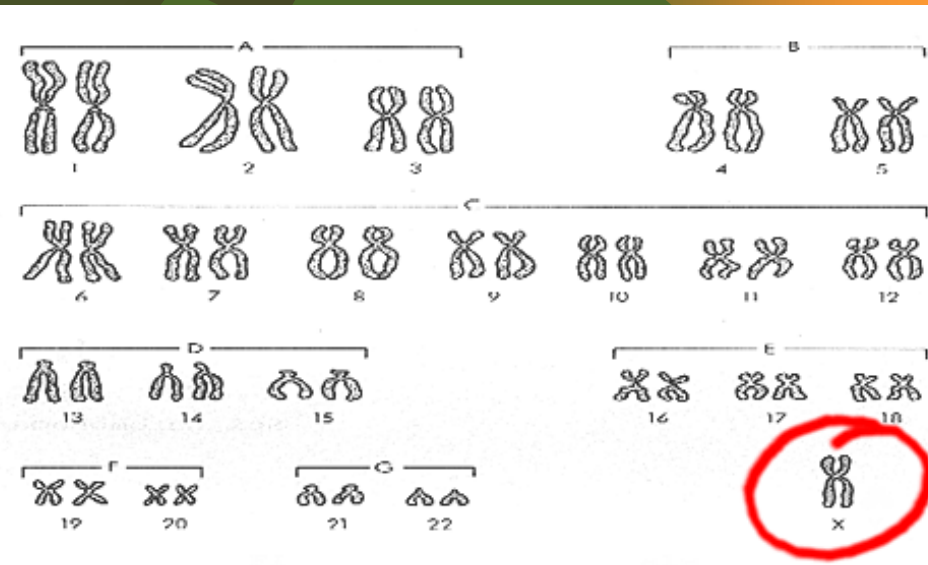
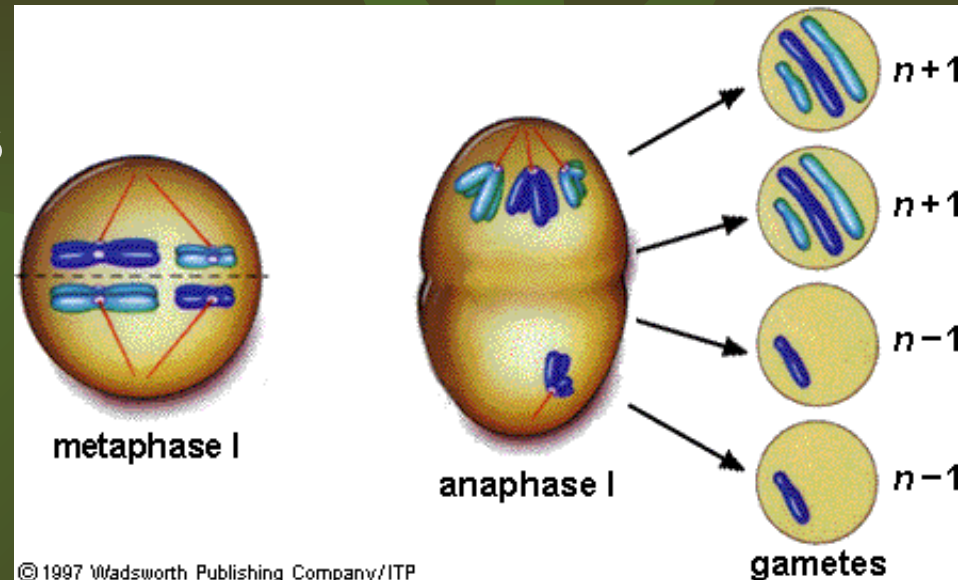
NONDISJUNCTION

Nondisjunction:

- ☀ chromosome pair fails to separate properly during meiosis

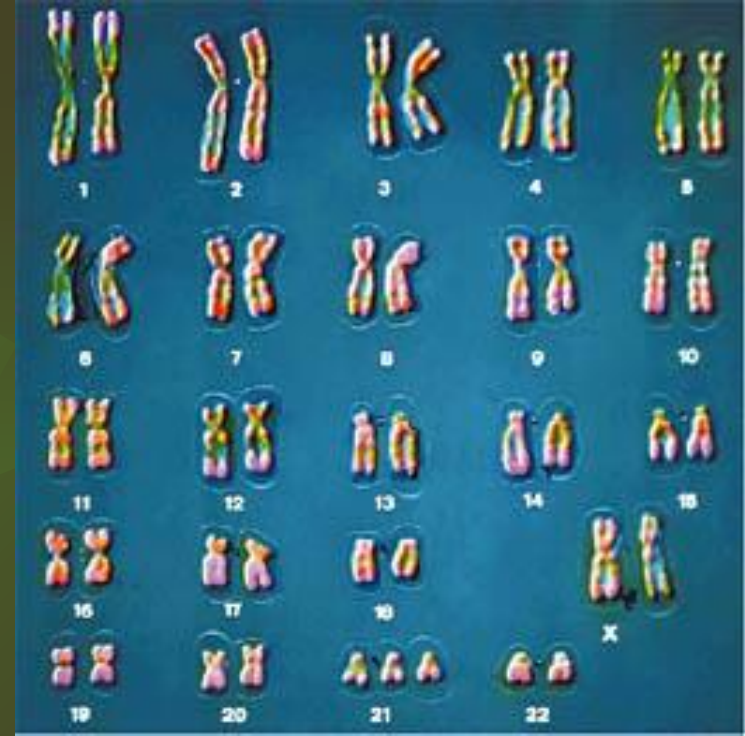
Monosomy:

- ☀ gamete has **1 less** chromosome than it should
- ☀ **45 chromosomes** is the result
- ☀ Ex: **Turner syndrome**
 - Missing a sex chromosome



Trisomy:

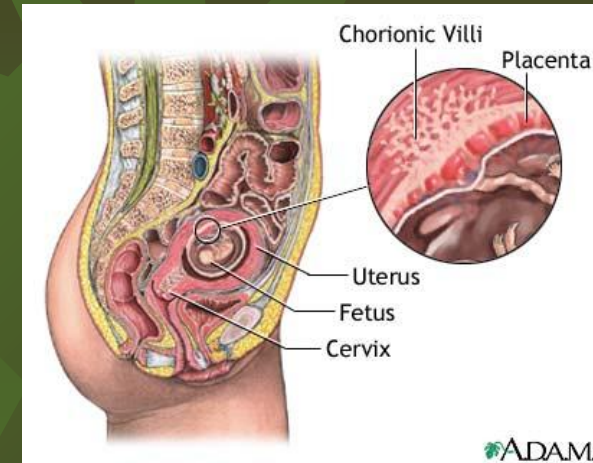
- ★ Gamete has **1 more** chromosome than it should
- ★ Result is **47** chromosomes
- ★ Ex: Down's Syndrome
 - ★ Extra #21 chromosome



Methods of Detection

Chorion villi sampling:

- Take sample of the chorion
 - (membrane surrounding fetus)
- Chemical tests and **Karyotyping** performed



Ultrasound:

- **Sound waves** are used to generate an image of the unborn child.
- Used to detect abnormalities of limbs, organs, etc.



Amniocentesis:

- **Fluid** surrounding the fetus is drawn out by needle
- Fetal cells are collected and grown in a lab.
- Chromosomes can be then Karyotyped

