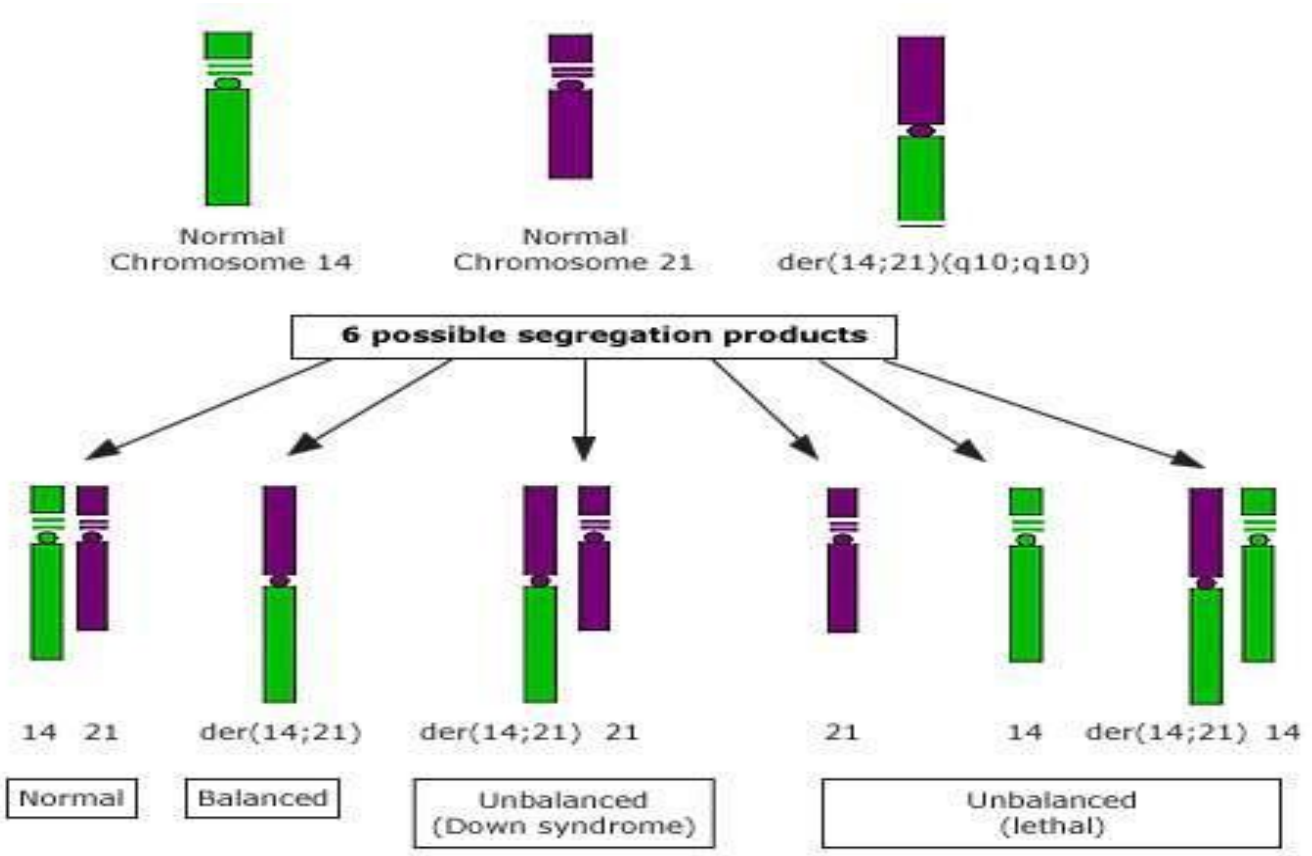
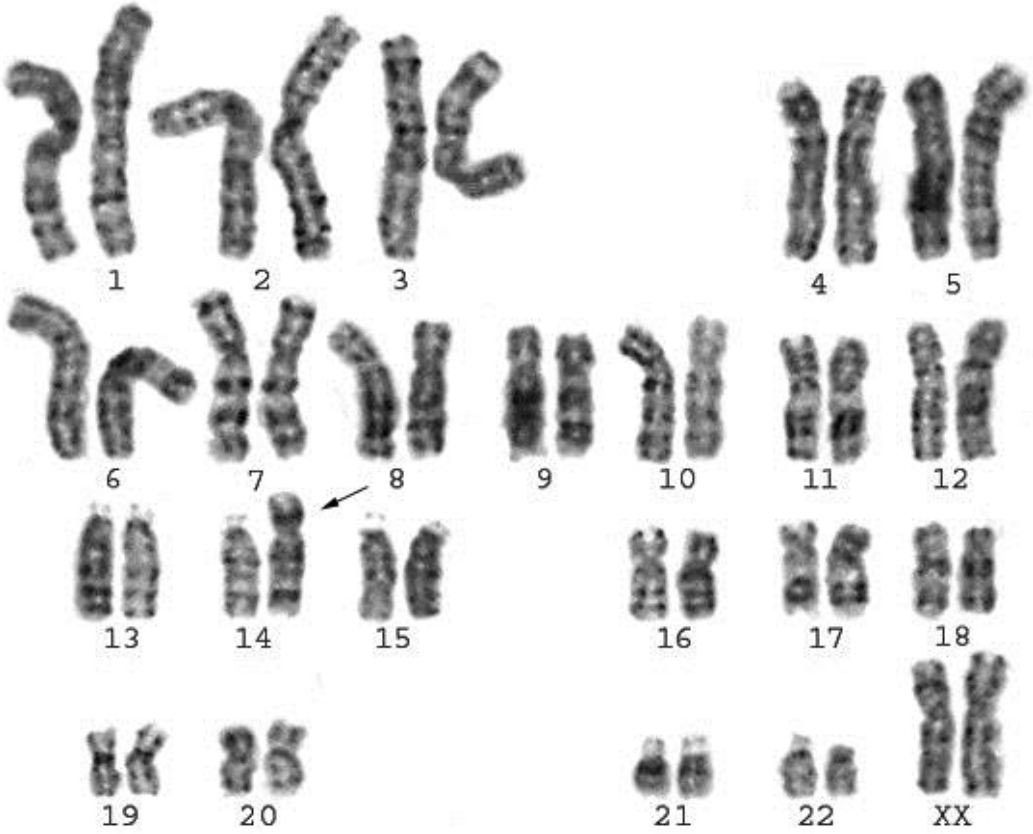
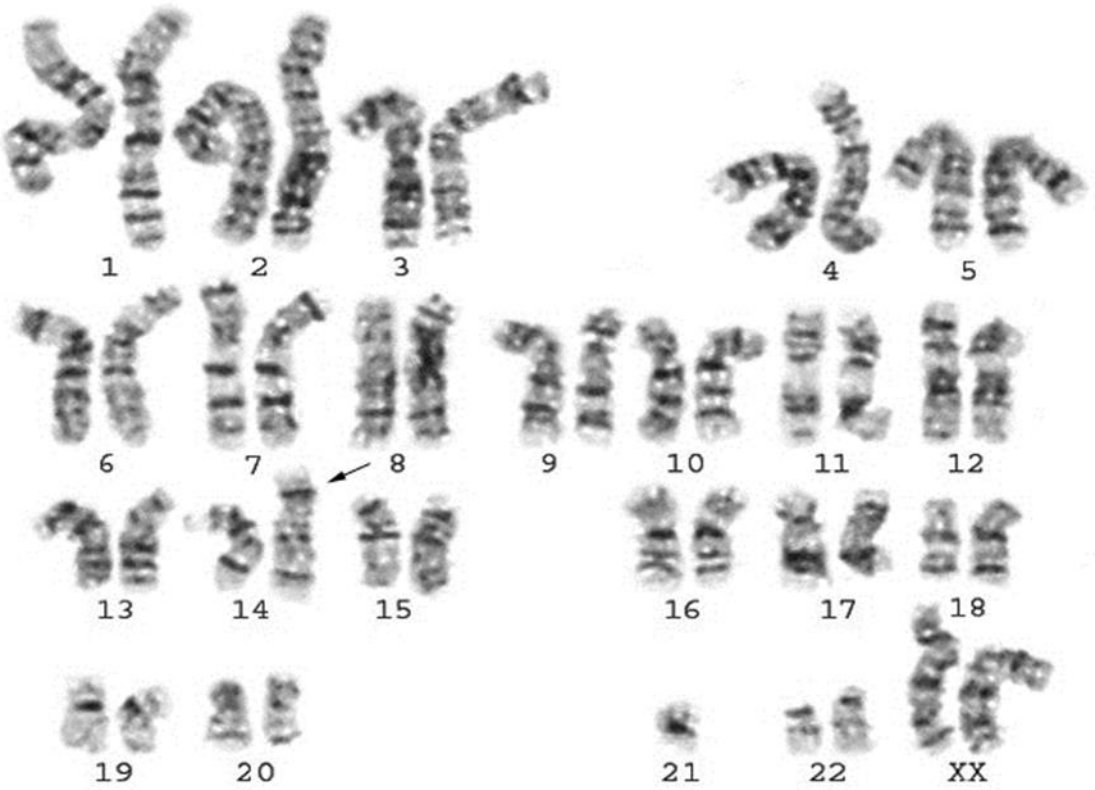
Learning Objectives

- Know some examples of numerical chromosomal abnormalities.
- What are structural chromosomal abnormalities?

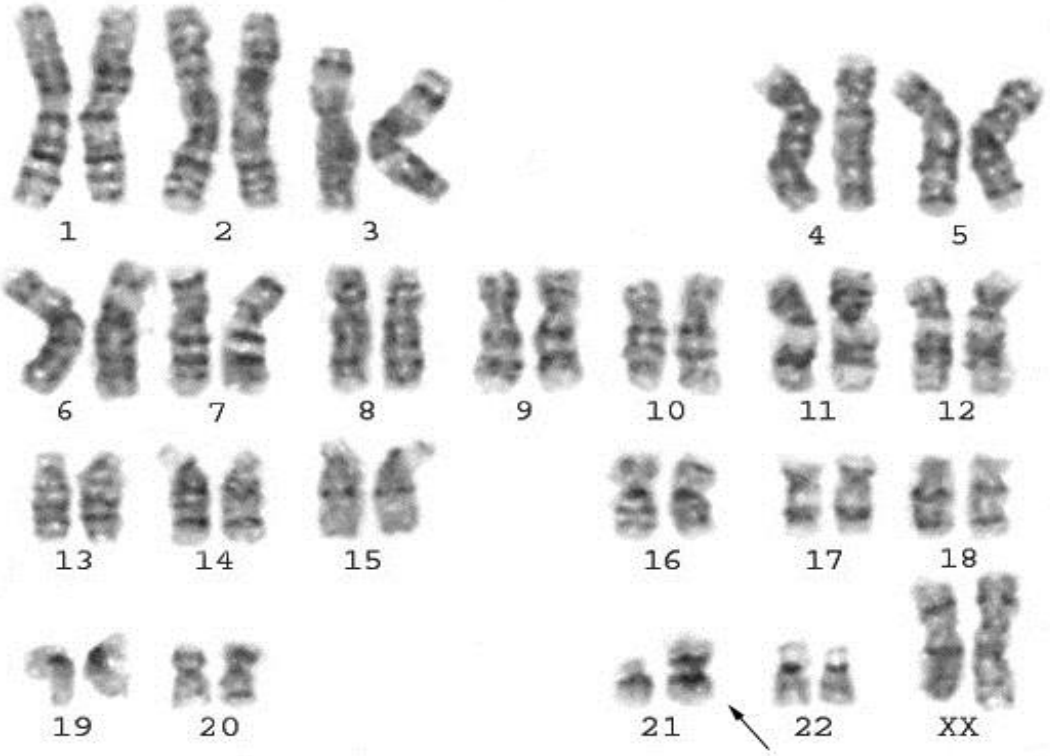

Normal
Chromosome 14
Normal
Chromosome 21
der(14;21)(q10;q10)
6 possible segregation products
14 21
der(14;21)
der(14;21) 21
21
14
der(14;21) 14
Normal
Balanced
Unbalanced
(Down syndrome)
Unbalanced
(lethal)



46,XX,t(14;21)(q10;q10),+21



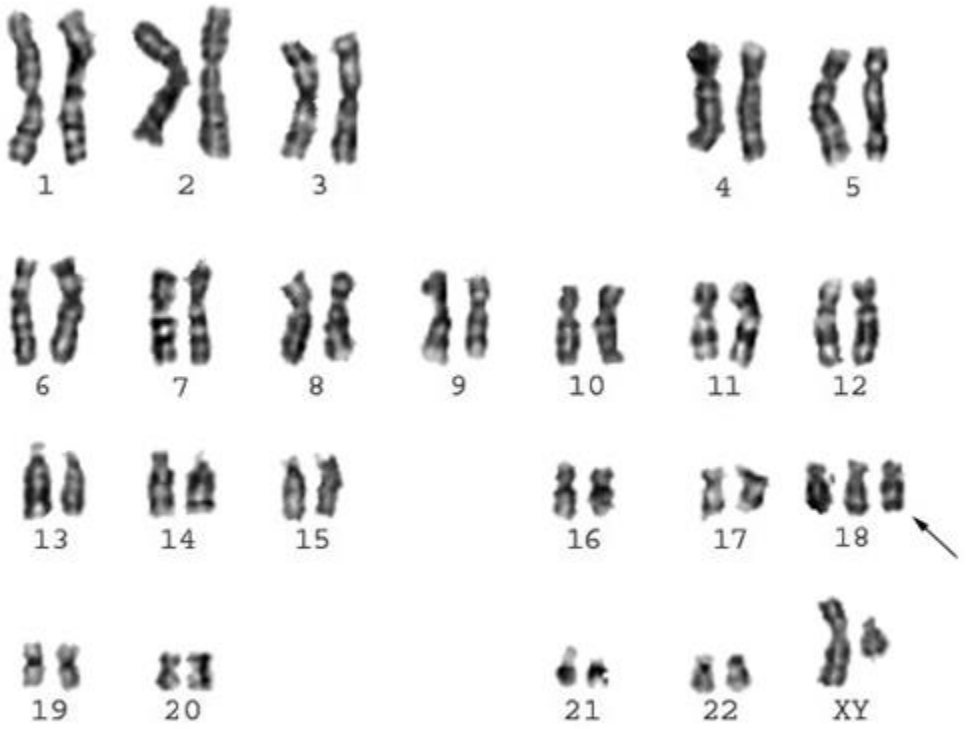
45,XX,t(14;21)(q10;q10)



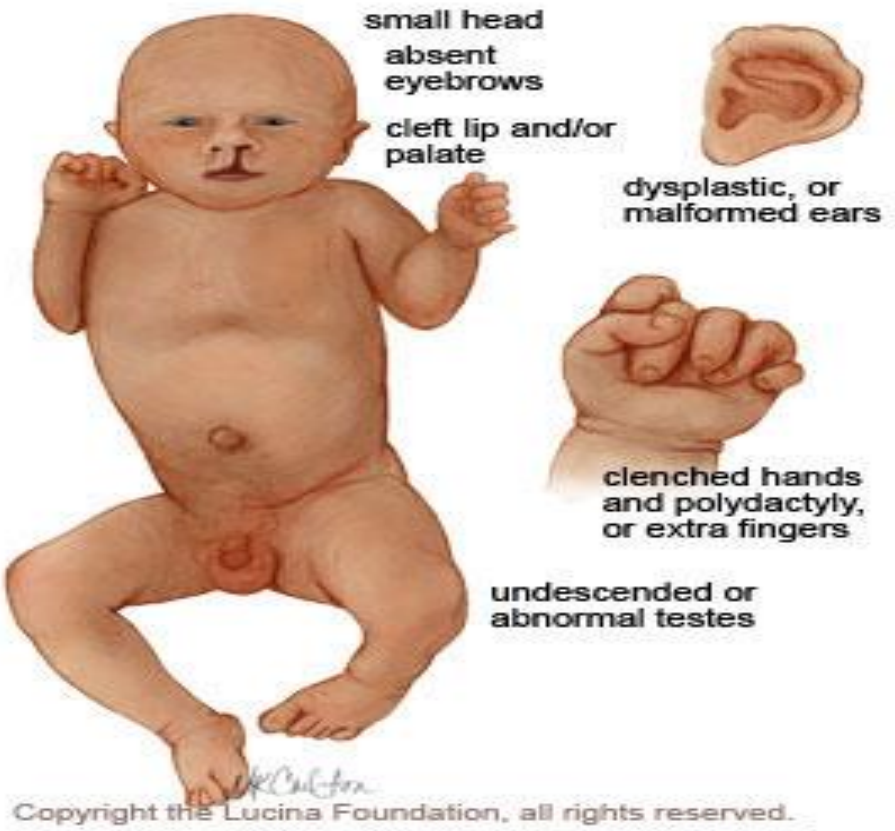
46,XX,+21,der(21;21)(q10;q10)

Trisomy 18(Edward Syndrome)

47,XY,+18

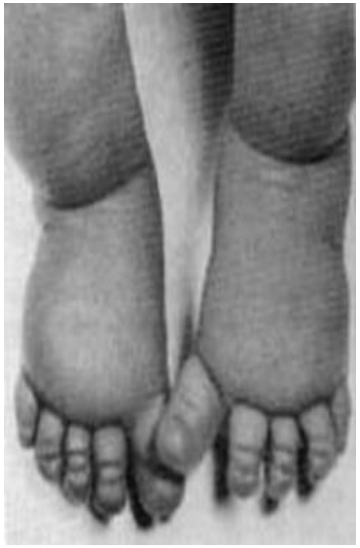


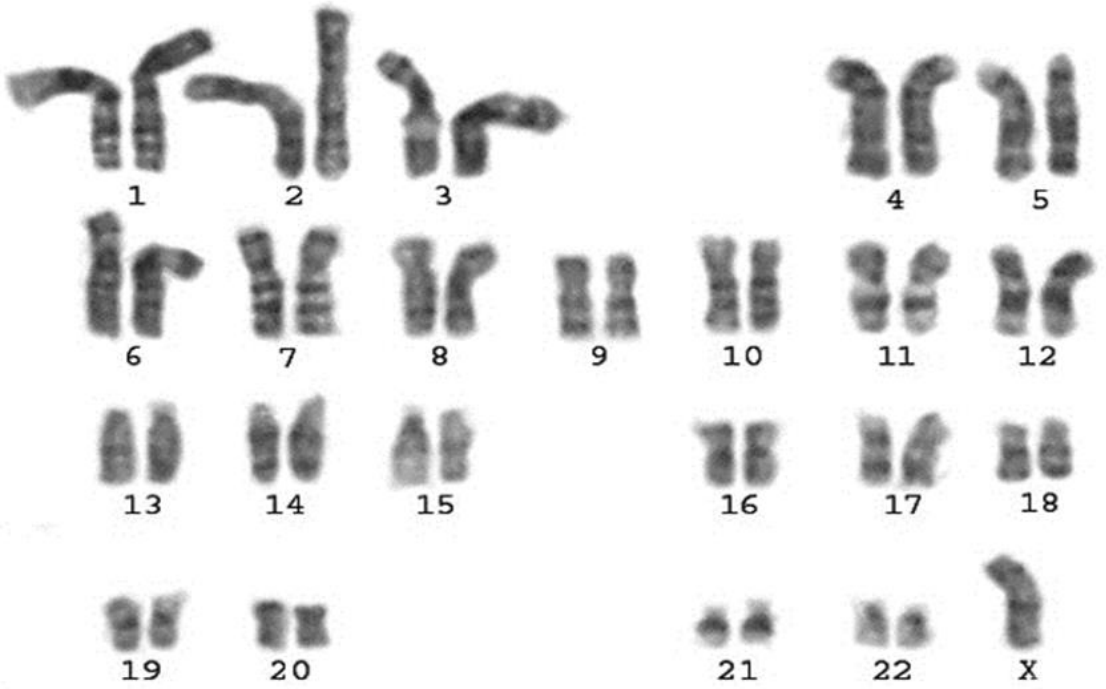
Trisomy 13 (Patau Syndrome)



Turner Syndrome

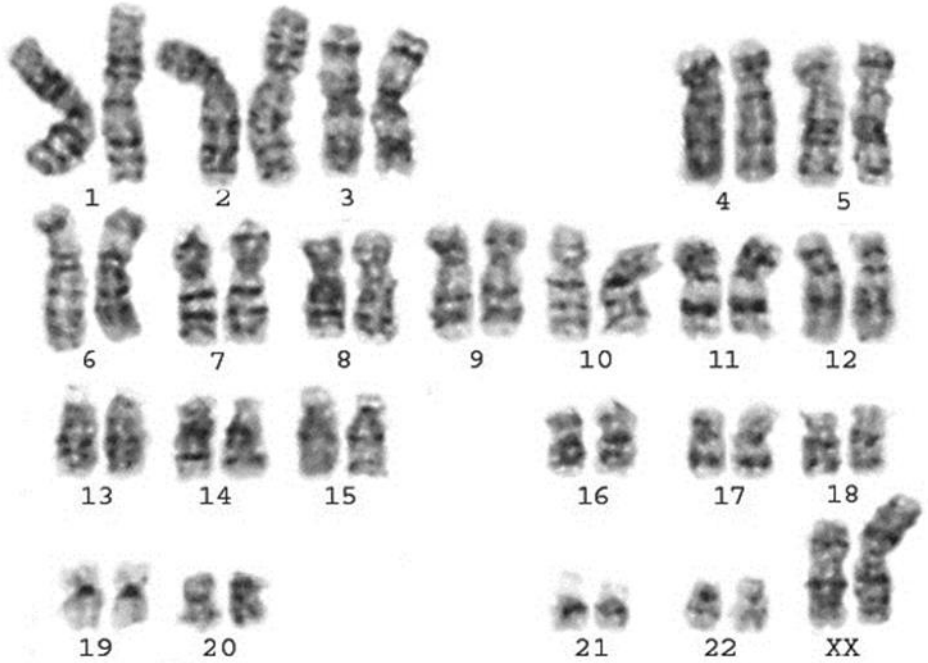
- This condition occurs in about 1 in 2,500 newborn girls worldwide, but it is much more common among pregnancies that do not survive to term (abortions).
- Turner syndrome is a chromosomal condition that affects development in females. The most common feature of Turner syndrome is short stature. An early loss of ovarian function is also very common, and most are infertile.
- About 30 percent of females with Turner syndrome have extra folds of skin on the neck (webbed neck), puffiness or swelling (lymphedema) of the hands and feet. One third to one half of individuals with Turner syndrome are born with a heart defect.
- Most girls and women with Turner syndrome have normal intelligence.





45,X

- About half of individuals with Turner syndrome have monosomy X.
- Turner syndrome can also occur if one of the sex chromosomes is partially missing or rearranged rather than completely absent.
- Some women with Turner syndrome have a chromosomal change in only some of their cells, which is known as mosaicism

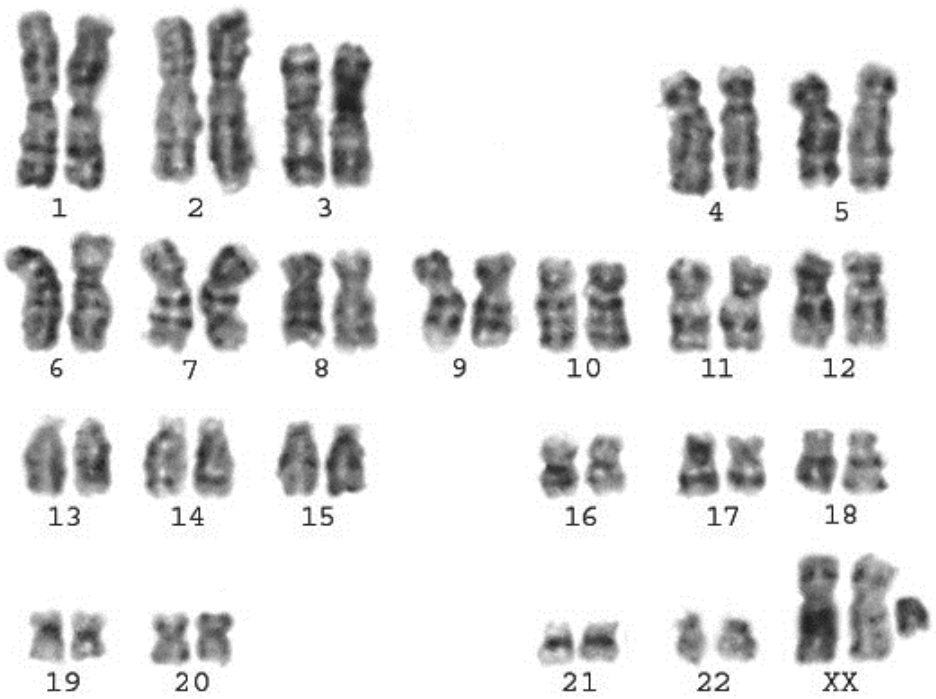


46,X,i(Xq)

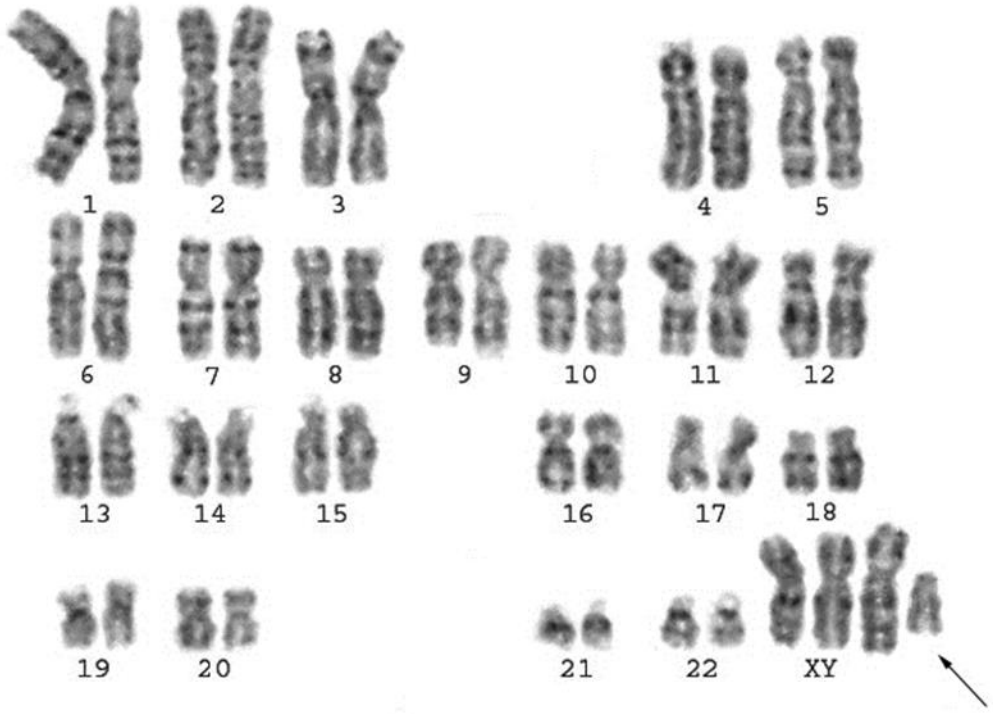
Klinefelter Syndrome

- Klinefelter syndrome is a chromosomal condition that affects male physical and cognitive development.
- Affected individuals typically have small testes that do not produce as much testosterone as usual which can lead to delayed or incomplete puberty, breast enlargement (gynecomastia), reduced facial and body hair, and infertility.
- Older children and adults with Klinefelter syndrome tend to be taller than their peers.
- Children with Klinefelter syndrome may have learning disabilities and delayed speech and language development.

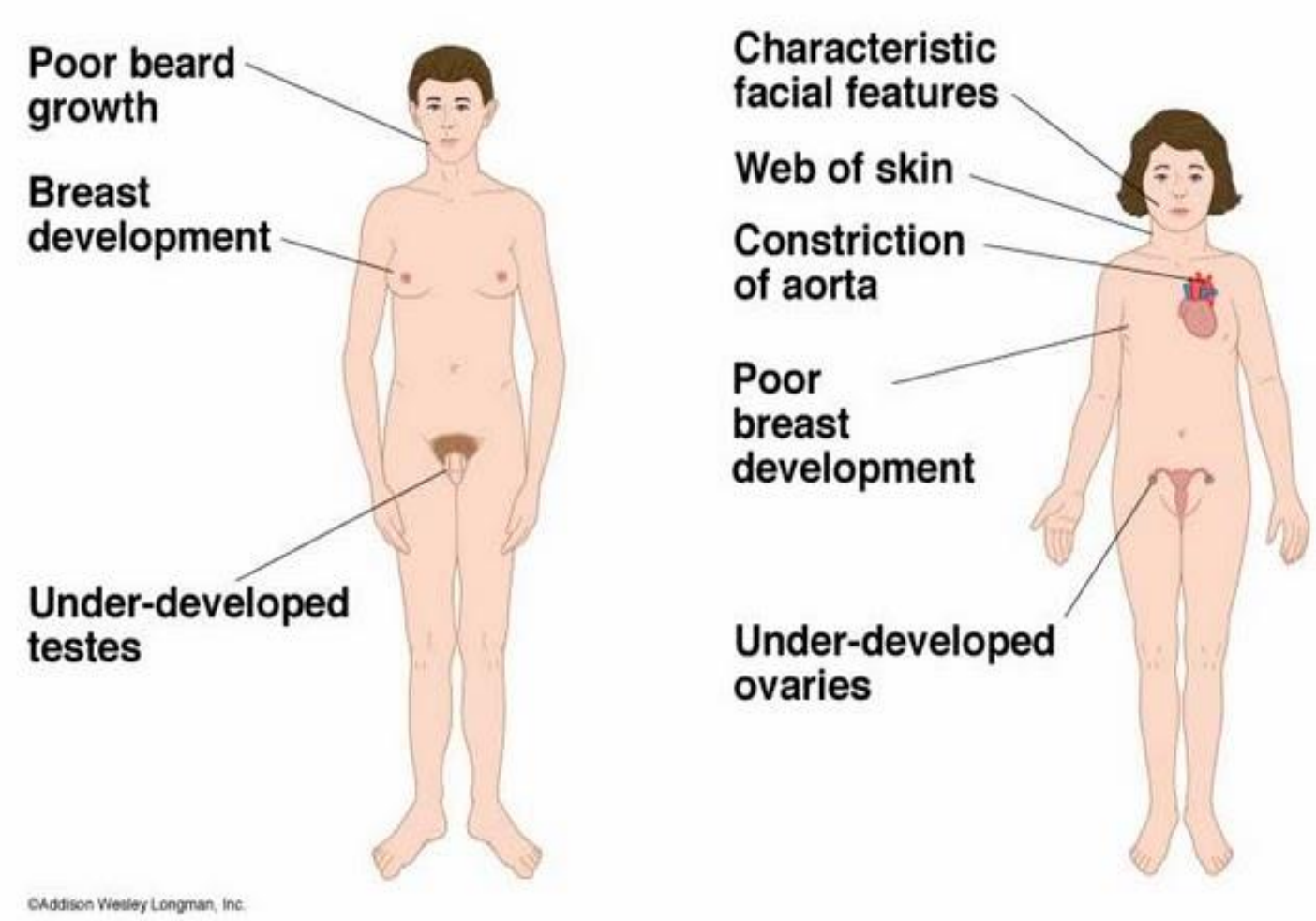
- Klinefelter syndrome affects 1 in 500 to 1,000 newborn males.
- Klinefelter syndrome results from the presence of one extra copy of the X chromosome in each cell (47,XXY).



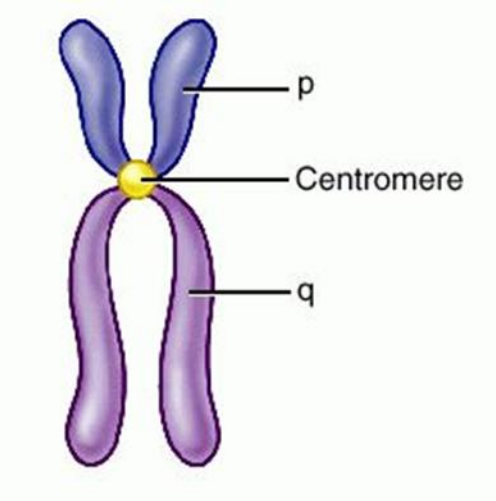
47,XXY



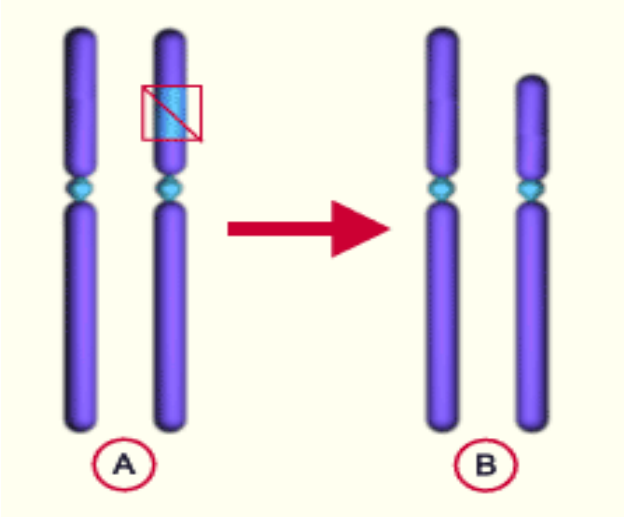
48,XXXY

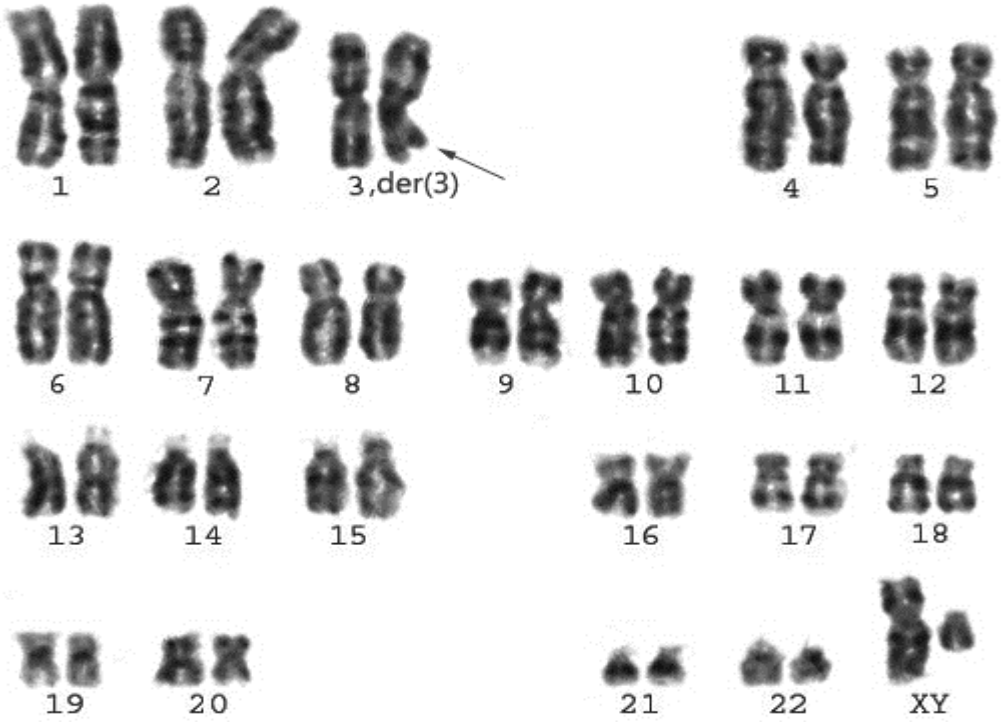

Poor beard growth
Breast development
Under-developed testes
©Addison Wesley Longman, Inc.
Characteristic facial features
Web of skin
Constriction of aorta
Poor breast development
Under-developed ovaries

Structural Abnormalities



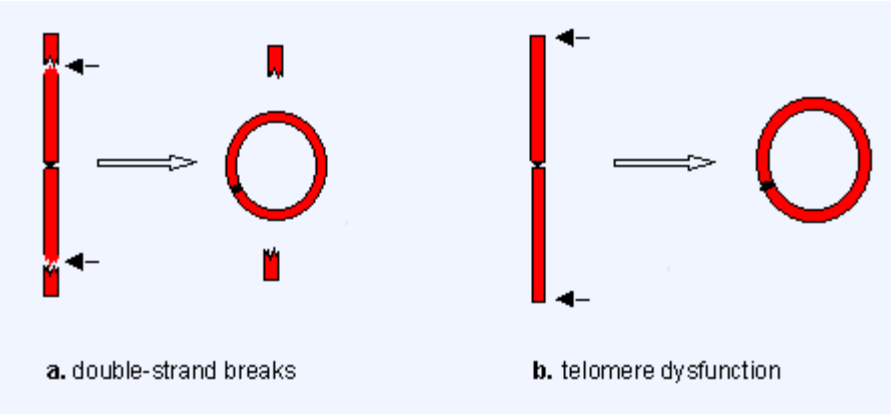
- Deletion: It means loss of a portion of chromosomes.



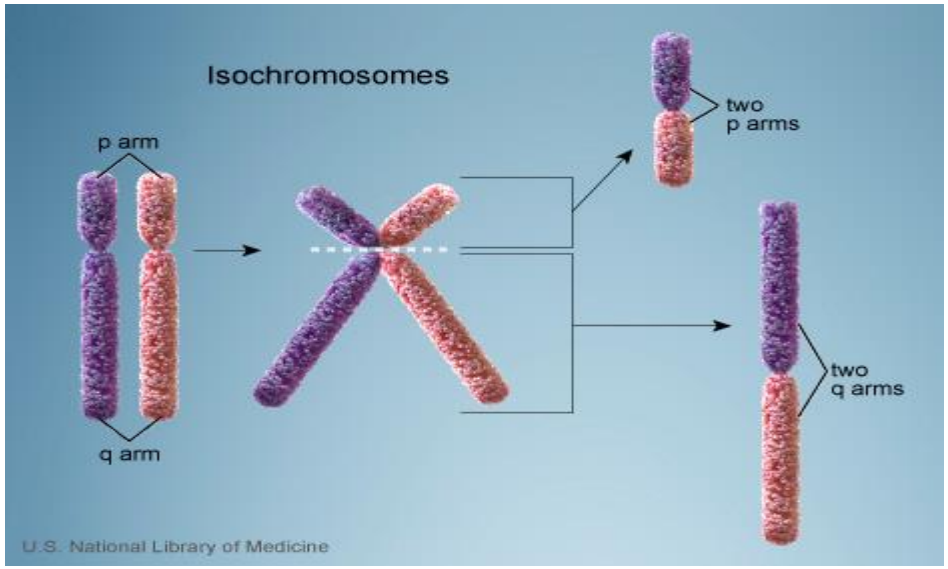


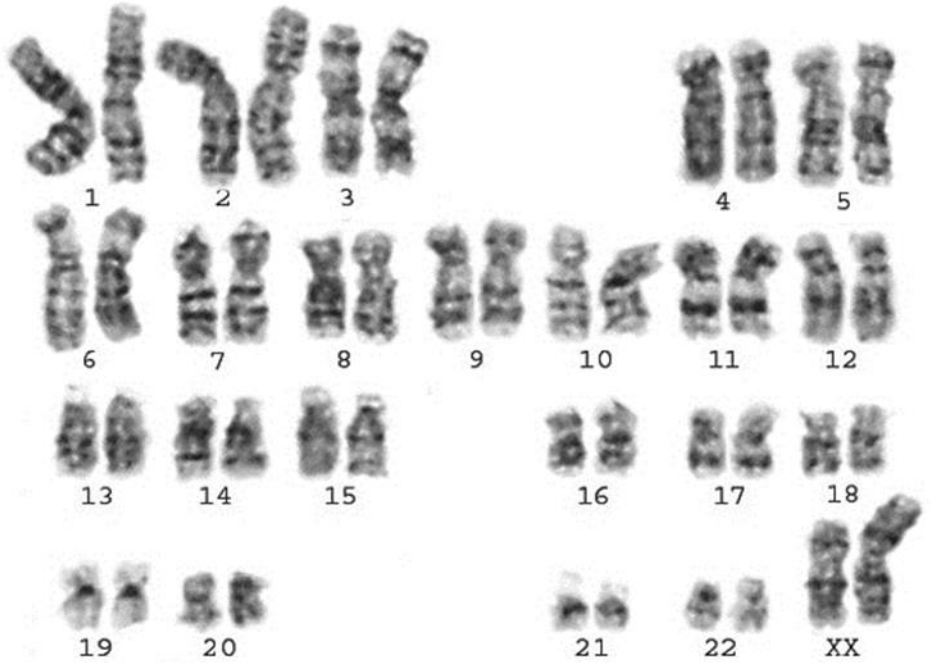
46,XY,del(3)(q26q27.2)

Ring chromosome: It means deletion of both ends of chromosomes with fusion of damaged ends.



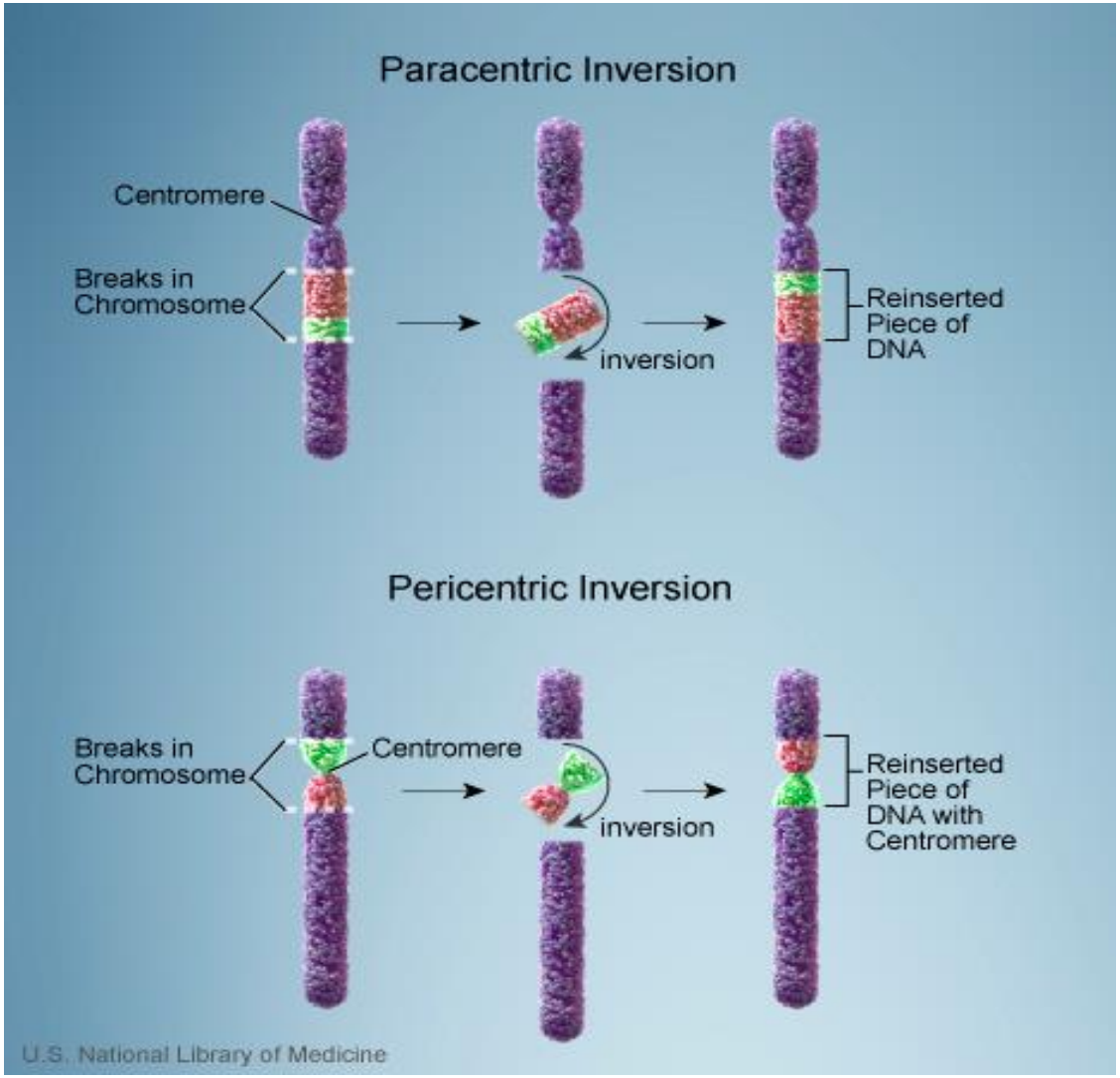
- **Isochromosome:** A chromosome produced by transverse splitting of the centromere so that both arms are from the same side of the centromere, are of equal length, and possess identical genes.

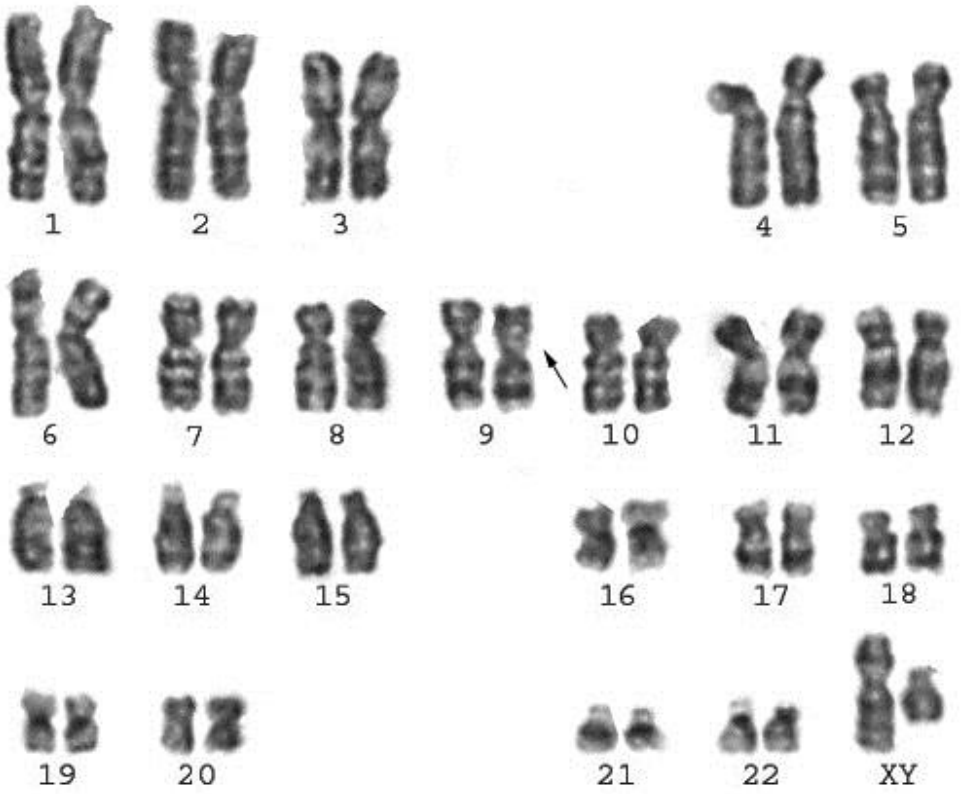




46,X,i(Xq)

- Inversion: A chromosomal rearrangement in which a segment of genetic material is broken away from the chromosome, inverted from end to end, and re-inserted into the chromosome at the same breakage site.


Paracentric Inversion
Centromere
Breaks in Chromosome
inversion
Reinserted Piece of DNA
Pericentric Inversion
Breaks in Chromosome
Centromere
inversion
Reinserted Piece of DNA with Centromere
U.S. National Library of Medicine



46,XY,inv(9)(p11q13)