MUTATIONS & GENETIC DISORDERS

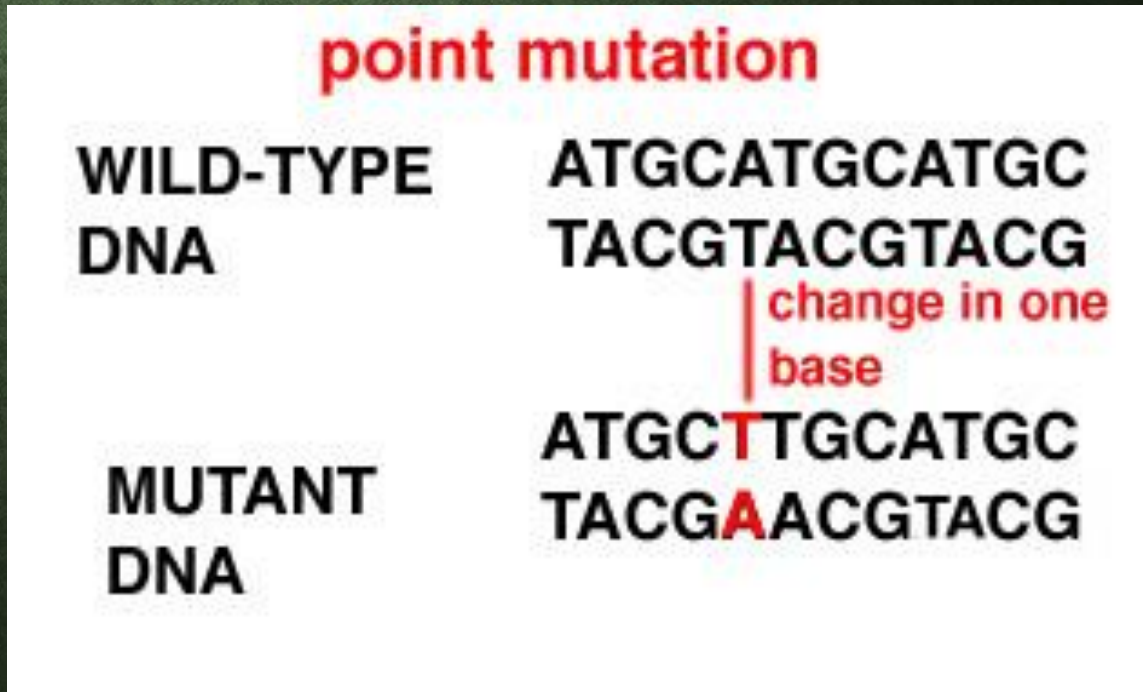
MUTATIONS

Mutation:

- Any mistake or change in the DNA sequence

Point mutation:

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



Chromosomal Mutation:

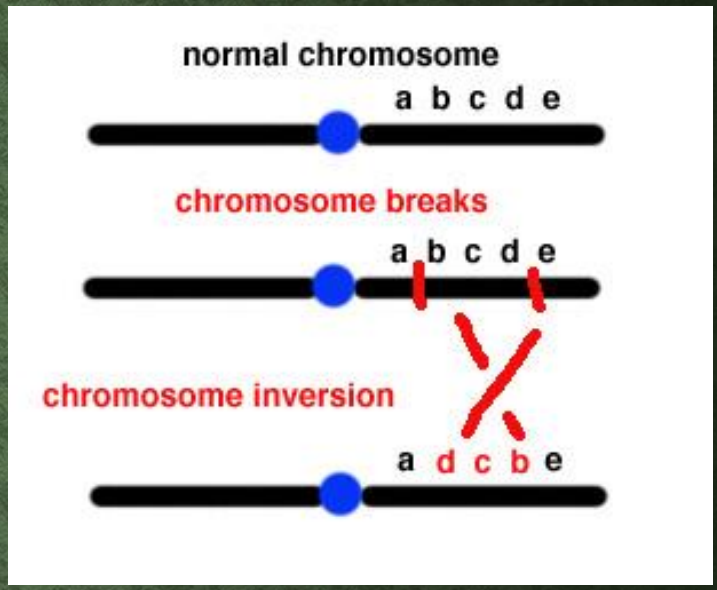
- Changes in chromosome structure. There are four types.

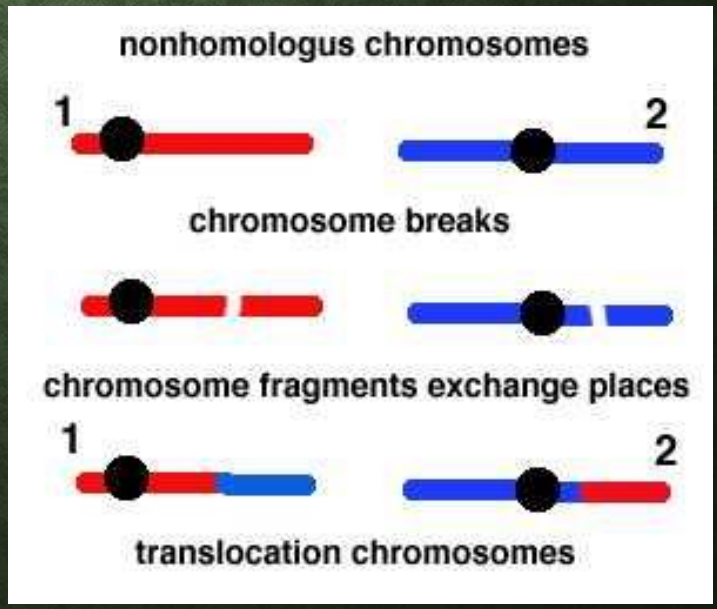
1) INVERSION:

- the order of genes on a chromosome is inverted

2) *TRANSLOCATION:*

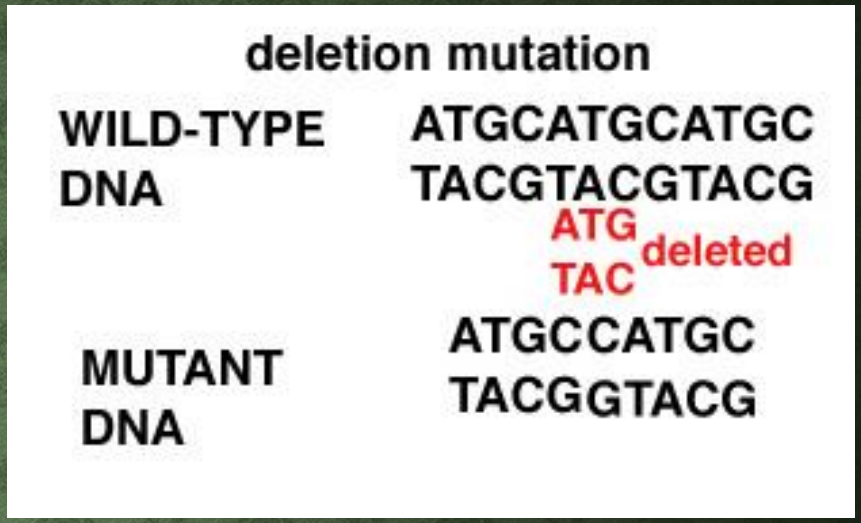
- the movement of a chromosome fragment to a nonhomologus chromosome





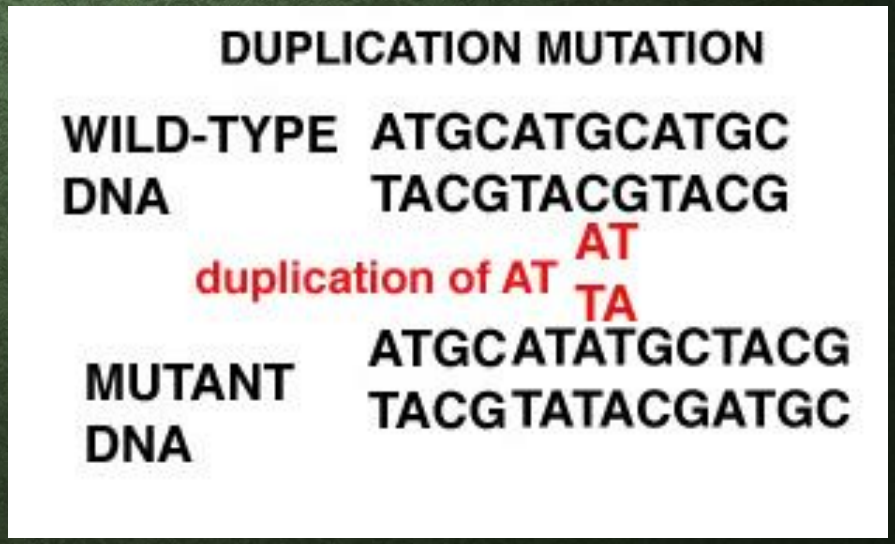
3. DELETION

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



4. DUPLICATION

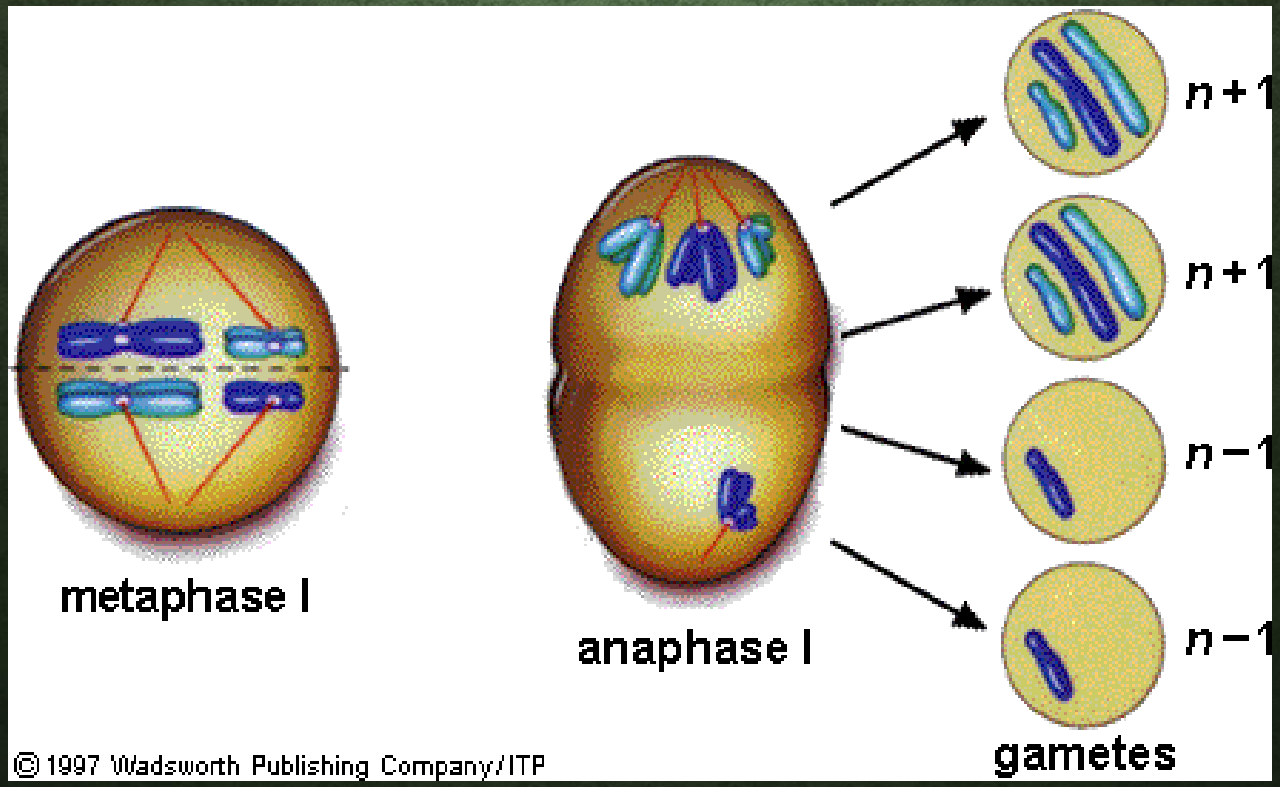
- Duplication of a few bases
- Duplication of large regions of a chromosome



NONDISJUNCTION

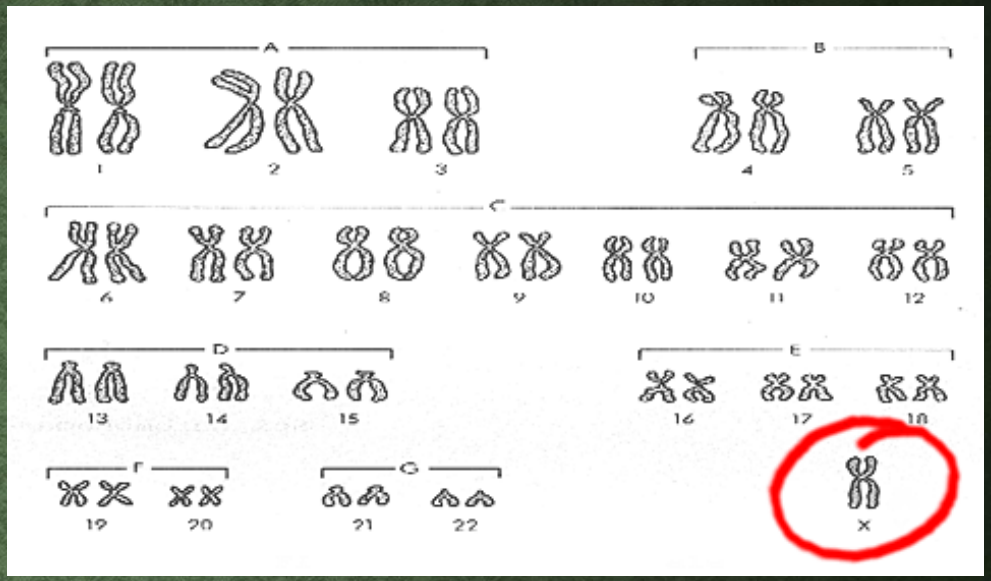
Nondisjunction:

- chromosome pair fails to separate properly during meiosis
- Two main types



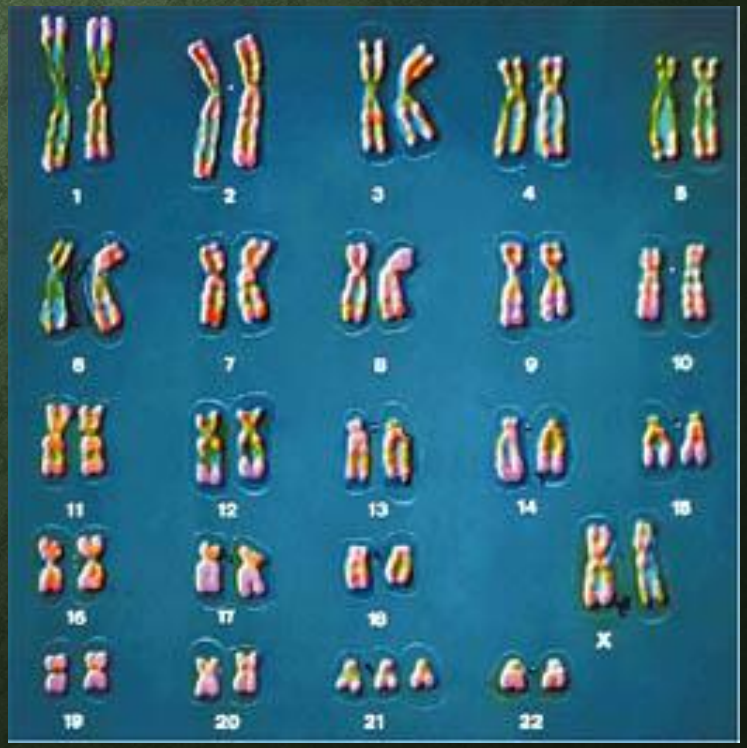
Monosomy:

- gamete has *1 less* chromosome than it should
- 45 chromosomes is the result



Trisomy:

- Gamete has *1 more* chromosome than it should
- Result is 47 chromosomes



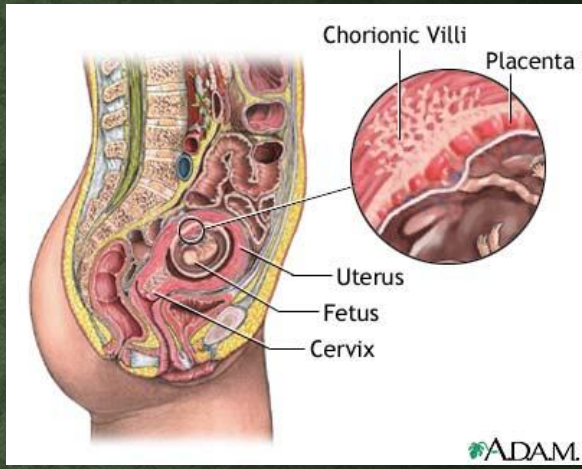
METHODS OF DETECTION

Chorion villi sampling:

•Take sample of the chorion

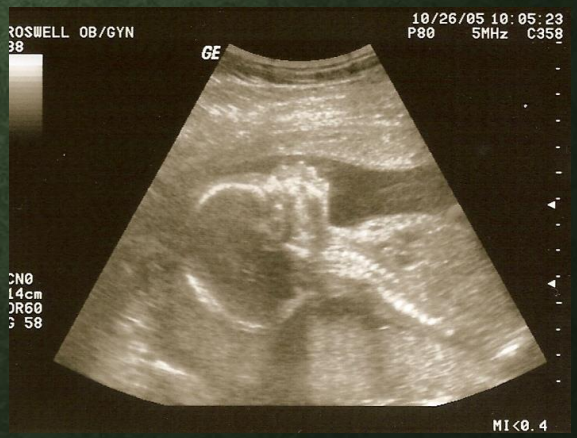
–(membrane surrounding fetus)

•Chemical tests and **Karyotyping** performed



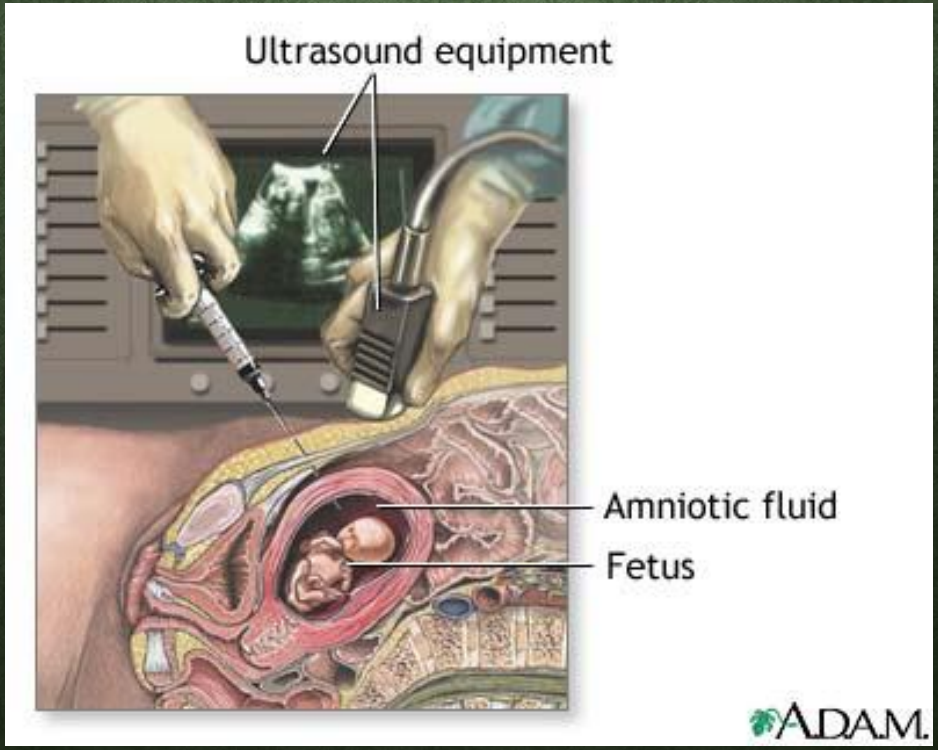
Ultrasound:

- 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



KARYOTYPE
(PICTURE OF AN INDIVIDUAL'S CHROMOSOMES)

One of the ways to analyze the amniocentesis is to make a Karyotype

What genetic disorder does this karyotype show?