Nondisjunction disorders

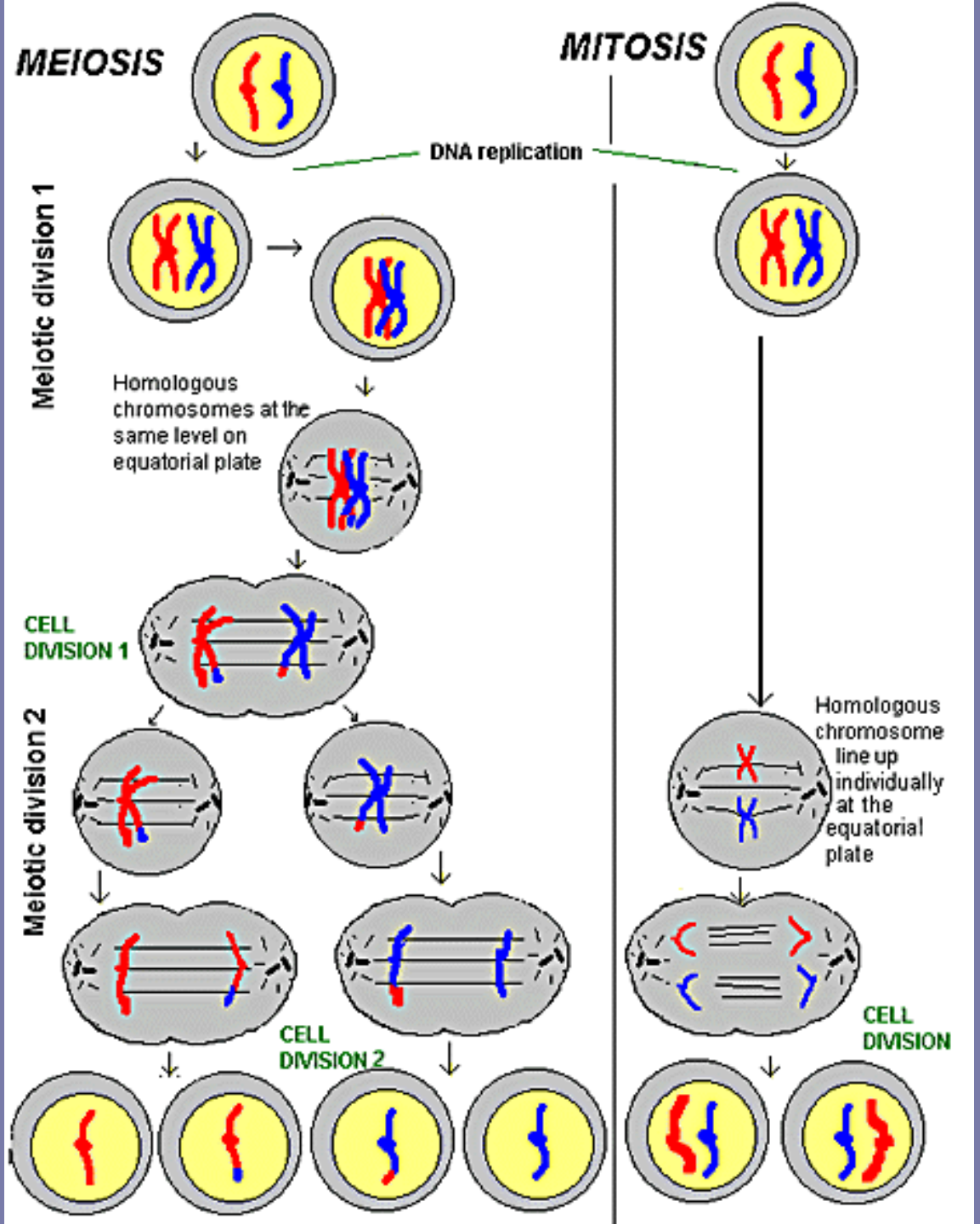
Mr.Yeung

Nondisjunction disorders

- Mutations where chromosomes fail to separate properly during Meiosis I or II

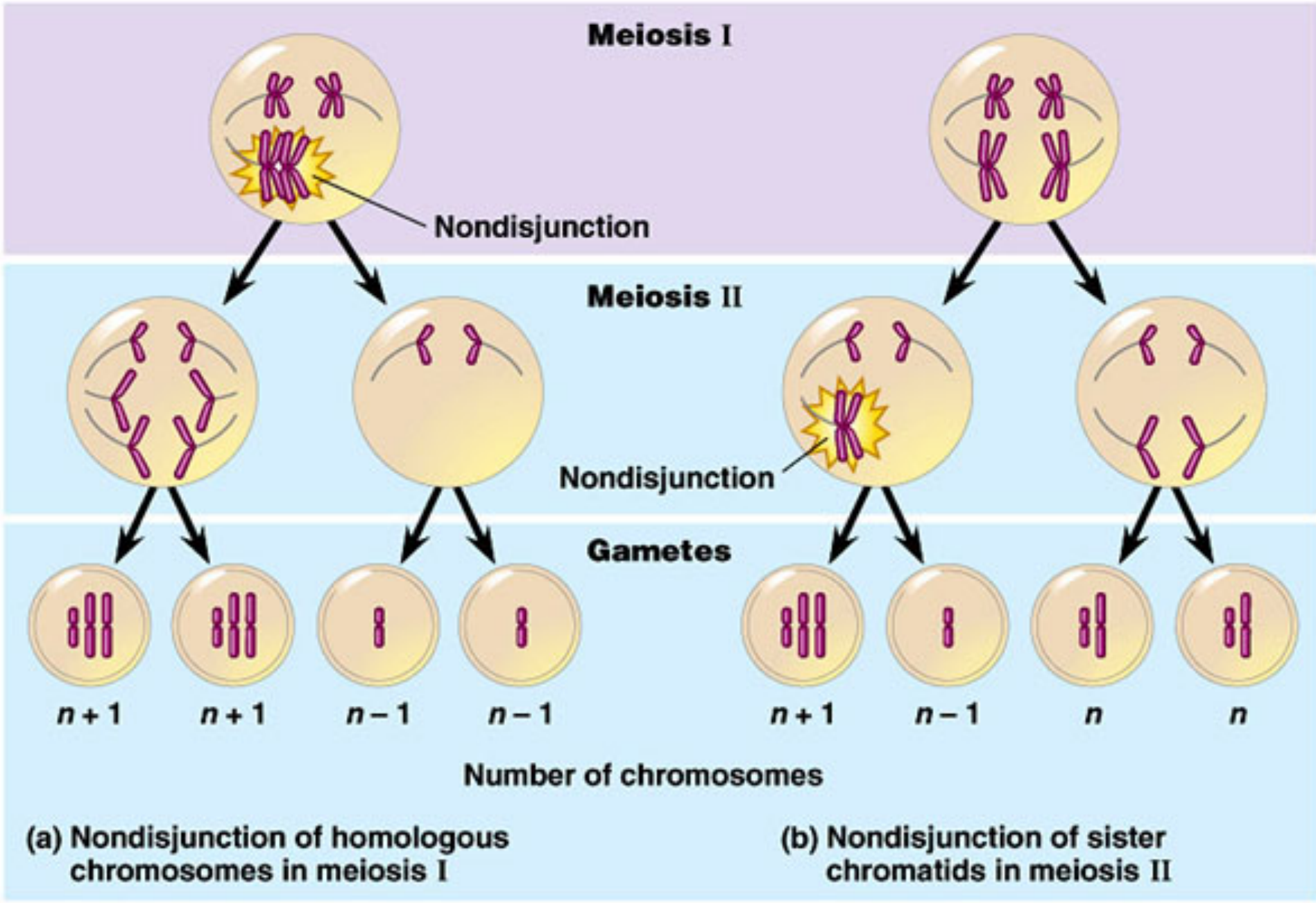
Review on Meiosis

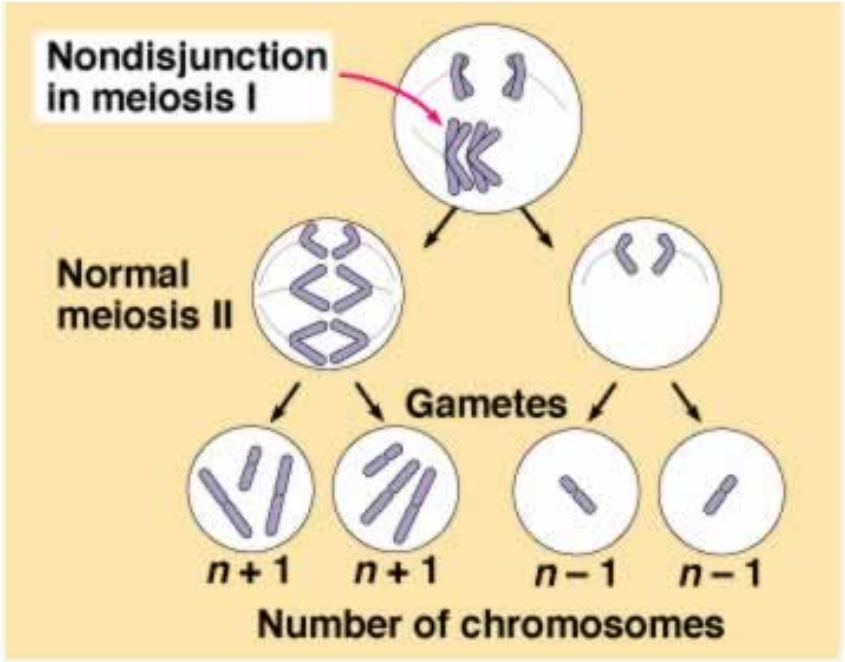
- Occurs in our sex cells (sperm and eggs)
- Give rise to haploid (1n) or half the number of the total amount of chromosomes
- In humans' sex cells, n = 23, or 23 chromosomes.
- After fertilization, the cell will have 2n = 46 due to egg (23 chromosomes) + sperm (23 chromosomes)

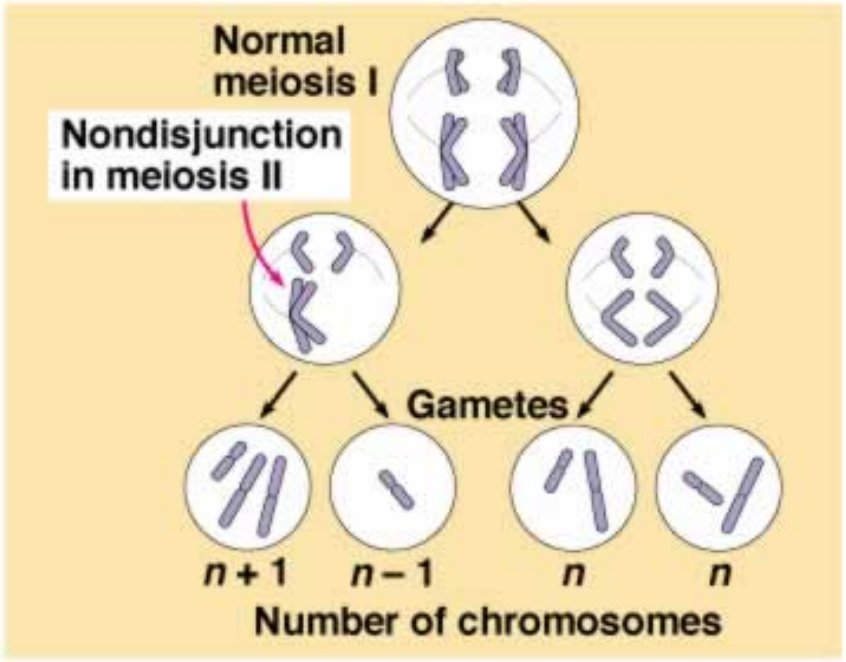

MEIOSIS
MITOSIS
DNA replication
Meiotic division 1
Homologous chromosomes at the same level on equatorial plate
CELL DIVISION 1
Meiotic division 2
Homologous chromosome line up individually at the equatorial plate
CELL DIVISION 2
CELL DIVISION

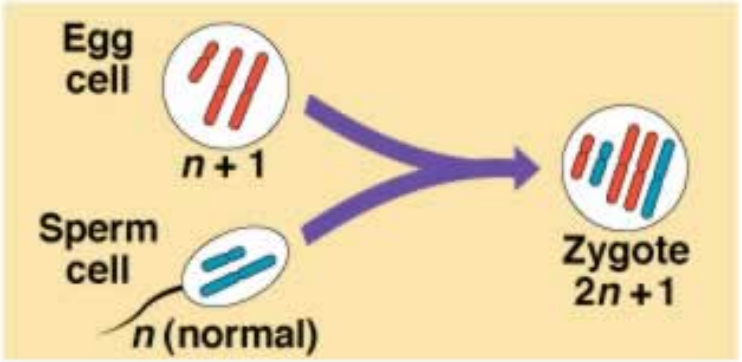
So… what happens when the chromosomes fail to separate?

- Instead of having 1 set of the chromosome in the final sex cell (sperm or egg)
- You might have one with
  - 1 extra chromosome
  - No chromosome(s)
  - 2 extra chromosomes







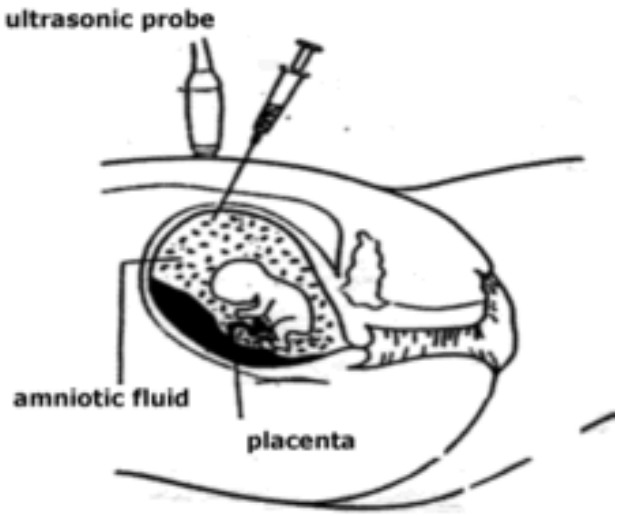


Disorders associated with nondisfunction mutations

- Down syndrome – Extra copy chromosome on chromosome #21
- Klinefelter Syndrome – Extra X (the sex chrosomosome) chromosome in their cells resulting in having XXY
- Turner Syndrome – Missing 1 X chromosome.

How do we detect these chromosomal mutations?

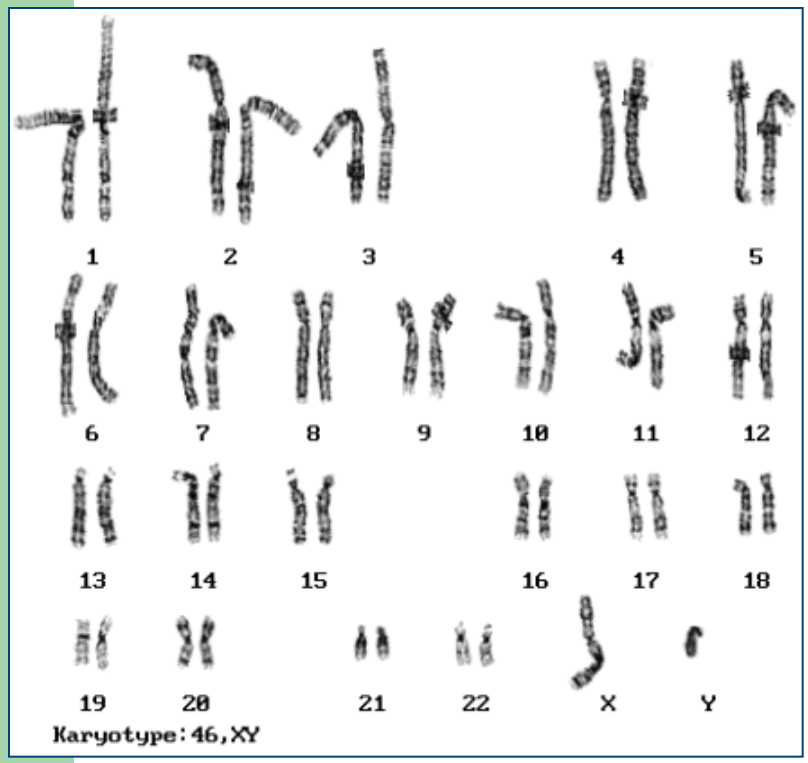
- Amniocentesis
  - Takes sample of amniotic fluid (the liquid the fetus grows in) from the mother
  - In the fluid, there are cells where the baby has shed from his/her skin and bladder
  - The cells are grown in the lab and then examined under a microscope



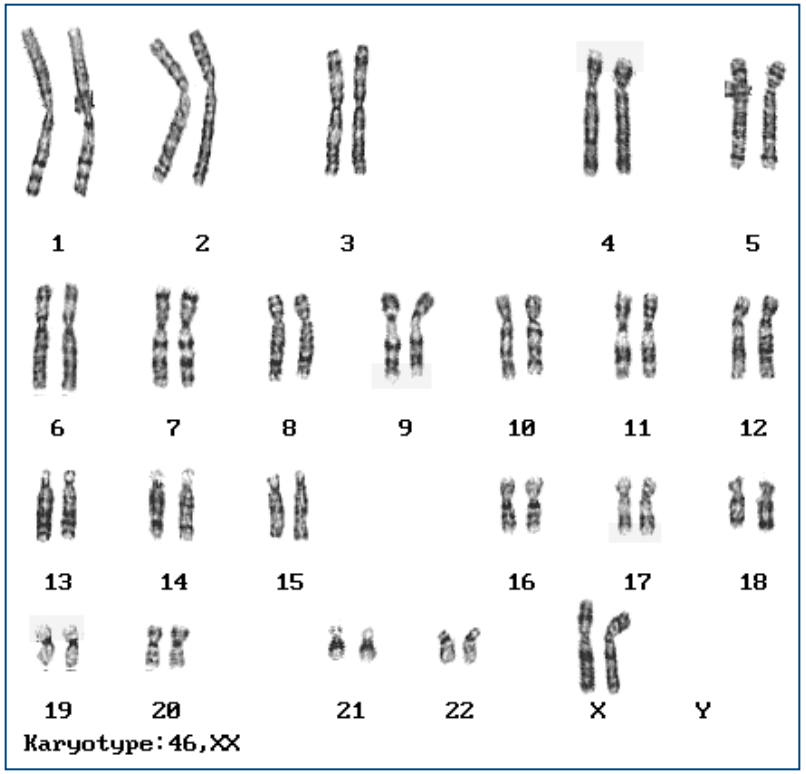
After the cells have been collected…

- The growth of the cells are stopped in metaphase where the chromosomes are clearly visible
- The cells are then stained so the chromosomes are visible.
- The chromosomes are identified and placed beside their respective homologous chromosome and analyzed.

Karyotypes

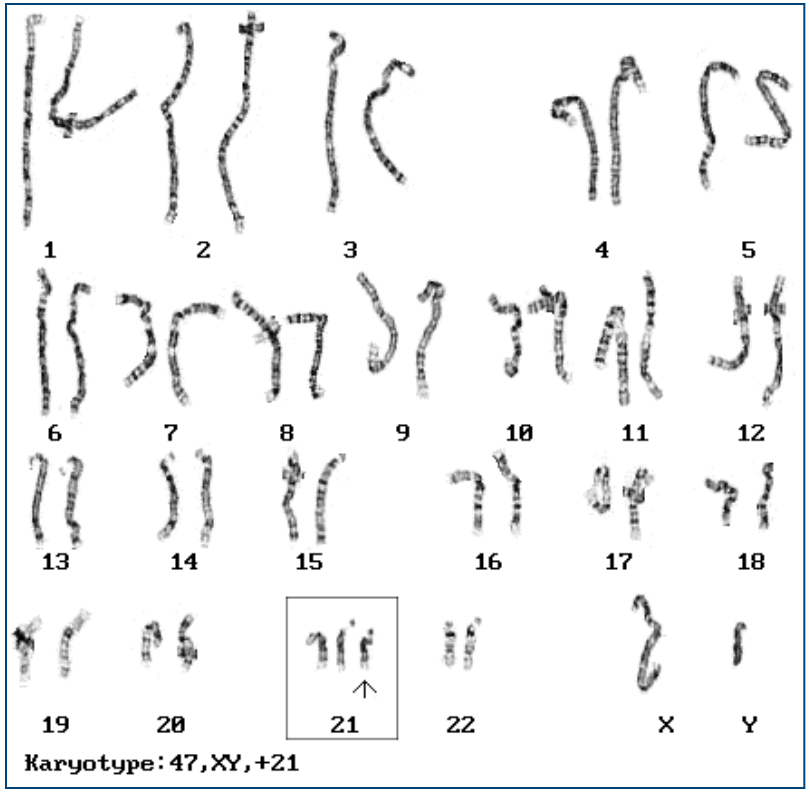


Male

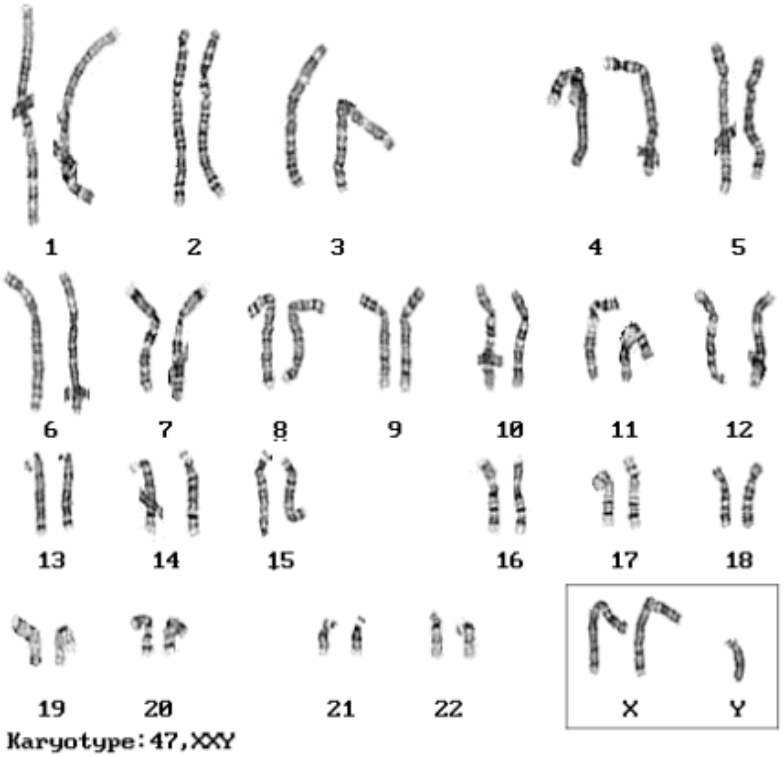


Female

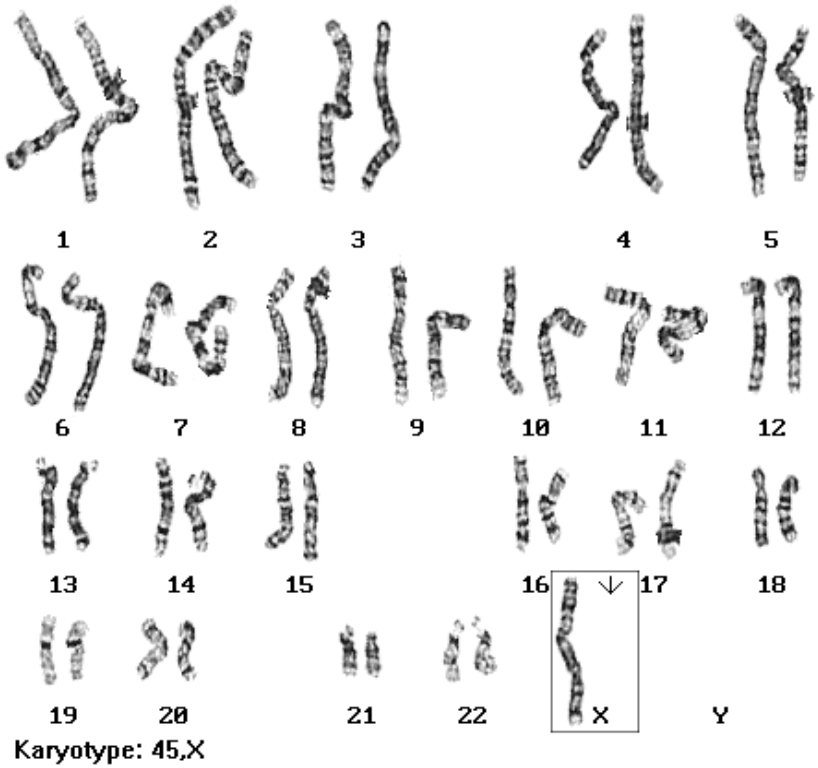
Downsyndrome (Trisomy 21)



Kleinfelter Syndrome XXY

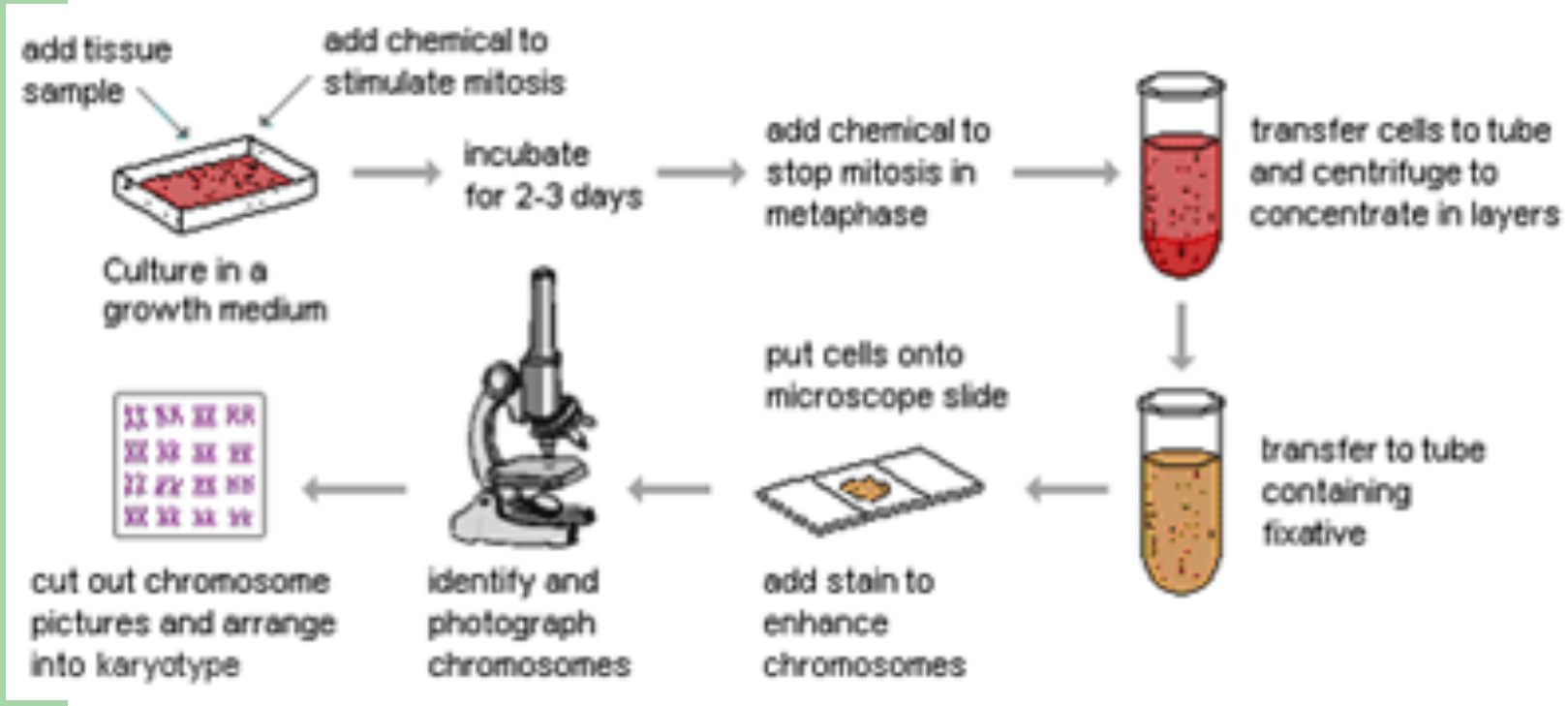


Turner Syndrome (X)



Karyotyping

- Stained chromosomes are matched with their homologous pairs
- The process allows doctors to visually see if there are extra or missing chromosomes in the fetus
- Down syndrome can be detected based on the extra chromosome on #21
- Other nondisjunction mutations are quite fatal, and the ones discussed are the few that can lead to a viable life.


add tissue sample
add chemical to stimulate mitosis
Culture in a growth medium
incubate for 2-3 days
add chemical to stop mitosis in metaphase
transfer cells to tube and centrifuge to concentrate in layers
transfer to tube containing fixative
put cells onto microscope slide
add stain to enhance chromosomes
identify and photograph chromosomes
cut out chromosome pictures and arrange into karyotype

Your task…

- Human Internet Karyotyping Assignment
- Submit end of class.