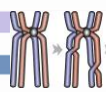
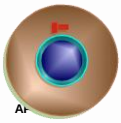
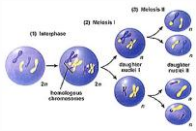


Errors of Meiosis
Chromosomal Abnormalities

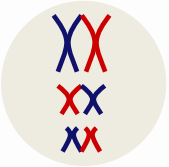






## Chromosomal abnormalities

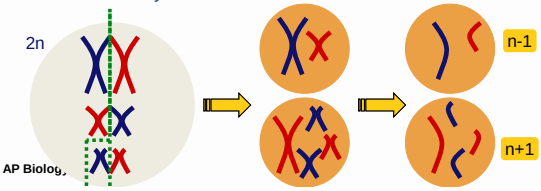
- Incorrect number of chromosomes
  - nondisjunction
    - chromosomes don't separate properly during meiosis
  - breakage of chromosomes
    - deletion
    - duplication
    - inversion
    - translocation





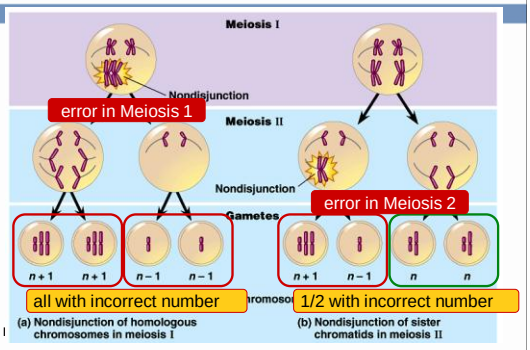
## Nondisjunction

- 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





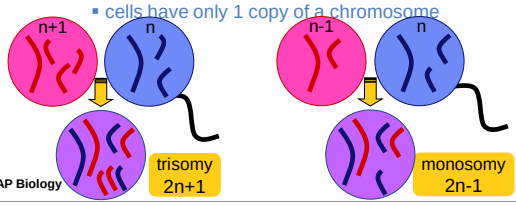

## Alteration of chromosome number






## Nondisjunction

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



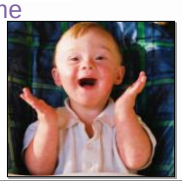



## Human chromosome disorders

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

## Down syndrome

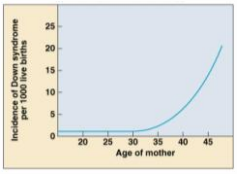
- Trisomy 21
  - 3 copies of chromosome 21
  - 1 in 700 children born in U.S.
- Chromosome 21 is the smallest human chromosome
  - but still severe effects
- Frequency of Down syndrome correlates with the age of the mother





## Down syndrome & age of mother

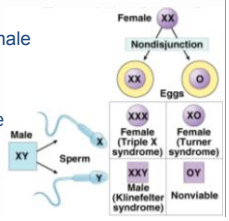
| Mother's age | Incidence of Down Syndrome |
|---|---|
| Under 30 | <1 in 1000 |
| 30 | 1 in 900 |
| 35 | 1 in 400 |
| 36 | 1 in 300 |
| 37 | 1 in 230 |
| 38 | 1 in 180 |
| 39 | 1 in 135 |
| 40 | 1 in 105 |
| 42 | 1 in 60 |
| 44 | 1 in 35 |
| 46 | 1 in 20 |
| 48 | 1 in 16 |
| 49 | 1 in 12 |






## Sex chromosomes abnormalities

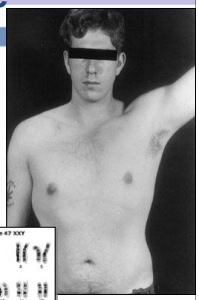
- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct syndromes in humans
  - XXY = Klinefelter's syndrome male
  - XXX = Trisomy X female
  - XYY = Jacob's syndrome male
  - XO = Turner syndrome female

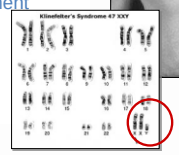





## Klinefelter's syndrome

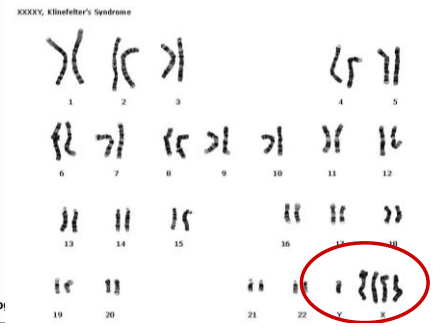
- XXY male
  - one in every 2000 live births
  - have male sex organs, but are sterile
  - feminine characteristics
    - some breast development
    - lack of facial hair
  - tall
  - normal intelligence







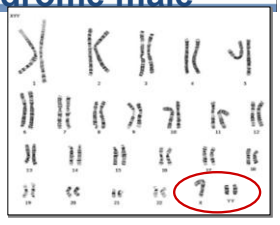
## Klinefelter's syndrome





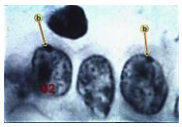

## Jacob's syndrome male

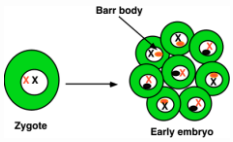
- XYY Males
  - 1 in 1000 live male births
  - extra Y chromosome
  - slightly taller than average
  - more active
  - normal intelligence, slight learning disabilities
  - delayed emotional maturity
  - normal sexual development



## Trisomy X

- XXX
  - 1 in every 2000 live births
  - produces healthy females
    - Why?
    - Barr bodies
      - all but one X chromosome is inactivated

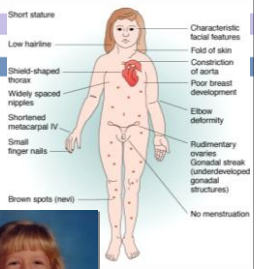







## Turner syndrome

- 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