

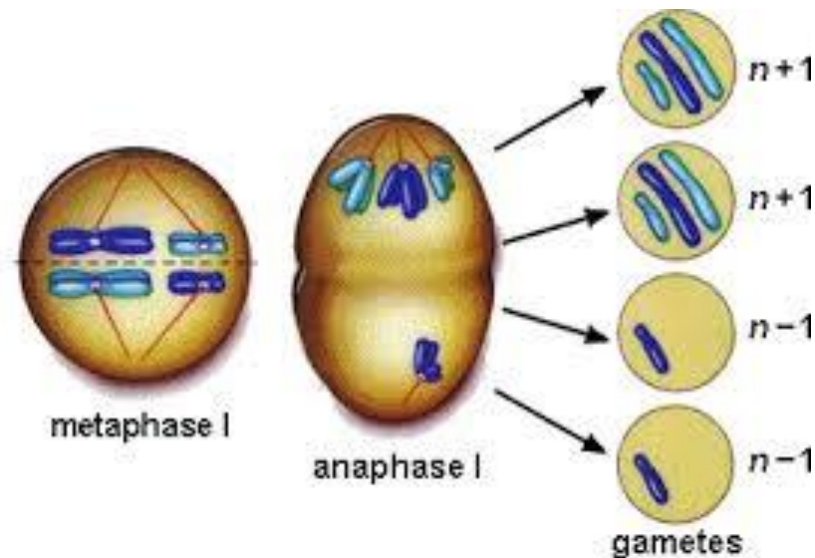
# Principles of Genetics (Zoo-352)

## Lecture 6

### Errors in Meiosis

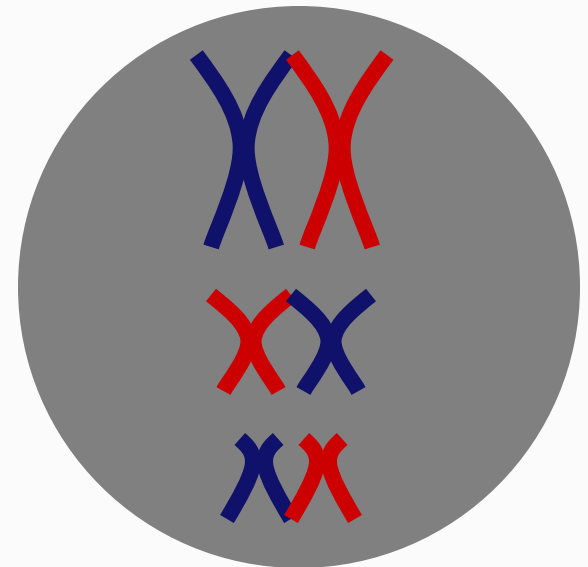
Department of Zoology, 1438-1439 H

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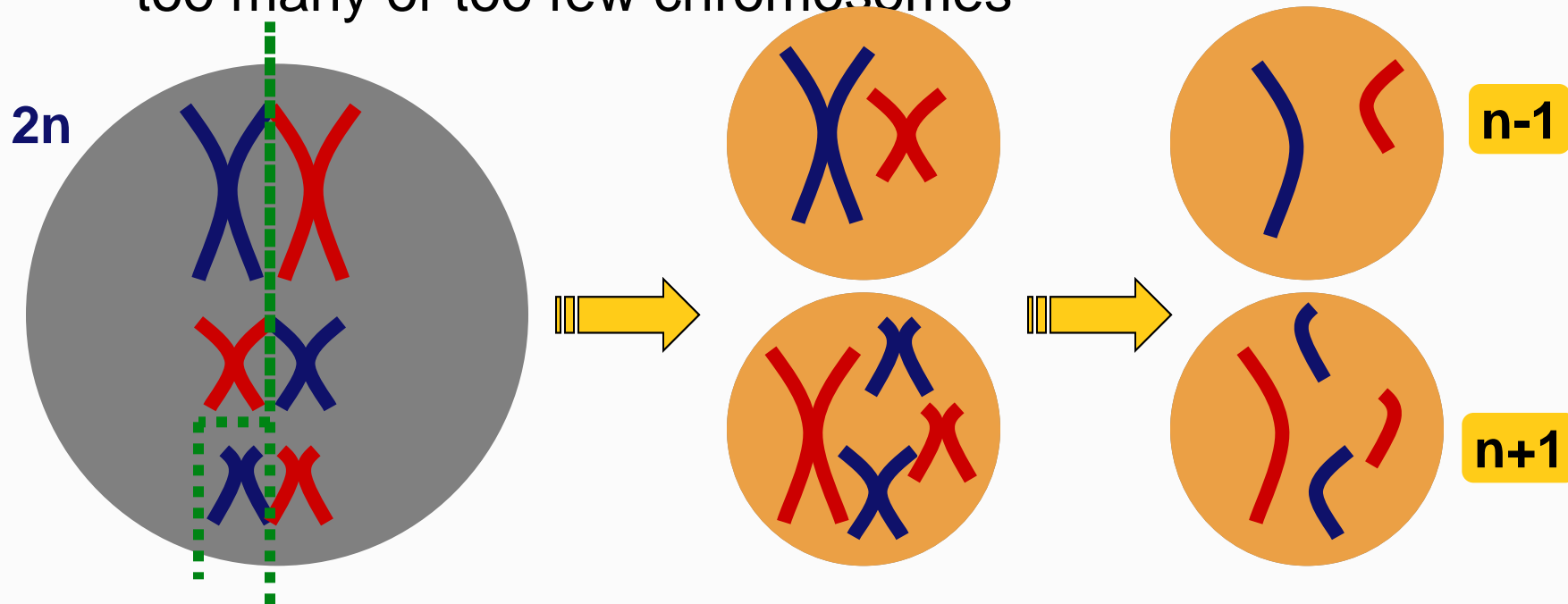
# Chromosomal abnormalities

- Incorrect number of chromosomes
  - nondisjunction
    - chromosomes don't separate properly during meiosis
  - breakage of chromosomes
    - deletion
    - duplication
    - inversion
    - translocation

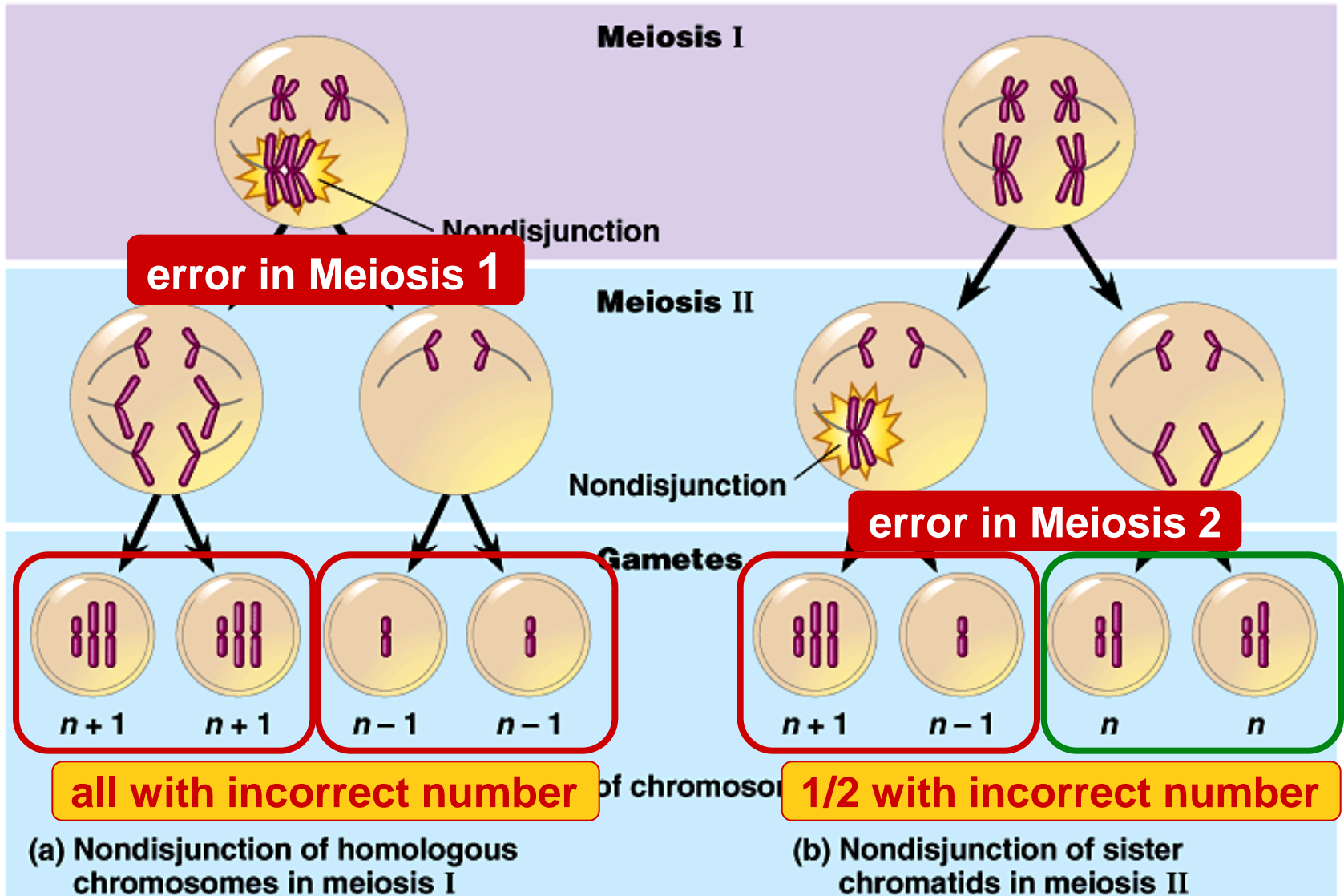


# Nondisjunction

- Problems with meiotic spindle cause errors in daughter cells
  - homologous chromosomes do not separate properly during Meiosis 1
  - sister chromatids fail to separate during Meiosis 2
  - too many or too few chromosomes

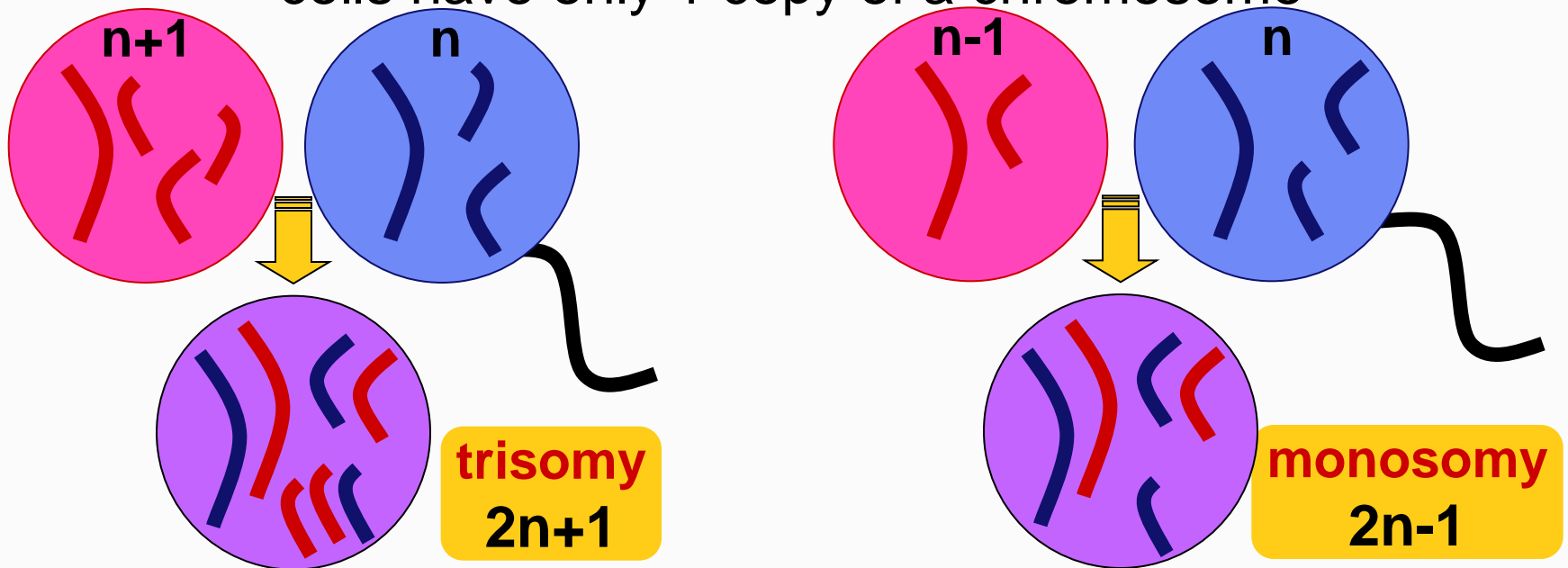


# Alteration of chromosome number



# Nondisjunction

- Baby has wrong chromosome number
  - trisomy
    - cells have 3 copies of a chromosome
  - monosomy
    - cells have only 1 copy of a chromosome

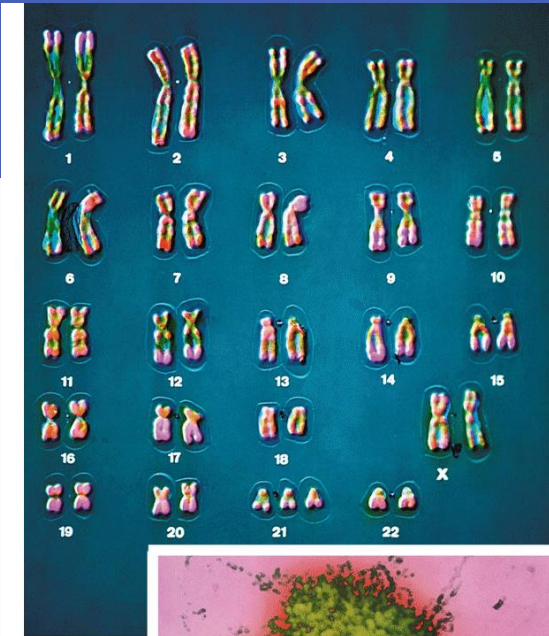


# Human chromosome disorders

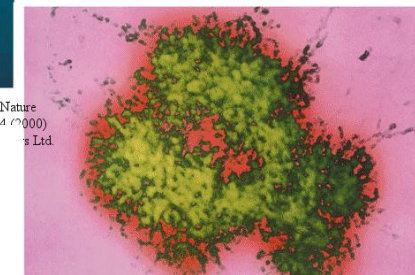
- High frequency in humans
    - most embryos are spontaneously aborted
    - alterations are too disastrous
    - developmental problems result from biochemical imbalance
      - imbalance in regulatory molecules?
        - hormones?
        - transcription factors?
  - Certain conditions are tolerated
    - upset the balance less = **survivable**
    - but characteristic set of symptoms = **syndrome**
-

# Down syndrome

- Trisomy 21
  - 3 copies of chromosome 21
  - 1 in 700 children born in U.S.
- Chromosome 21 is the smallest human chromosome
  - but still severe effects
- Frequency of Down syndrome correlates with the age of the mother



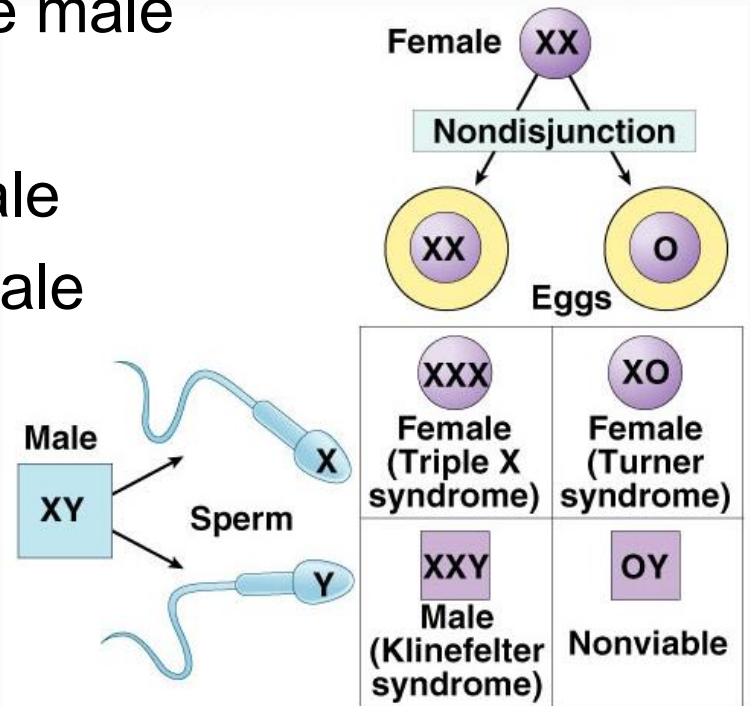
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# Sex chromosomes abnormalities

- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct syndromes in humans
  - **XXY** = Klinefelter's syndrome male
  - **XXX** = Trisomy X female
  - **XYY** = Jacob's syndrome male
  - **XO** = Turner syndrome female

**Know inheritance and characteristics of each!**

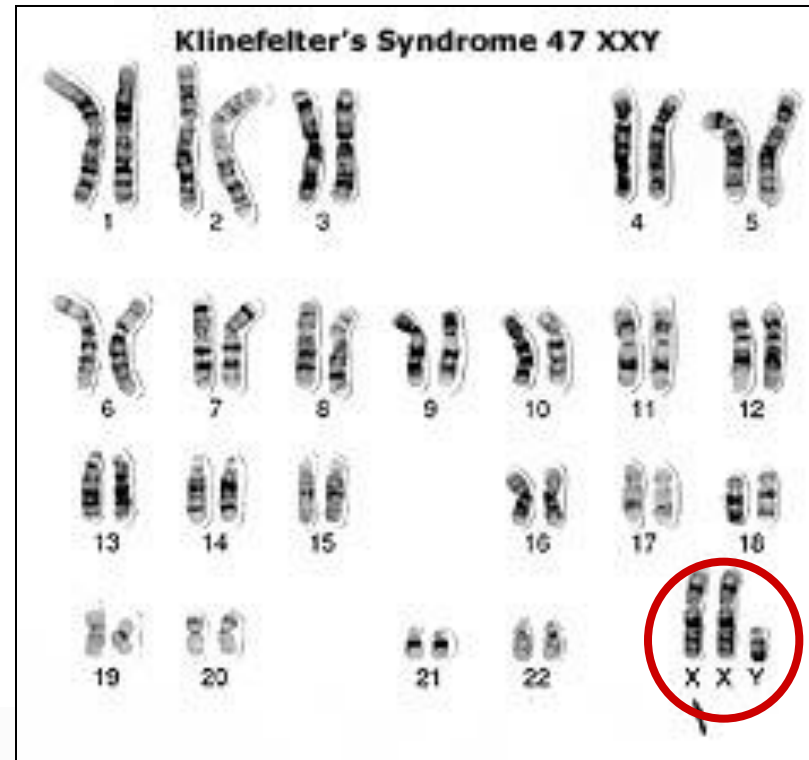




# Klinefelter's syndrome

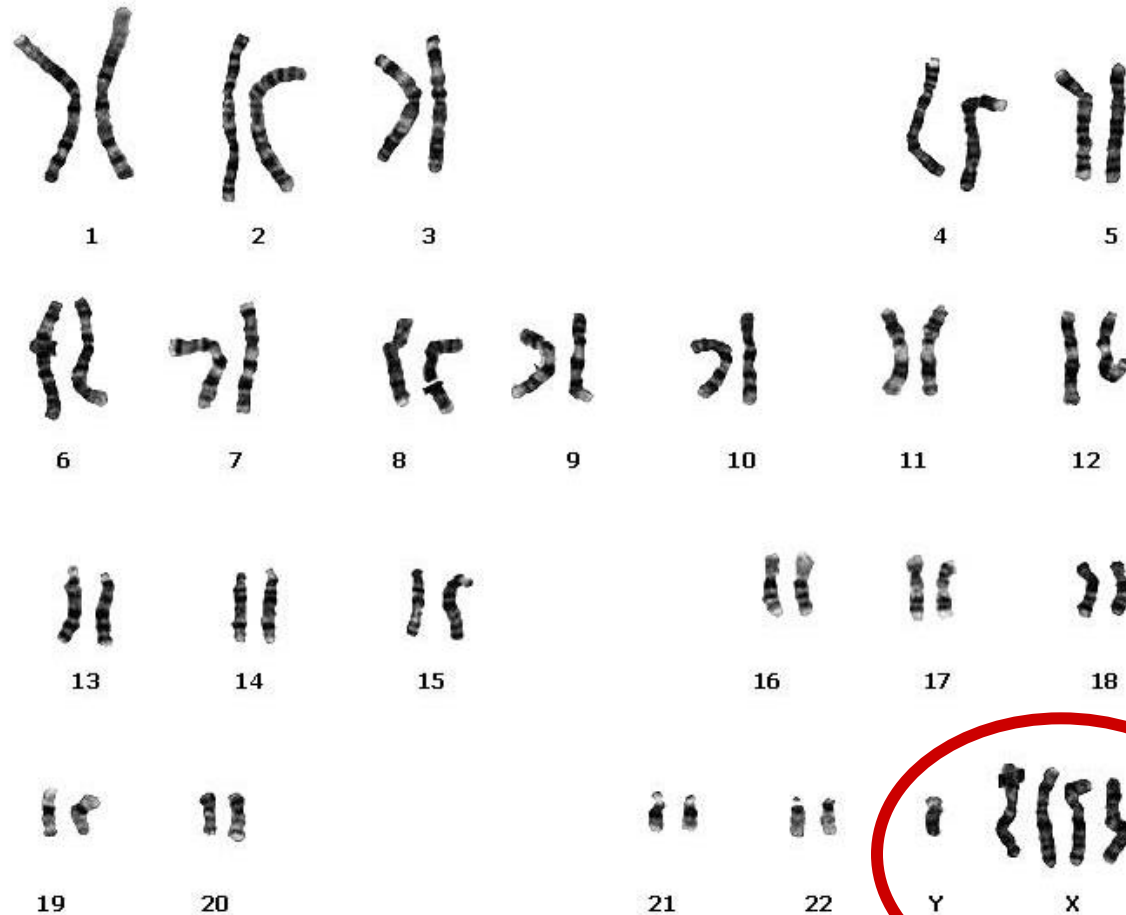
## ■ XXY male

- one in every 2000 live births
- have male sex organs, but are sterile
- feminine characteristics
  - some breast development
  - lack of facial hair
- tall
- normal intelligence



# Klinefelter's syndrome

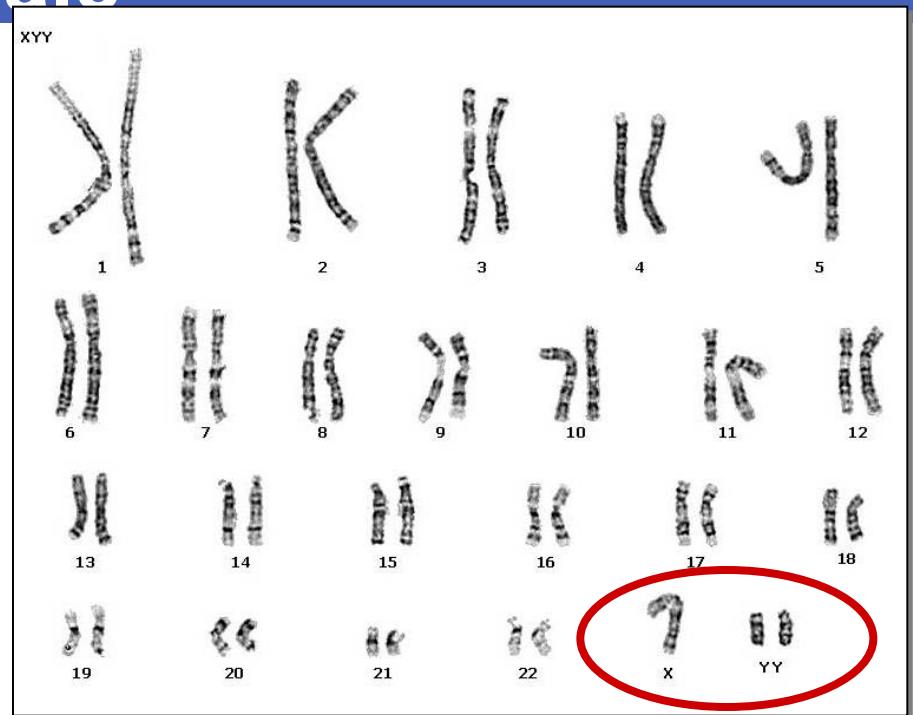
XXXXY, Klinefelter's Syndrome



# Jacob's syndrome male

## XYY Males

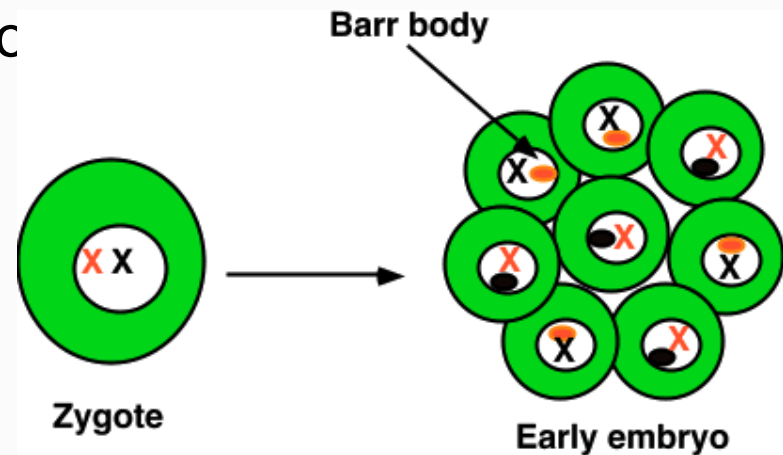
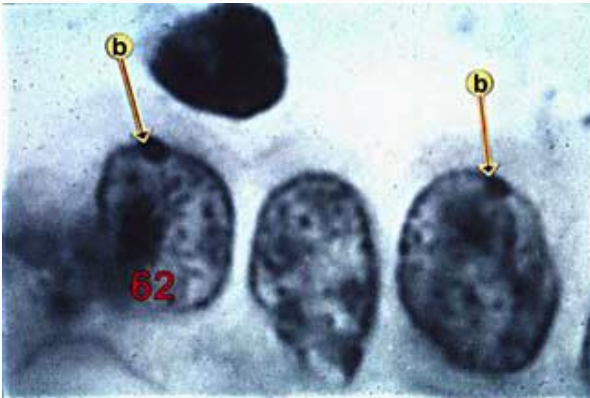
- 1 in 1000 live male births
- extra Y chromosome
- slightly taller than average
- more active
- normal intelligence, slight learning disabilities
- delayed emotional maturity
- normal sexual development



# Trisomy X

## ■ XXX

- 1 in every 2000 live births
- produces healthy females
  - Why?
  - **Barr bodies** [A **Barr Body** is an inactivated, condensed X chromosome found in female cells.]
    - all but one X chromoc



# Turner syndrome

- Monosomy X or X0
  - 1 in every 5000 births
  - varied degree of effects
  - webbed neck
  - short stature
  - sterile

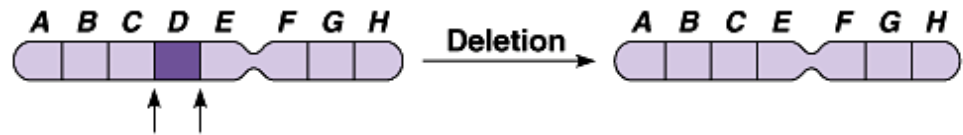


# Changes in chromosome structure

error of replication

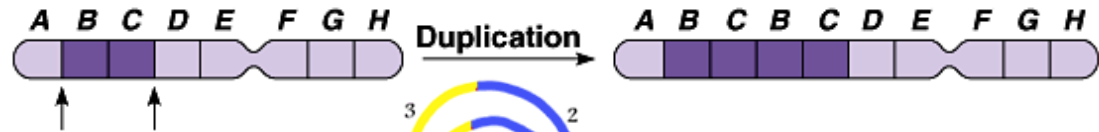
- deletion

- loss of a chromosomal segment



- duplication

- repeat a segment



error of crossing over

- inversion

- reverses a segment



- translocation

- move segment from one chromosome to another

