Chromosomal Mutations

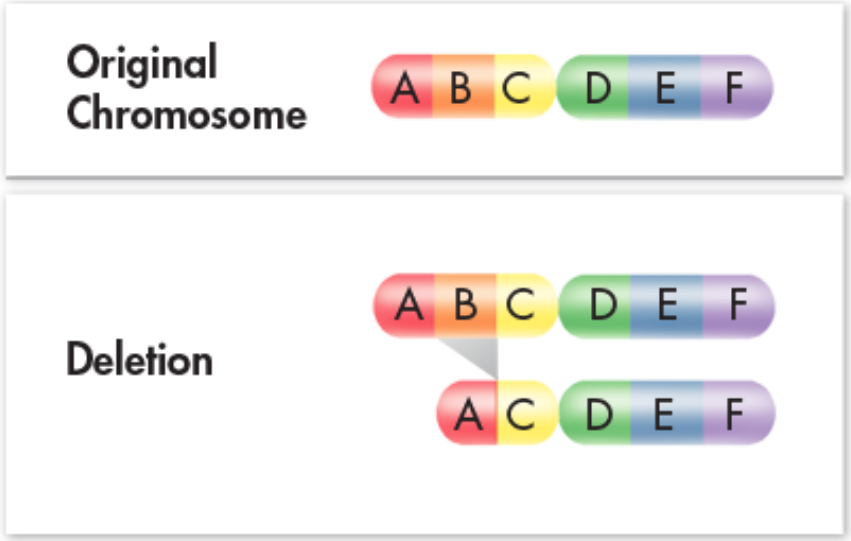
Chromosomal mutations involve changes in the number or structure of chromosomes.

These mutations can change the location of genes on chromosomes and can even change the number of copies of some genes.

There are four types of chromosomal mutations: deletion, duplication, inversion, and translocation.

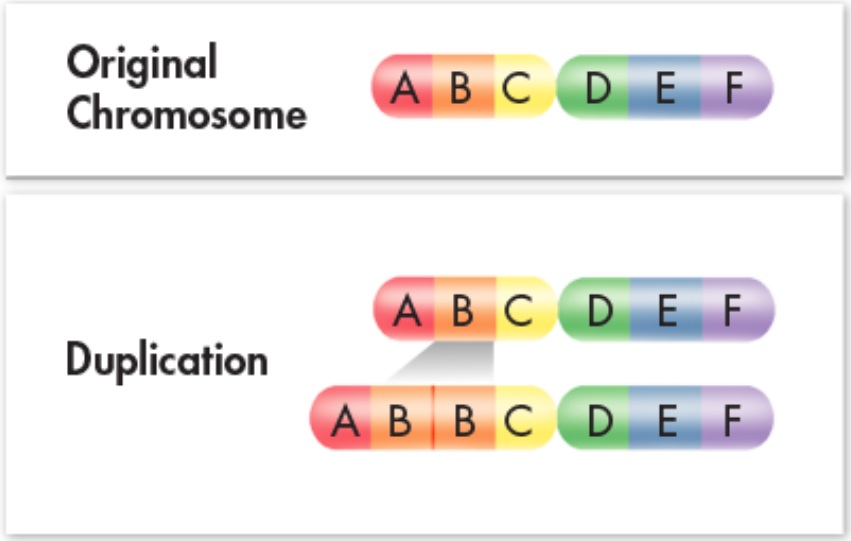
Chromosomal Mutations

Deletion involves the loss of all or part of a chromosome.



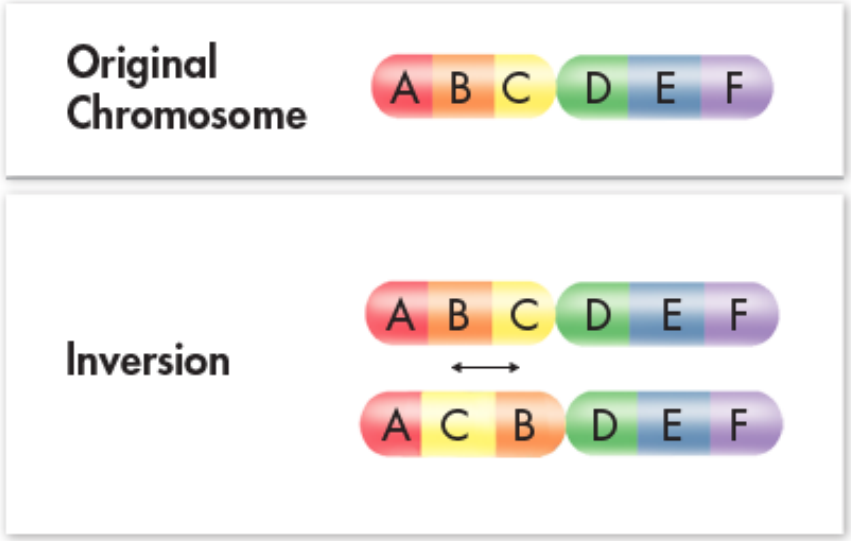
Chromosomal Mutations

Duplication produces an extra copy of all or part of a chromosome.



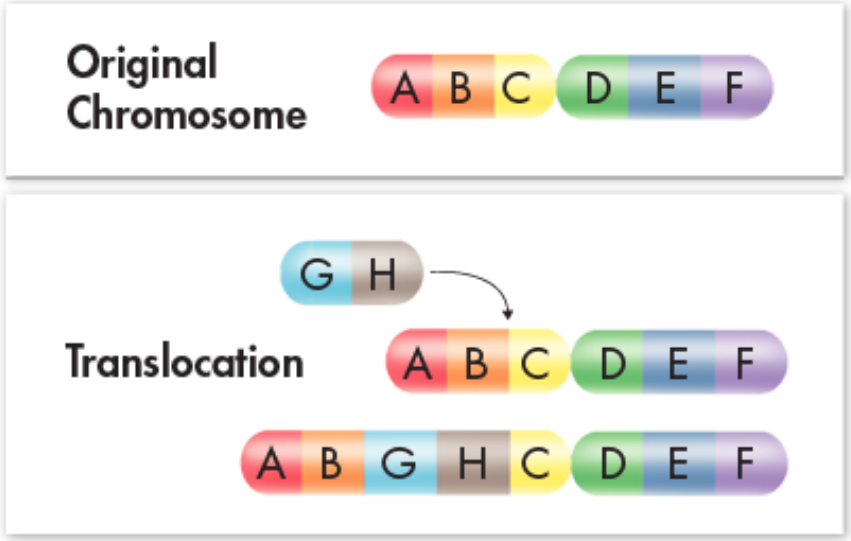
Chromosomal Mutations

Inversion reverses the direction of parts of a chromosome.



Chromosomal Mutations

Translocation occurs when part of one chromosome breaks off and attaches to another.



Mutagens

Some mutations arise from **mutagens**, chemical or physical agents in the environment.

Chemical mutagens include certain pesticides, a few natural plant alkaloids, tobacco smoke, and environmental pollutants.

Physical mutagens include some forms of electromagnetic radiation, such as X-rays and ultraviolet light.

Mutagens

If these mutagens interact with DNA, they can produce mutations at high rates.

Some compounds interfere with base-pairing, increasing the error rate of DNA replication.

Others weaken the DNA strand, causing breaks and inversions that produce chromosomal mutations.

Cells can sometimes repair the damage; but when they cannot, the DNA base sequence changes permanently.

Harmful and Helpful Mutations

The effects of mutations on genes vary widely. Some have little or no effect; and some produce beneficial variations. Some negatively disrupt gene function.

Whether a mutation is negative or beneficial depends on how its DNA changes relative to the organism's situation.

Mutations are often thought of as negative because they disrupt the normal function of genes.

However, without mutations, organisms cannot evolve, because mutations are the source of genetic variability in a species.

Harmful Effects

Some of the most harmful mutations are those that dramatically change protein structure or gene activity.

The defective proteins produced by these mutations can disrupt normal biological activities, and result in genetic disorders.

Some cancers, for example, are the product of mutations that cause the uncontrolled growth of cells.

Harmful Effects

Sickle cell disease is a disorder associated with changes in the shape of red blood cells. Normal red blood cells are round. Sickle cells appear long and pointed.

Sickle cell disease is caused by a point mutation in one of the polypeptides found in hemoglobin, the blood's principal oxygen-carrying protein.

Among the symptoms of the disease are anemia, severe pain, frequent infections, and stunted growth.

Beneficial Effects

Some of the variation produced by mutations can be highly advantageous to an organism or species.

Mutations often produce proteins with new or altered functions that can be useful to organisms in different or changing environments.

For example, mutations have helped many insects resist chemical pesticides.

Some mutations have enabled microorganisms to adapt to new chemicals in the environment.

Beneficial Effects

Plant and animal breeders often make use of "good" mutations.

For example, when a complete set of chromosomes fails to separate during meiosis, the gametes that result may produce triploid (3N) or tetraploid (4N) organisms.

The condition in which an organism has extra sets of chromosomes is called **polyploidy.**

Polyploid plants are often larger and stronger than diploid plants.

Important crop plants—including bananas and limes—have been produced this way.

Chromosomal disorders in humans:

A chromosomal disorders list can be seen below:

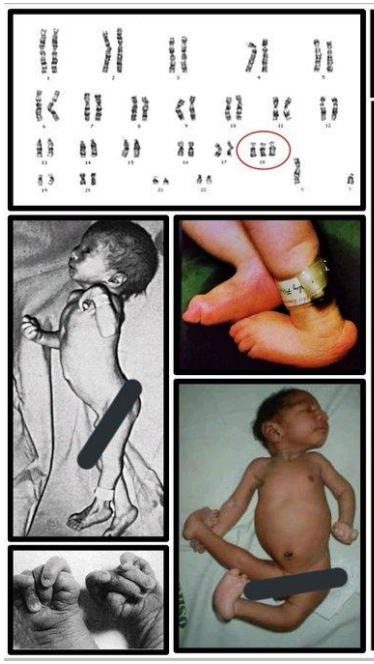
- Trisomy 21: Down Syndrome.
- Trisomy 18.
- Trisomy 13.
- Klinefelter Syndrome.
- XYY Syndrome.
- Turner Syndrome.
- Triple X Syndrome.

Trisomy 18 or Edwards' syndrome

Each cell in your body usually contains 23 pairs of chromosomes, which carry the genes you inherit from your parents.

A baby with Edwards' syndrome has 3 copies of chromosome number 18 instead of 2. This affects the way the baby grows and develops. Having 3 copies of chromosome 18 usually happens by chance, because of a change in the sperm or egg before a baby is conceived.

Your chance of having a baby with Edwards' syndrome increases as you get older, but anyone can have a baby with Edwards' syndrome. The condition does not usually run in families and is not caused by anything the parents have or have not done.



2) Trisomy 18 (Edwards syndrome)

❖ **Clinical features**
- ✓ **Rocker bottom feet**
- ✓ **Overlapping fingers**
- ✓ **Clenched fist**
- ✓ Short sternum
- ✓ Hypoplastic nails
- ✓ Hypoplastic nasal alae
- ✓ IUGR
- ✓ Intellectual disability
- ✓ Microcephaly
- ✓ Hypertonia
- ✓ Prominent occiput
- ✓ Micrognathia
- ✓ Limited hip abduction
- ✓ Cleft lip / palate

2nd most common autosomal trisomy

❖ **Most common cardiac defect ?**
- ✓ **Ventricular septal defect (VSD)**
- ✓ Atrial septal defect (ASD)
- ✓ PDA

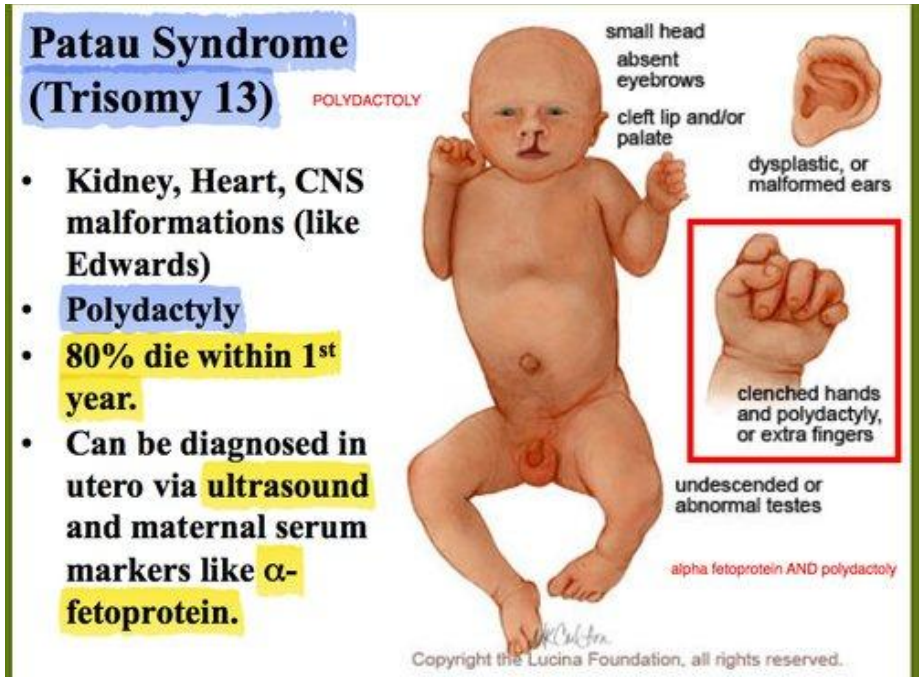
❖ **Most common cause of death?**
- ✓ Central apnea

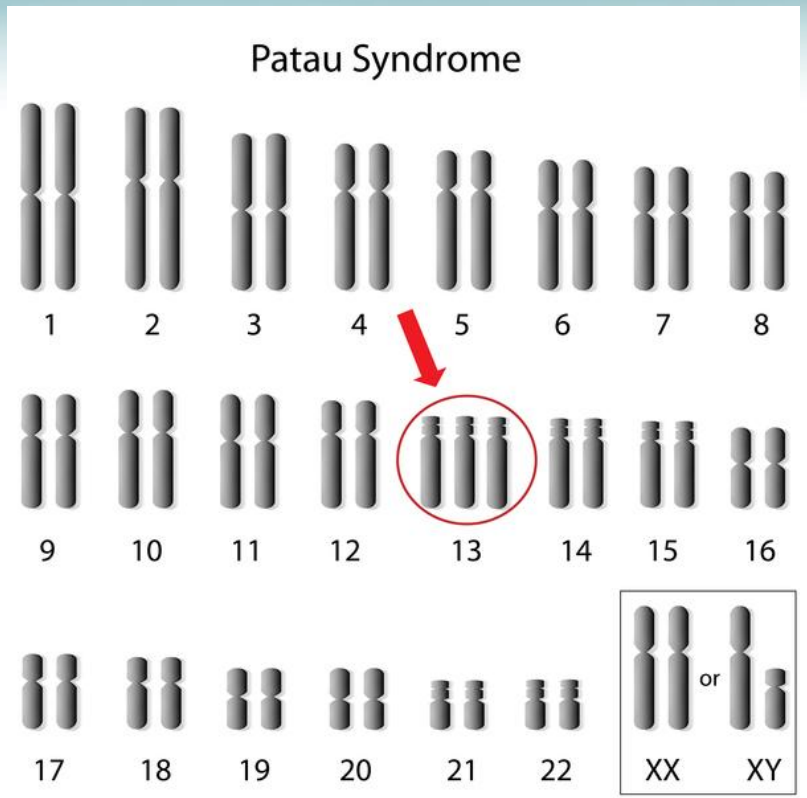
❖ **Prenatal screen lab finding?**
- ✓ Low α feto protein
- ✓ Low estriol
- ✓ Low hCG

Pediatric On Squares

❖ **Prognosis**
- ✓ 50% die in the 1st week of life
- ✓ 90% die by 1 year of age

Trisomy 13 or Patau Syndrome





Klinefelter syndrome

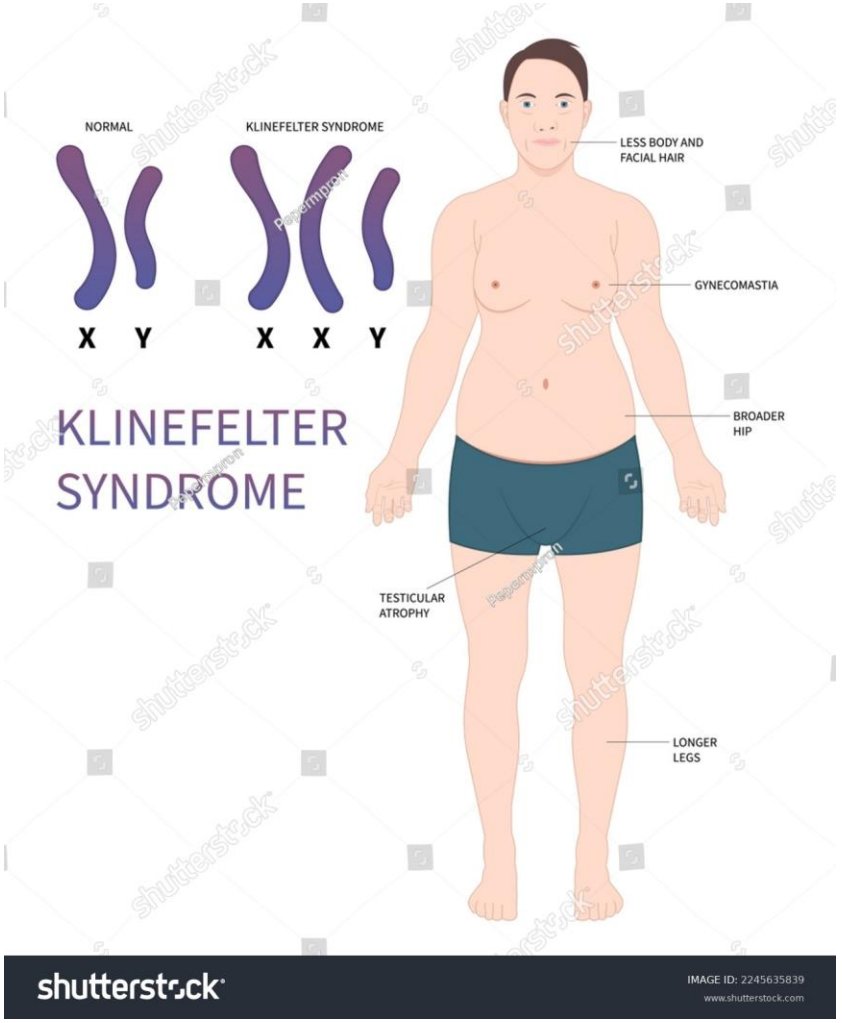
Klinefelter syndrome (sometimes called Klinefelter's, KS or XXY) is where boys and men are born with an extra X chromosome.

Chromosomes are packages of genes found in every cell in the body. There are 2 types of chromosome, called the sex chromosomes, that determine the genetic sex of a baby. These are named either X or Y.

Usually, a female baby has 2 X chromosomes (XX) and a male has 1 X and 1 Y (XY). But in Klinefelter syndrome, a boy is born with an extra copy of the X chromosome (XXY).

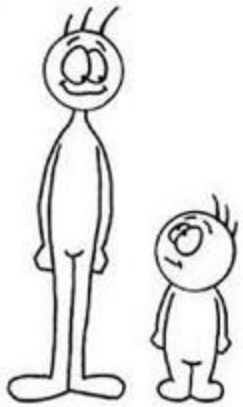
The X chromosome is not a "female" chromosome and is present in everyone. The presence of a Y chromosome denotes male sex.

Boys and men with Klinefelter syndrome are still genetically male, and often will not realise they have this extra chromosome, but occasionally it can cause problems that may require treatment.



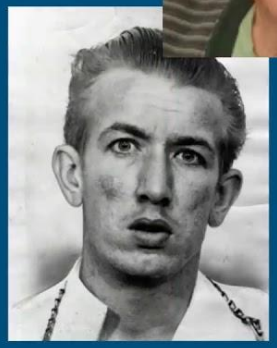
XYY Syndrome

- Males only.
- Affects pair 23.
- Extra copy of the Y chromosome.
- Taller than average.
- Increased risk of learning disabilities.
- Prone to severe acne.
- May include antisocial or behavioral problems.



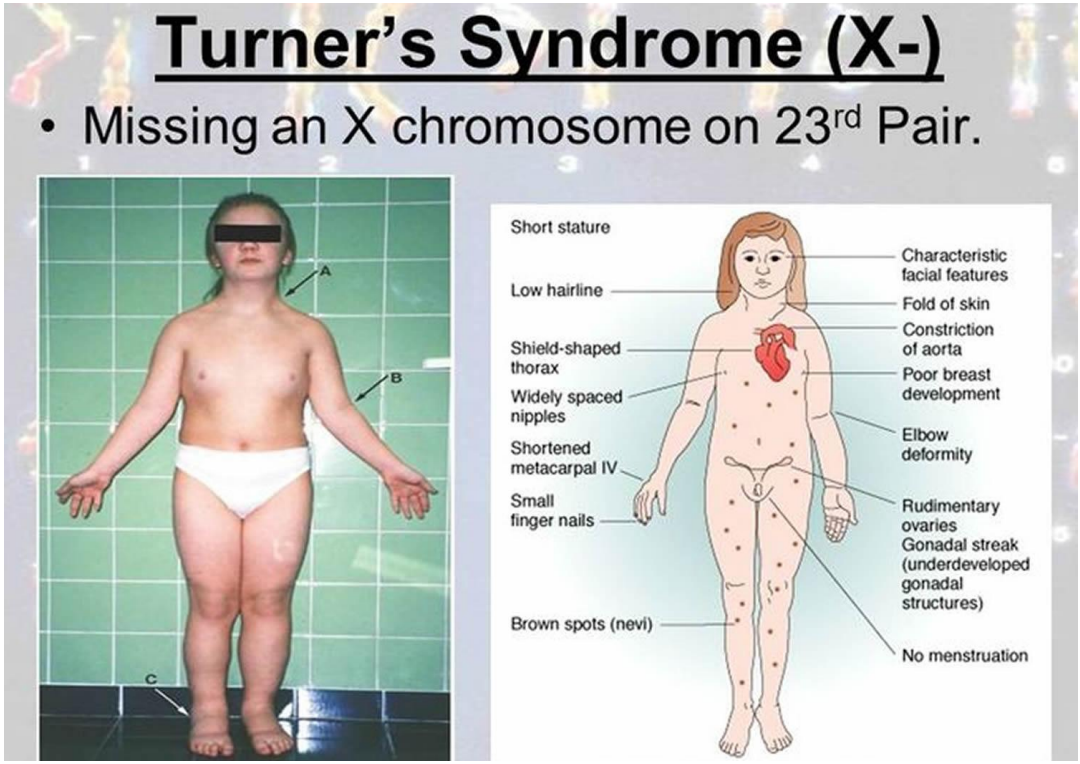
History of XYY

- First recorded case in July 1961, found by Dr. Avery Sandberg
- Studies linked those with XYY to criminality
- Richard Speck, mass murderer, was thought to have XYY
  - Later genetic testing found he was XY



Turner syndrome

Turner syndrome, a condition that affects only females, results when one of the X chromosomes (sex chromosomes) is missing or partially missing. Turner syndrome can cause a variety of medical and developmental problems, including short height, failure of the ovaries to develop and heart defects.



Triple X syndrome

Triple X syndrome, also called trisomy X or 47,XXX, is a genetic disorder that affects about 1 in 1,000 females. Females normally have two X chromosomes in all cells — one X chromosome from each parent. In triple X syndrome, a female has three X chromosomes.