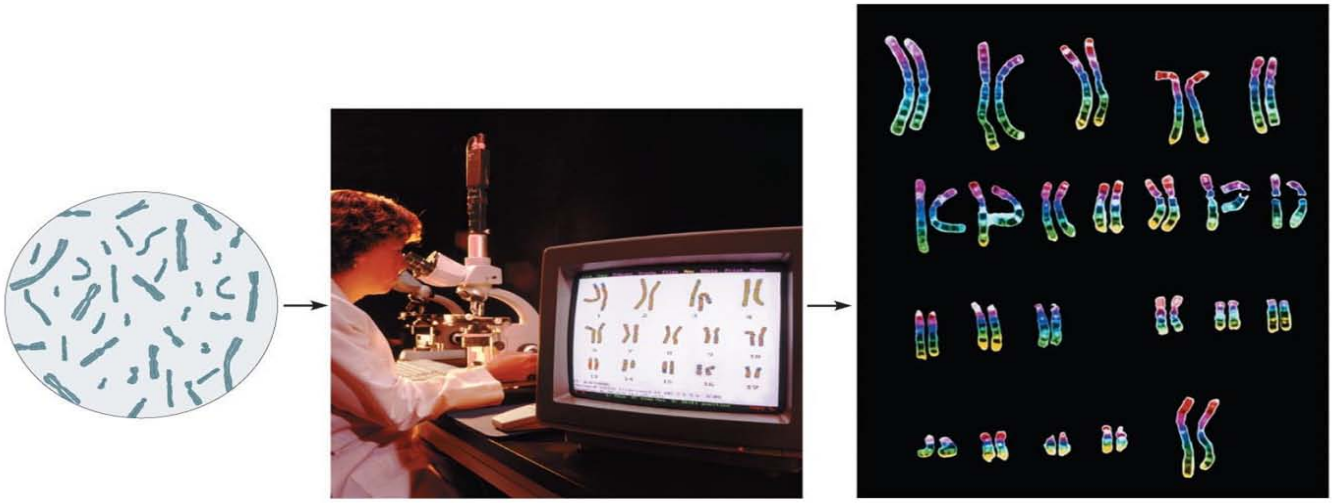
Heredity



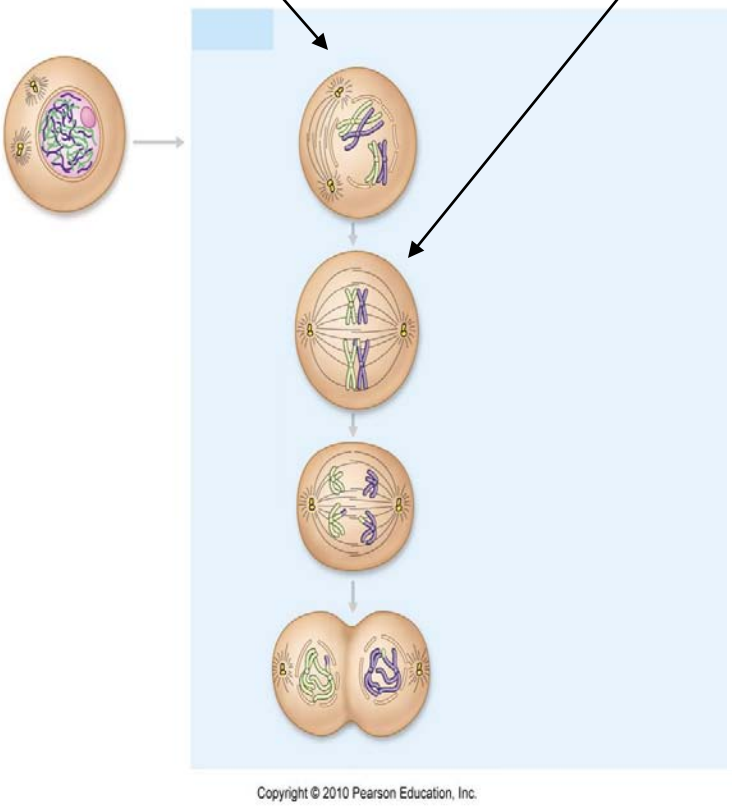
Sexual Reproduction

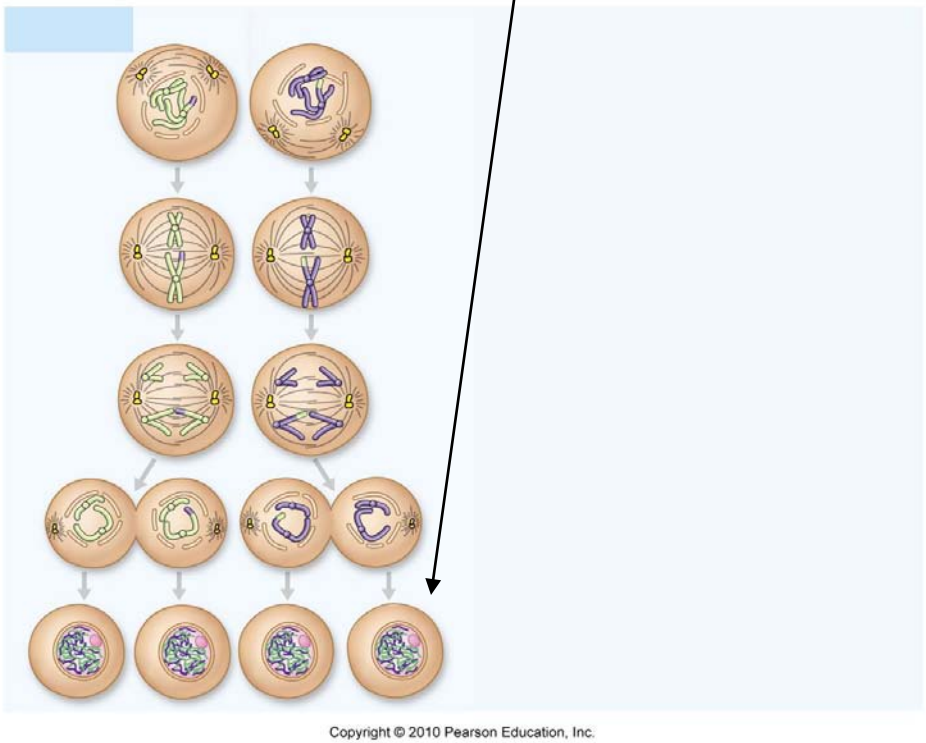
- unique organism (unique genotype & phenotype)
sexual reproduction
-> genetic variation between individuals

genetic variation factors:
1) crossover
- allele exchanges between homologous chrom.
2) independent assortment
- genetic variation in gametes
3) random sperm fertilization
- hundreds of millions of sperm per ejaculation, only 1-2 will fertilize the egg

Genetic Variation

crossover **independent assortment** **random sperm fertilization**





Pioneers of Heredity

1) **Darwin – how are organisms changed?**
   - **genetic var. -> bio-diversity**
   - **natural selection & biodiversity -> evolution**
   - **bird studies**
2) **Mendel - how are changes passed?**
   - **deduced existence of genes**
   - **segregation, independent assortment**
   - **garden pea experiments**
3) **Watson & Crick - how are changes saved?**
   - **DNA molecular structure**

Darwin – how are organisms changed?

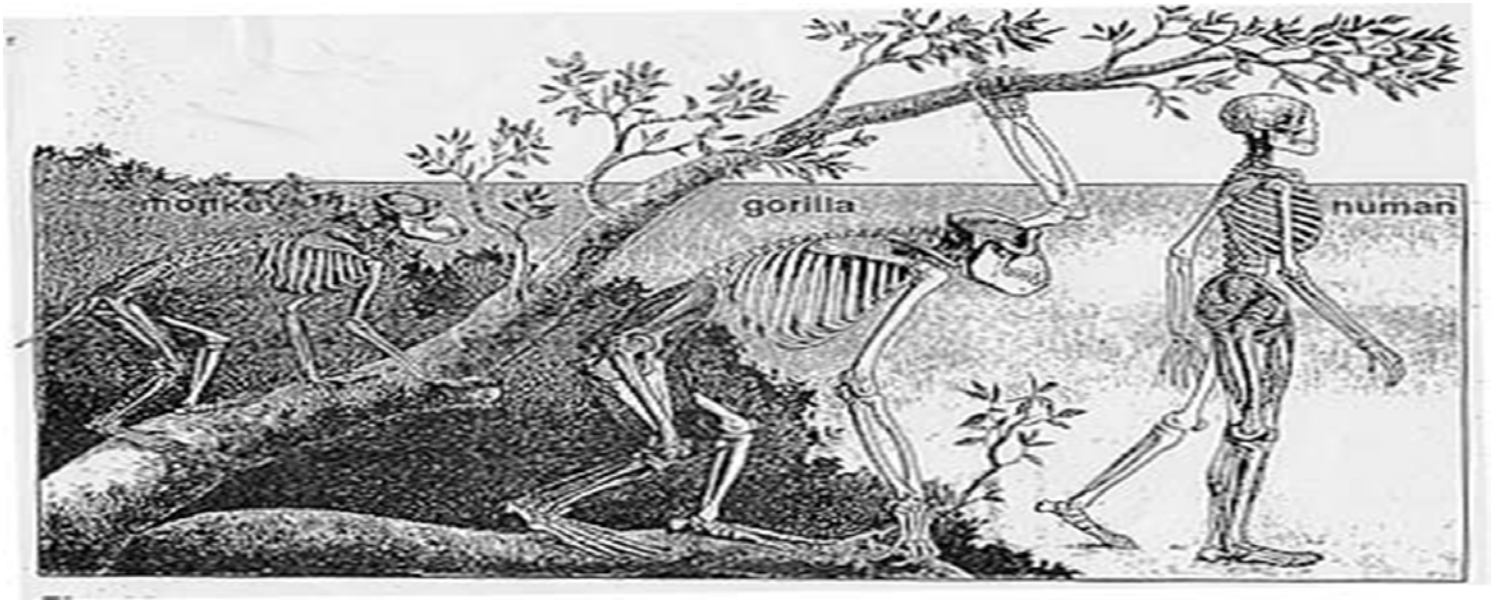
<u>Theory of evolution</u> (1859)

- descent with modif. in successive generations

<u>Natural selection</u>

- environ. change ->survival of fittest

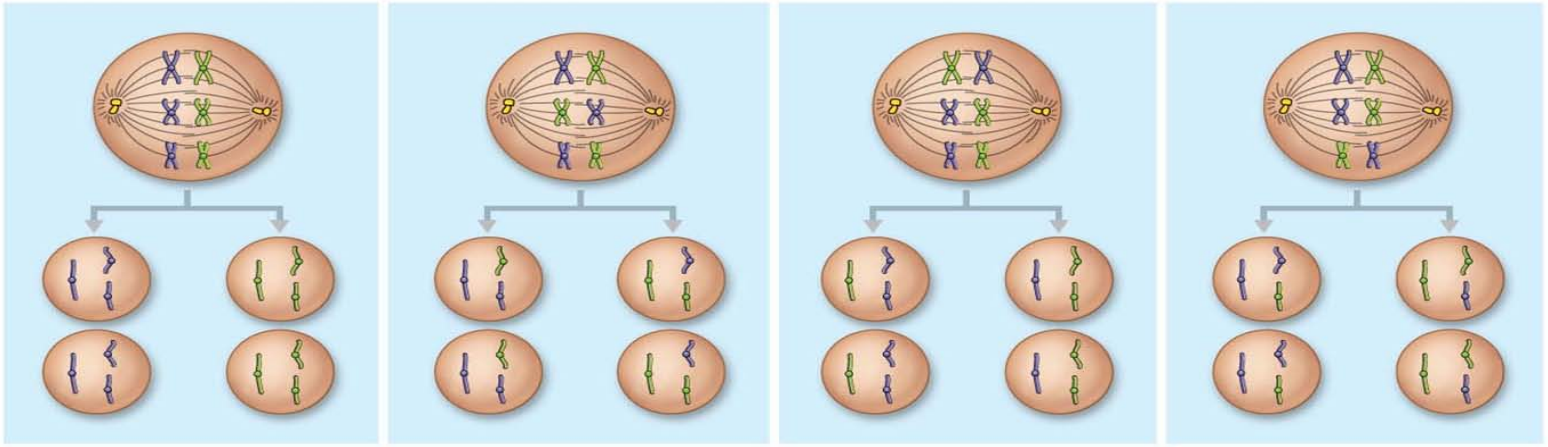
(from the bio-diversity prod. thru genetic variation)



Mendel - how are changes passed?

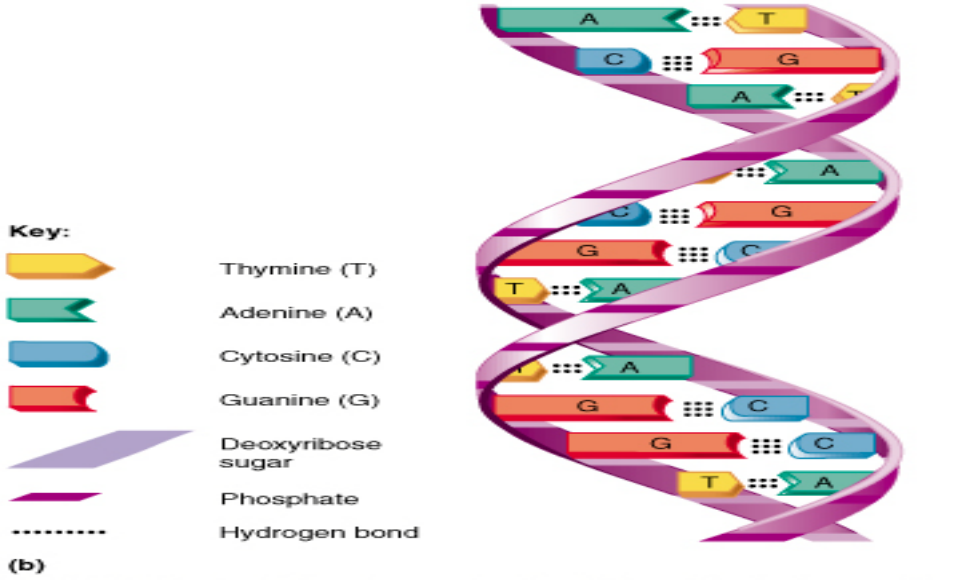
1) **segregation - chromosomes separated**
2) **independent assortment – chromo. indep. distributed**

16 possibilities



Watson & Crick - how are changes saved?

molecular structure of DNA
gene = trait, specific section & locus of chromosome
human genome project: about 25,000 genes (only)



Dominant / Recessive Heredity

- dominant alleles:
eg brown eye color, tongue rolling, widow's peak, non-albino, unattached earlobes, etc.

Trait	Phenotype	Genotype
1) brown eyes	E	EE (homozygous dom.)
2) brown eyes	E	Ee (heterozygous)
3) blue eyes	e	ee (homozygous rec.)

Incomplete or Co-Dominance Heredity

sickle cell anemia (sickling gene)
- amino acid substitution in HB -> sickle-shape RBC
benefit: malarial protection; ↑ levels in africans

Ss: sickle cell anemia trait
- make normal and sickled Hb
1) healthy unless in prolonged low oxygen air
2) can transmit sickling gene to offspring

ss: sickle cell anemia disease
- make only sickled Hb
- OK until breathing problems or excess exercise
- sickle RBC's jam & fragment in capillaries
-> breathing problems & pain

Multiple-Allele Heredity

3 alleles: I^A, I^B, i

4 phenotypes (blood groups)

A & B are co-dominant

TABLE 29.2 ABO Blood Groups

BLOOD GROUP (PHENOTYPE)	FREQUENCY (% OF U.S. POPULATION)			
	GENOTYPE	WHITE	BLACK	ASIAN
O	*ii*	45	49	40
A	I^AI^A or I^Ai	40	27	28
B	I^BI^B or I^Bi	11	20	27
AB	I^AI^B	4	4	5

Sex-Linked Heredity

<u>X-linked</u> (X chrom. - 2500 genes)

gene on X chrom.,

“unmasked” in males

eg hemophilia,

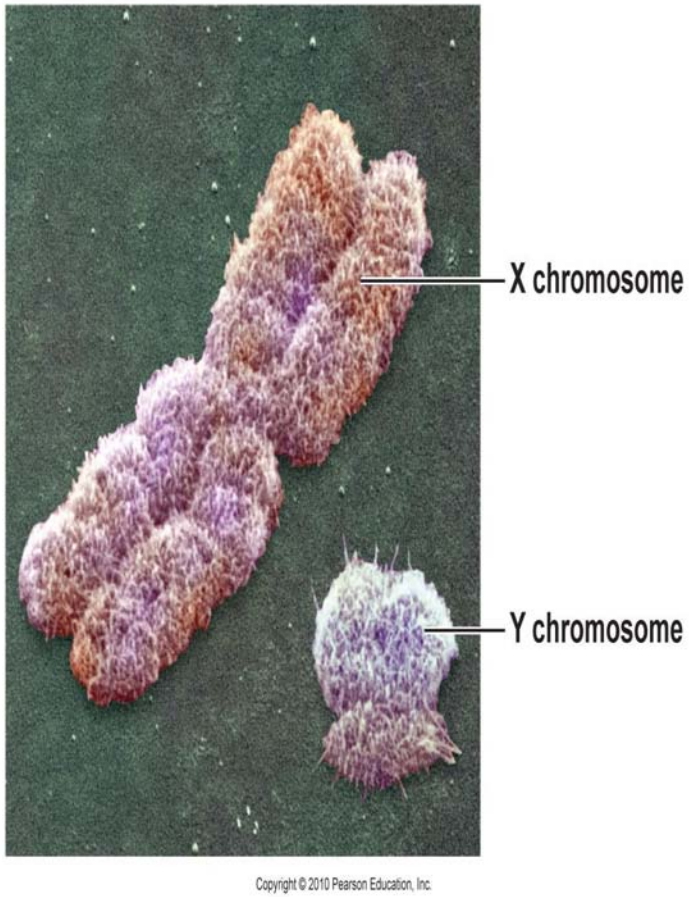
red-green color blindness

<u>Y-linked (</u>Y chrom. - 15 genes)

gene on Y chrom,

only males, father to son

eg SRY (male dev.), hairy ear pinnae



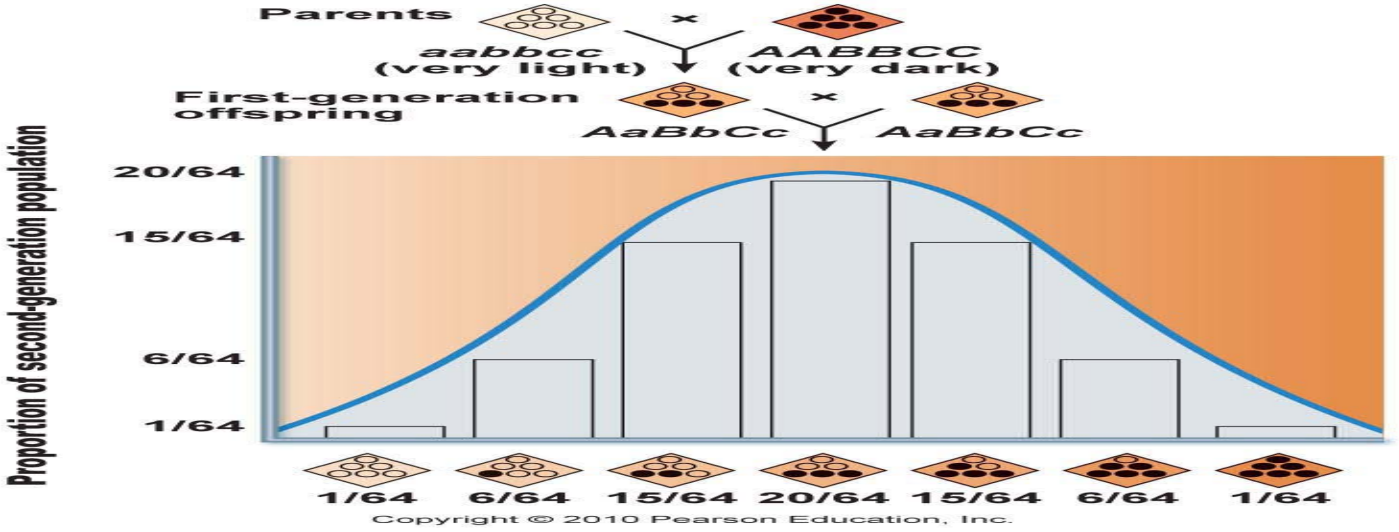
Polygene Heredity

skin color, height, metabolic rate, intelligence

- phenotype expressed by several gene pairs,
at different sites, acting in tandem

- results in graded difference between 2 extremes

3 gene pairs: skin color spectrum from light to dark



Terms Assign. (not collected)

1) chromosome
2) homologous chromosome
3) allele
4) trait
5) dominant trait
6) recessive trait
7) co-dominant trait
8) gene
9) gene locus
10) homozygous genes
11) heterozygous genes
12) phenotype
13) genotype

Maternal Contributions

phenocopies = environmentally produced phenotypes

- like genetic mutations

1) thalidomide (sedative)

-> flipperlike appendages

2) low childhood hormones (TH)

-> abnormal skeletal proportions (cretinism/ dwarfism)

3) poor nutrition & infections

-> stunts "tall genes", brain & body development

"RNA-only Genes"

“junk genes” = non protein coding DNA

- small RNA molecules that act directly on DNA, RNA, and/or protein synthesis

3 examples:

1) **anti-sense RNA: prevent protein synthesis**
2) **micro-RNA: silence mRNA’s**
3) **ribo-switches: start/stop protein synthesis**

research area:

- gene therapy in Parkinson’s, cancer, genetic disorders

Genomic imprinting

certain genes are tagged (imprinted) in gametogenesis

- tagged genes are expressed; untagged genes are silent

- same allele has different effect, depends on source parent

<u>Chromosome Pair #15</u>

Angelman Syndrome (mom)

- retarded, incoherent speech, jerky movement, uncontrolled laughter

Prader-Willi Syndrome (dad)

- less retardation, short, grossly obese

Maternal DNA

- both parents: nuclear DNA

mother: yolk, cytoplasm, organelles, maternal DNA

maternal DNA:

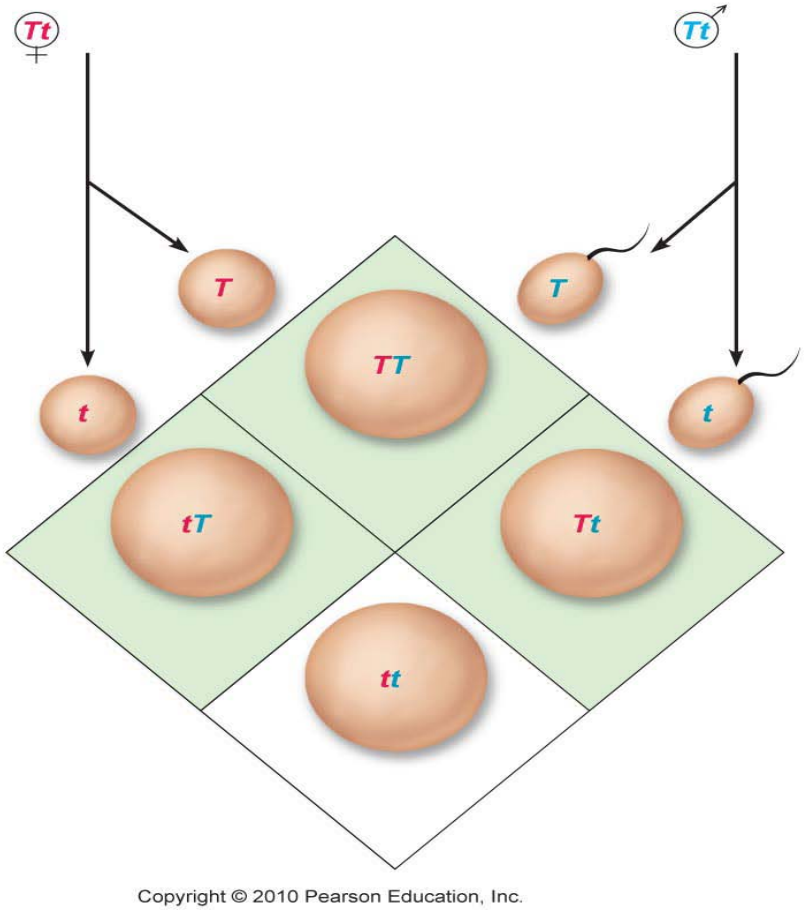
1) mitochondrial DNA (mtDNA)

- identical fragment in all human mitochondria (basis for "Eve", placental studies)

- defects may involve muscle & neural disorders

2) centriole DNA

Punnet Square (1)



Prediction of traits:

1) **phenotype**
2) **genotype**
3) **Punnett Sq.**
4) **genotype %**
5) **phenotype %**

Punnet Square (2)

Case: blue-eyed woman mates with
brown-eyed man (homozygous dominant)
Question: What are the chances of blue eyed child?

1) Phenotypes: E and e
2) Genotypes: EE and ee
3) Punnett Square:

	E	**E**
e	**eE**	**eE**
e	**eE**	**eE**

4) Genotype %: EE = 0%, eE = 100%, ee = 0%
5) Phenotype %: E = 100%, e = 0%

Answer: 0% chance of a blue-eyed child.

Carrier Recognition

2 ways to identify carriers of specific genes:

1) **blood tests for recessive genes**
   - **sickle cell anemia, Tay-Sachs, cystic fibrosis**
2) **pedigree**
   - **trace genetic trait through several generations**