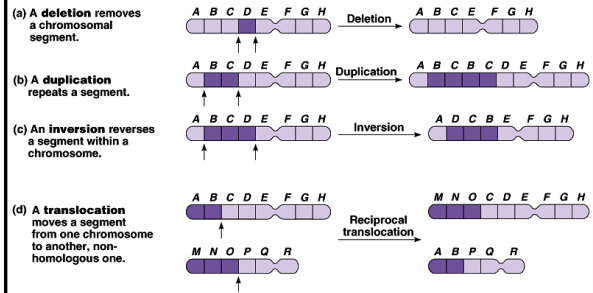


Chromosomal Abnormalities

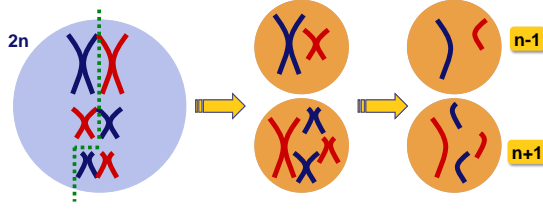
- **Incorrect number of chromosomes**
 - ◆ **nondisjunction**
 - chromosomes don't separate properly during meiosis
 - ◆ **breakage of chromosomes**
 - deletion
 - duplication
 - inversion
 - translocation

Changes in Chromosome Structure

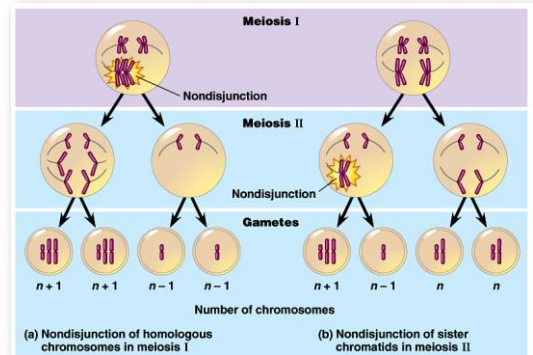


Nondisjunction

- **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

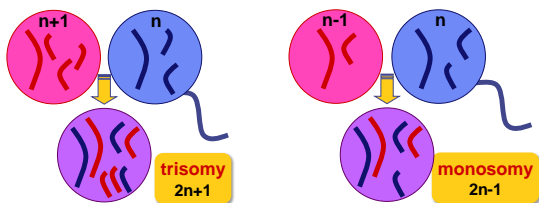


Alteration of Chromosome Number



Nondisjunction

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

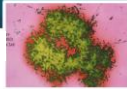


Human Chromosome Disorders

- **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**

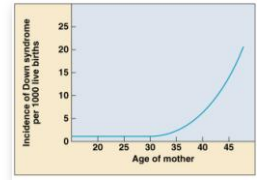
Down Syndrome

- **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

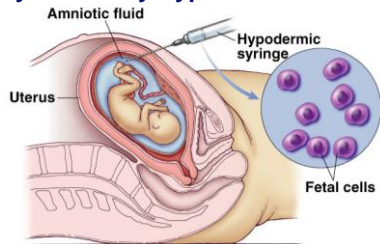


Rate of miscarriage due to amniocentesis:

- **1970s data**
0.5%, or 1 in 200 pregnancies
- **2006 data**
<0.1%, or 1 in 1600 pregnancies

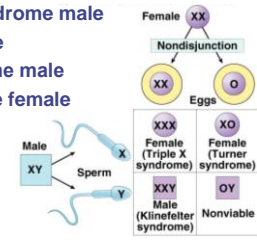
Genetic Testing

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



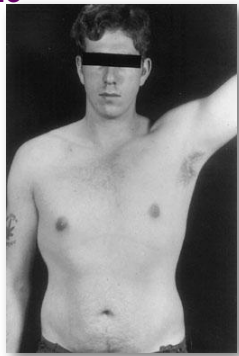
Human Sex Chromosomes

- **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

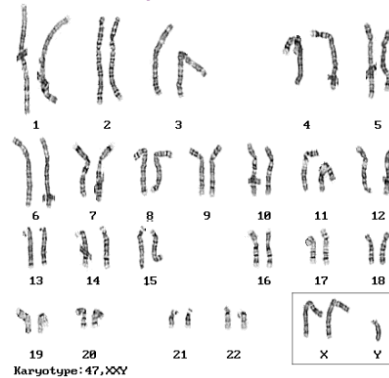


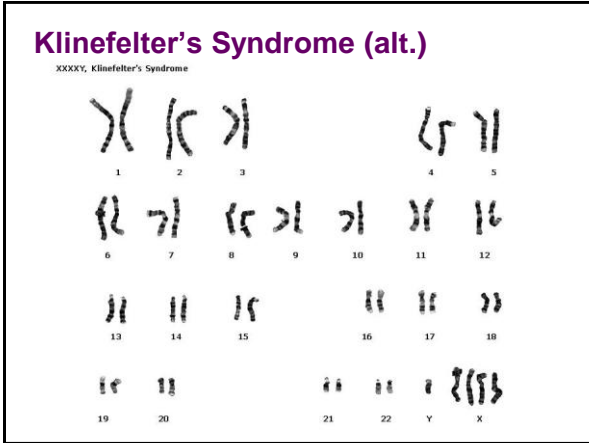
Klinefelter's Syndrome

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



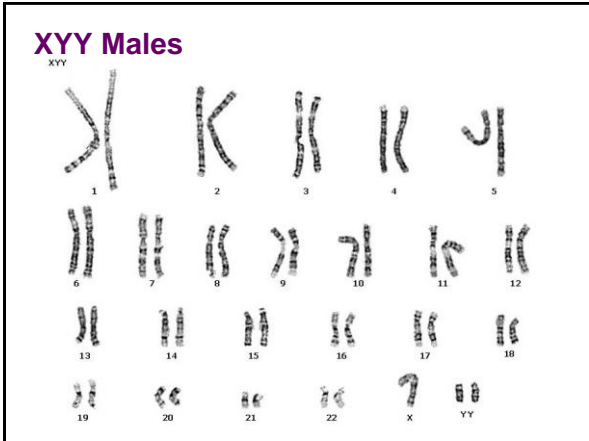
Klinefelter's Syndrome





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