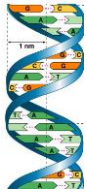
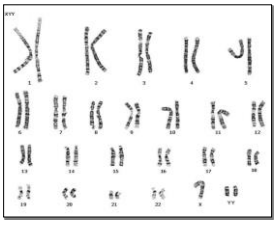
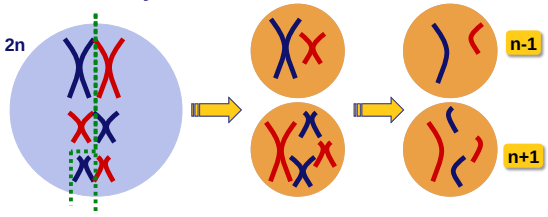
Chromosomal Abnormalities in Humans



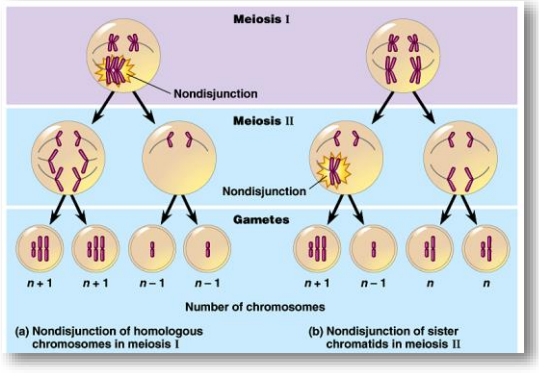


Nondisjunction

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

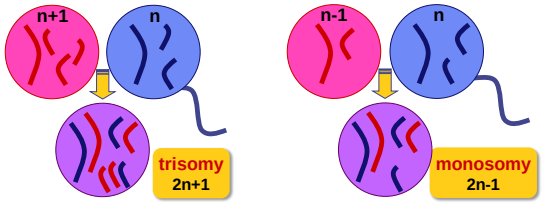


Alteration of Chromosome Number



Nondisjunction

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

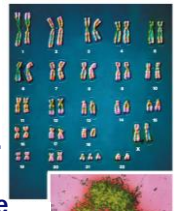


Human Chromosome Disorders

- 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 = survive
  - characteristic set of symptoms = syndrome

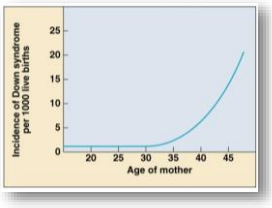
Down Syndrome

- 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 if affected
- Frequency of Down syndrome correlates with the age of the mother



Down Syndrome & Mother's Age

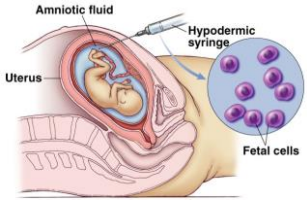
Mother's age	Incidence of Down Syndrome
Under 30	<1 in 1000
30	1 in 900
35	1 in 400
36	1 in 300
37	1 in 230
38	1 in 180
39	1 in 135
40	1 in 105
42	1 in 60
44	1 in 35
46	1 in 20
48	1 in 16
49	1 in 12



Genetic Screening

Genetic Testing

- Amniocentesis in 2nd trimester
  - sample of embryo cells
  - stain & photograph chromosomes
- Analysis of karyotype



Rate of miscarriage due to amniocentesis:
- 1970s data
  0.5%, or 1 in 200 pregnancies
- 2006 data
  <0.1%, or 1 in 1600 pregnancies

Down Syndrome & Mother's Age

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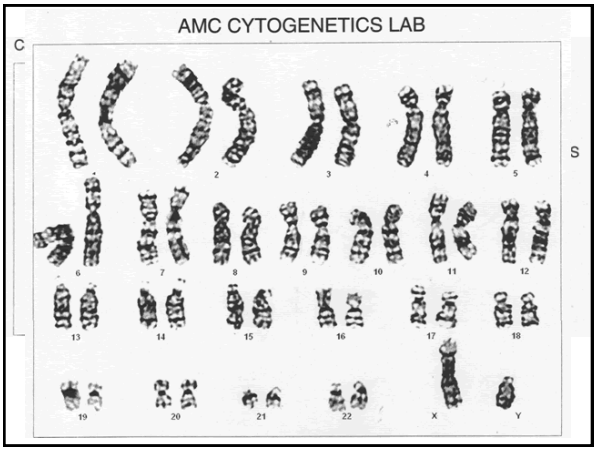
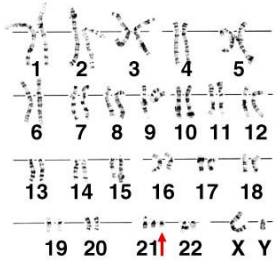
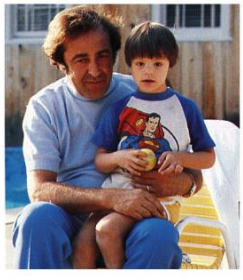
Incidence of Down syndrome per 1000 live births
25
20
15
10
5
0
20 25 30 35 40 45
Age of mother

Genetic Screening



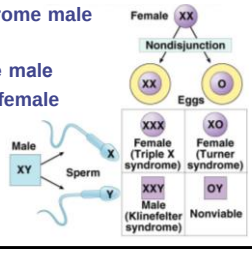
Trisomy 21





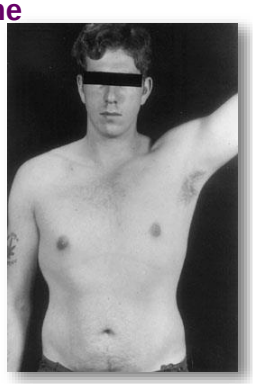
Human Sex Chromosomes

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

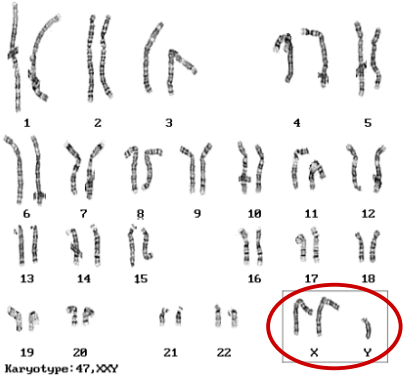


Klinefelter's Syndrome

- XXY male
  - one in every 2000 live births
  - have male sex organs, but are sterile
  - slight feminine characteristics
  - tall
  - normal intelligence



Klinefelter's Syndrome

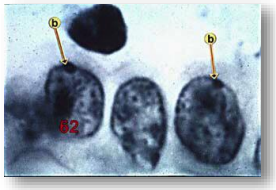


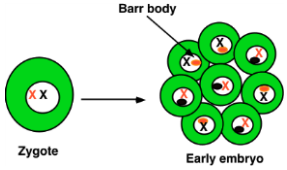
Jacob's Syndrome

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

Trisomy X

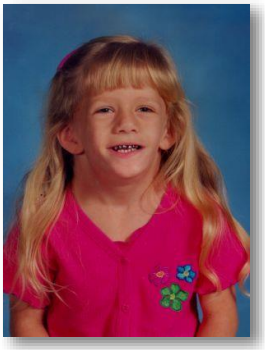
- XXX
  - 1 in every 2000 live births
  - produces healthy females
    - Why?





Turner Syndrome

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



Turner Syndrome