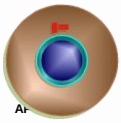
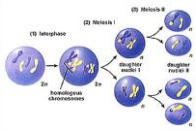
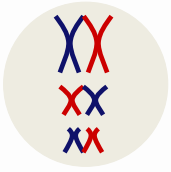
Errors of Meiosis
Chromosomal Abnormalities





Chromosomal abnormalities

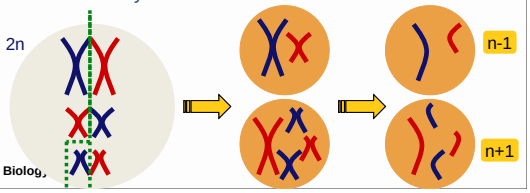
- Incorrect number of chromosomes
  - nondisjunction
    - chromosomes don't separate properly during meiosis
  - breakage of chromosomes
    - deletion
    - duplication
    - inversion
    - translocation



AP Biology

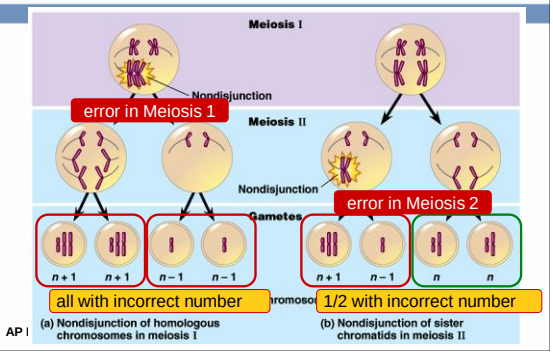
Nondisjunction

- 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



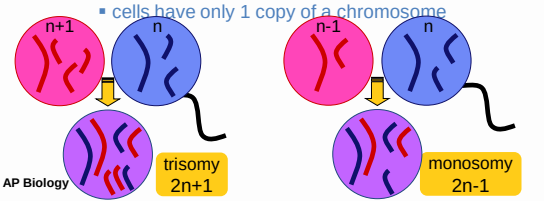
AP Biology

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

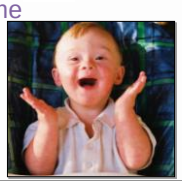


Human chromosome disorders

- 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

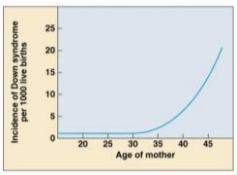
Down syndrome

- 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



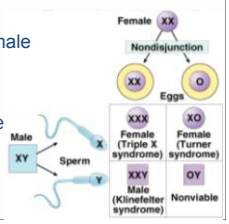
Down syndrome & age of mother

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



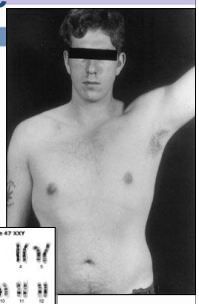
Sex chromosomes abnormalities

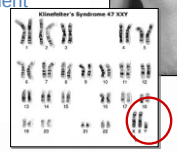
- Human development more tolerant of wrong numbers in sex chromosome
- But produces a variety of distinct syndromes in humans
  - XXY = Klinefelter's syndrome male
  - XXX = Trisomy X female
  - XYY = Jacob's syndrome male
  - XO = Turner syndrome female



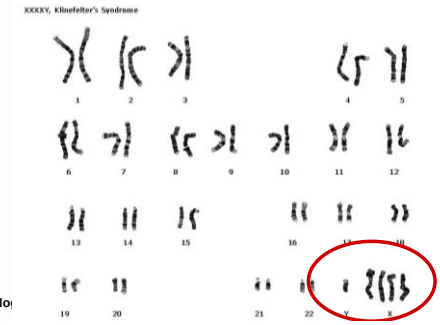
Klinefelter's syndrome

- 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



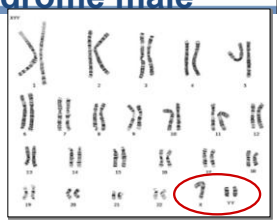


Klinefelter's syndrome



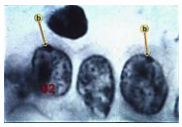
Jacob's syndrome male

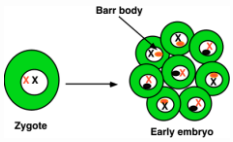
- XYY Males
  - 1 in 1000 live male births
  - extra Y chromosome
  - slightly taller than average
  - more active
  - normal intelligence, slight learning disabilities
  - delayed emotional maturity
  - normal sexual development



Trisomy X

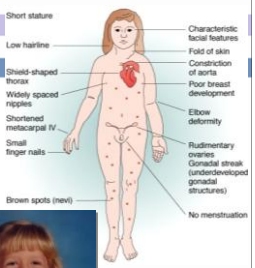
- XXX
  - 1 in every 2000 live births
  - produces healthy females
    - Why?
    - Barr bodies
      - all but one X chromosome is inactivated





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