

Human Genetic Diseases I

- Most genetic diseases are caused by recessive alleles on autosomes
- Harmful recessive alleles can persist if they are not too common, despite natural selection, because homozygous recessive individuals will be rare
- Examples
 - Phenylketonuria or PKU
 - Cystic fibrosis - 4% of whites are carriers
 - Sickle-cell anemia - 10% of African-Americans are carriers

Probability and Human Genetics

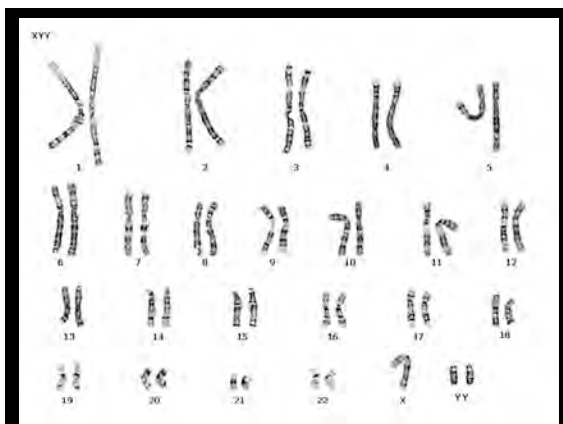
- If one in 25 whites carries the defective allele that causes CF, then what is the chance that a given white child will have CF?
- If the defective allele that causes red-green colorblindness is on one out of every 100 human X chromosomes, what proportion of men will be colorblind? What proportion of women will be colorblind?

Human Genetic Diseases II

- Some rare genetic diseases are caused by dominant alleles - e.g., Huntington's Disease
- Others (e.g., hemophilia) caused by sex-linked genes

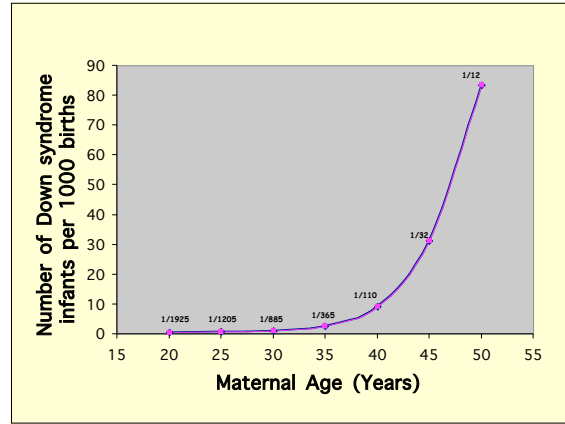
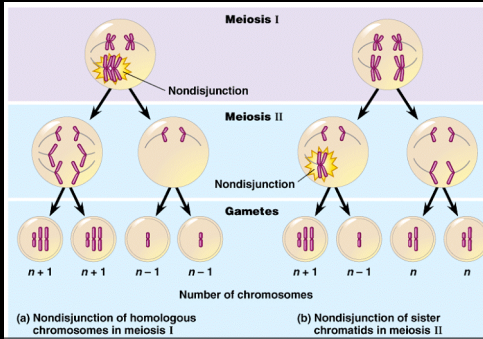
Human Genetic Diseases III

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



TRISOMY 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		
XXY	4	46
XXY	4	44
XO	1350	8
XXX	21	44
POLYPOIDY		
Triplody	1275	0
Tetraploidy	490	0

Causes of aneuploidy: nondisjunction of chromosomes in meiosis



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

Genetic testing of adults

For recessive traits (e.g., sickle cell)

For dominant traits or other genes (e.g., Huntington's disease, BRCA1)