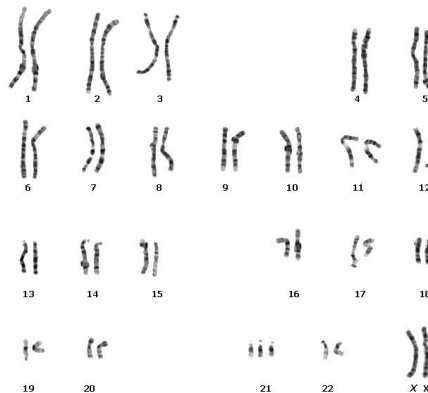


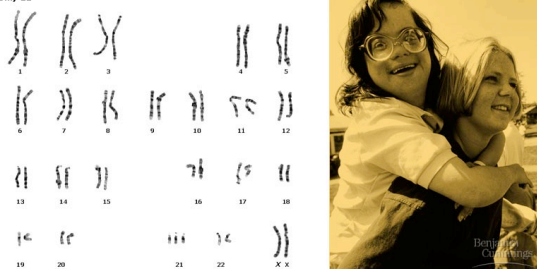
Human Genetic Diseases

- Chromosomal disorders cause some genetic diseases
- Aneuploidy - having the wrong number of chromosomes

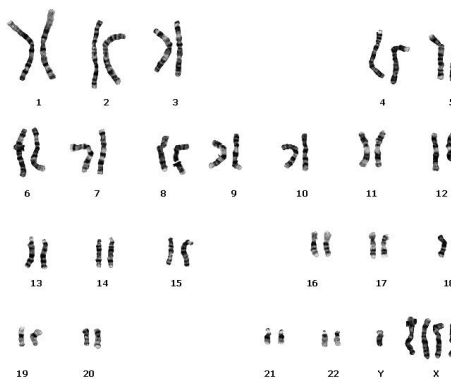
Trisomy 21



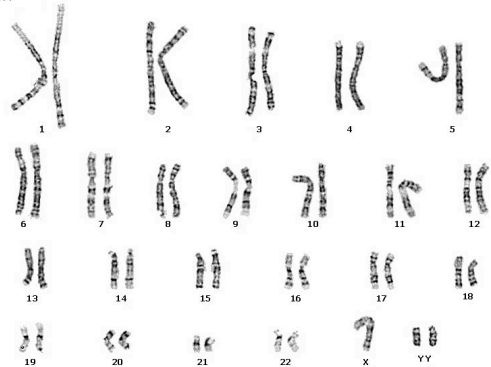
Trisomy 21



XXXXY, Klinefelter's Syndrome



XYY



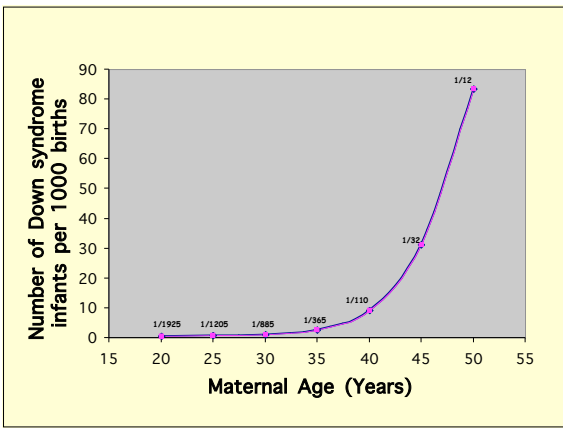
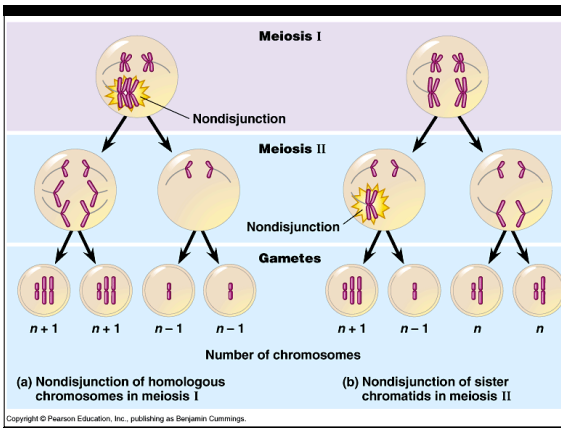
Human Genetic Diseases

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- Aneuploidy - having the wrong number of chromosomes
- Effects of aneuploidies - table

TREASONY FOR	# among spontaneous abortions	# among live births
1	0	0
2	159	0
3	53	0
4	95	0
5	0	0
6-12	561	0
13	128	17
14	275	0
15	318	0
16	1229	0
17	10	0
18	223	13
19-20	52	0
21	350	113
22	424	0
SEX CHROMOSOMES		
XYY	4	46
XXY	4	44
XO	1350	8
XXX	21	44
POLYPLOIDY		
Trisomy	1275	0
Tetraploidy	450	0

Human Genetic Diseases

- Chromosomal disorders cause some genetic diseases
- Aneuploidy - having the wrong number of chromosomes
- Effects of aneuploidies - table
- Causes of aneuploidies - homologous chromosomes fail to separate during the first meiotic division



Two Hypotheses Might Explain This Pattern

- "Old Eggs" hypothesis
- "Relaxed Filter" hypothesis

Which one is right? What kind of evidence would help you decide?

Genetic testing

- During pregnancy
 - Amniocentesis
 - Chorionic villus sampling
- Of adults
 - For recessive traits (e.g., sickle cell)
 - For dominant traits or other genes (e.g., Huntington's disease, BRCA1)

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