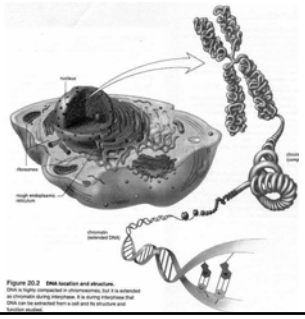
Human Chromosomes

- **Chromosome** = linear DNA wrapped around proteins in the membrane bound nucleus.



Chromosomal Genetics

- Genes (DNA) on chromosomes in cells "direct" cell activities.
- Most human cells have TWO copies of every gene / chromosome. There are 23 chromosomes, thus 46. (DIPLOID)
- One copy from each parent.
- Sperm or egg have ONE copy of every gene / chromosome. 23 chromosomes (HAPLOID)

Eukaryotic Cell Processes

MITOSIS

46 → 46 46

MEIOSIS

46 → 23 23 23 23

FERTILIZATION

23 23 → 46

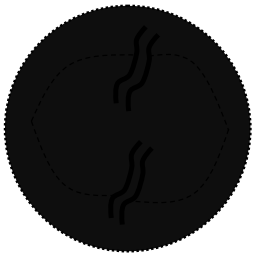
Mitosis

- **Mitosis** = eukaryotic cell division
- One eukaryotic cell becomes two genetically identical offspring (daughter) cells.
- Chromosomes duplicate.
- The nuclear membrane disappears.
- 46 chromosomes move to each end of the cell.
- The cell splits in two.

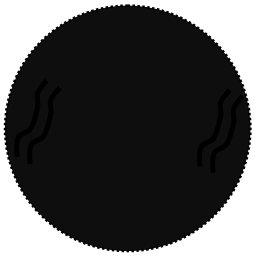
Mitosis

Mitosis

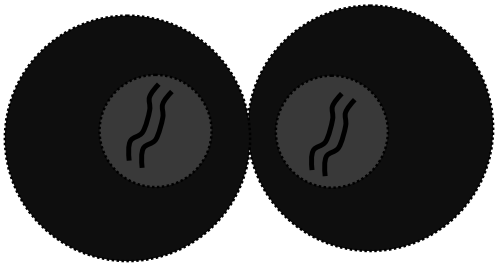
Mitosis



Mitosis



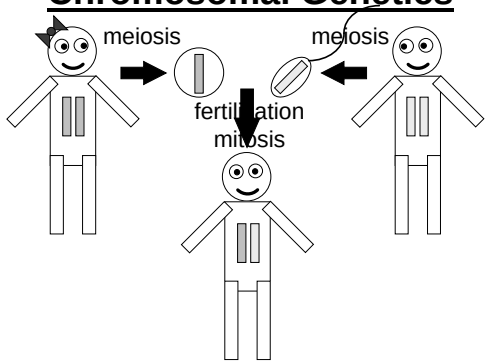
Mitosis



Meiosis

- **Meiosis** = production of eukaryotic sex cells
- One eukaryotic cell becomes **four** cells with 1/2 the number of original chromosomes.
- Sperm and egg are formed by meiosis.
- All other human cells are formed by mitosis.

Chromosomal Genetics



Human Chromosomes

- Human chromosomes numbered 1-23.
- Chromosomes in pairs not numbered separately.
- Paired chromosomes are visually identical with one exception...

  the sex chromosomes (#**23**), **X** & **Y**.
- X chromosome = large w/ many genes.
- Y chromosome = small with few genes.

Human Sex Determination

- Women = **XX**; Men = **XY**.
- Because the Y chromosome contains few genes…

  men are **functionally haploid** for most X chromosome genes.
- Increase in certain genetic disorders in men:

  Hemophilia, color blindness, muscular dystrophy

Chromosomal Disorders

- Sometimes meiosis does not work right…

  ova or sperm with too many or too few chromosomes.
- **Down syndrome** = 3 chromosomes #21 (47 chromosomes total).

  Symptoms - mental retardation and specific physical characteristics

  Not fatal but reduced life span.

Chromosomal Disorders

- **Turner syndrome** = Only one sex chromosome, X. (45 total)

  Female but infertile. Not fatal.
- **Kleinfelter syndrome** = Three sex chromosomes, XXY. (47 total)

  Male but infertile. Reduced secondary sexual characteristics. Not fatal.
- **Jacob syndrome** = Three sex chromosomes, XYY. (47 total)

  Male. Some learning disabilities.

Basic Genetics

- **Genotype** = the genes of an individual.
- **Phenotype** = the physical appearance of an individual.
- Two different genotypes can cause the same phenotype.
- The relationship between genotype and phenotype is known for some genes.

Basic Genetics

- The same gene can have different forms (**Alleles**).
- The eye color gene has 2 alleles brown and blue.
- An individual can have any two of those forms.
  Remember, people have 2 copies of every gene.
- Same 2 alleles = **homozygous**.
- 2 different alleles = **heterozygous**.

Genetics

- Gene “B” has 2 alleles, “B” and “b.”
- BB or bb = homozygous.
- Bb = heterozygous.
- BB, Bb, and bb = genotypes.
- **Genotype** = an organism’s alleles

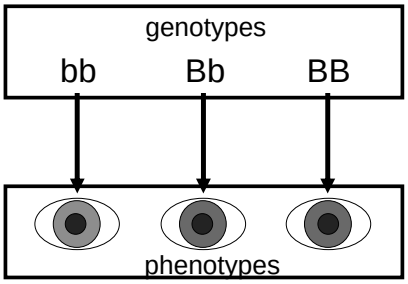
Genetics

- **Dominant Allele** = allele that is expressed in the phenotype whenever present. (capital letter)

  BB or Bb (homozygous dominant or heterozygous)
- **Recessive Allele** = allele that is expressed in the phenotype only when present with another allele. (lowercase letter)

  bb (homozygous recessive)

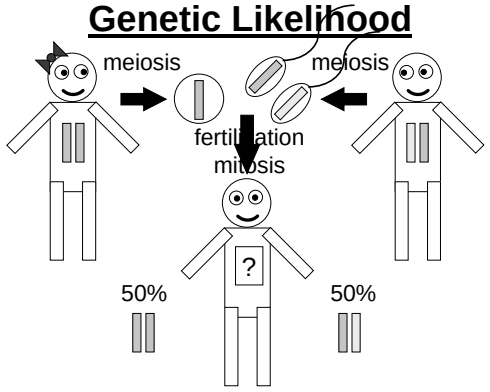
Genetics



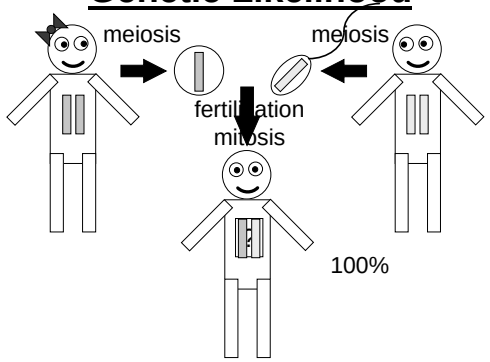
Genetic Likelihood

- What is the likelihood that children will have a specific genotype or phenotype based on parental genotype?
- Why care?
- Likelihood of inheriting genetic disorders and other traits such as pattern baldness.

Genetic Likelihood



Genetic Likelihood



Genetic Likelihood

- **Punnett Square** = table used to determine the likelihood of various genotypes in offspring.

  Possible genetic composition of gametes along axes.

  Combine in center of table.
- Gives you the **probable** distribution of **genotypes**.

Genetic Likelihood

- **Mom's genotype** = Bb
- Gametes mom can produce = B & b
- **Dad's genotype** = bb
- Gametes dad can produce = b & b

	B	b
b	Bb	bb
b	Bb	bb

50% = Bb
50% = bb

Genetic Likelihood

- **Mom's genotype** = Bb
- Gametes mom can produce = B & b
- **Dad's genotype** = Bb
- Gametes dad can produce = B & b

75% = brown
25% = blue

	B	b
B	BB	Bb
b	Bb	bb

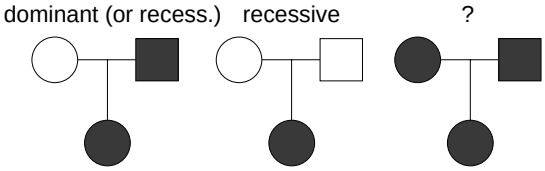
25% = BB
50% = Bb
25% = bb

Pedigree

- **Pedigree Chart** (geneology) = Traces pattern of mating and phenotype for a particular characteristic.

  Female = circle

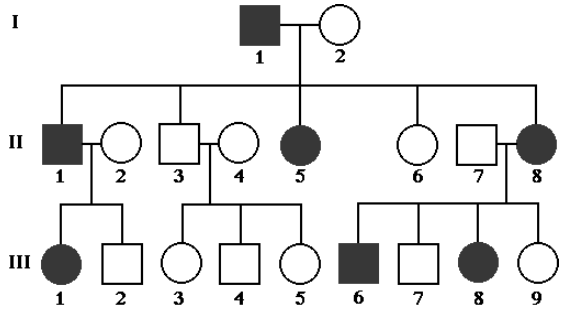
  Male = square



Pedigree

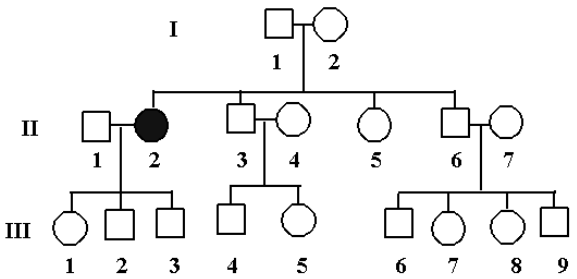
- Traits that show up every generation + never reappear if they disappear = most likely dominant.
- Traits in men and women that “skip” a generation = recessive.

Dominant



Pedigree 1. An idealized pedigree of a family with hypercholesterolemia, an autosomal dominant disease where the heterozygote has a reduced number of functional low density lipoprotein receptors.

Recessive



Sex Linkage

- **Sex Linked** = characteristics controlled by genes of the sex chromosomes are sex linked.
- **X-linked** = X-chromosome genes
- **Y-linked** = Y-chromosome genes (very very few genes are Y-linked)
- In men the X chromosome comes from the mother and the Y chromosome comes from the father.

Sex Linkage

- **Carrier** = individual with a normal phenotype but carry a gene for a (recessive) genetic disorder.
- **X-linked recessive disorders**

  color blindness

  hemophilia A

  hemophilia B

  Duchennes muscular distrophy

X-linked Recessive Disorders

- More males than females affected.
- The disorder often “skips” generations.
- Affected male can have unaffected parents.

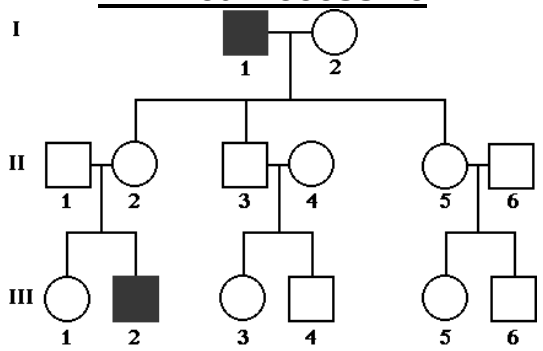
  (mother a carrier)
- Affected female must have an affected father.
- All the male children of an affected female will have the disorder.

Pedigree & X linkage

- An X-linked recessive will...
- Be able to “skip” a generation ***through a female carrier***.

  An affected male can have unaffected parents.
- **Females showing the trait MUST have a father who showed the trait.**
- All male offspring of females showing the trait will also show the trait.

X-linked Recessive

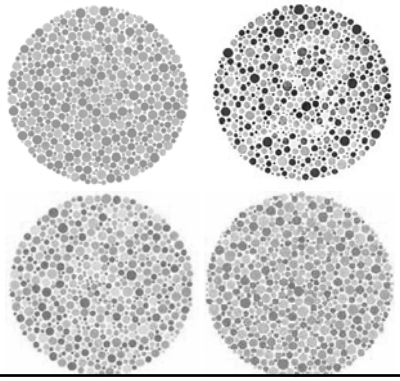


Pedigree 7. X-linked recessive inheritance.

Red-Green Color Blindness

- Green and red color receptor genes are on the X-chromosome.
- The disorder is caused by a recessive allele.
- Individuals cannot or have trouble distinguishing red and green.
- 8% of men of European descent

Red-Green Color Blindness



Hemophilia

- Caused by a defective allele that causes an important *blood clotting* protein to not be formed.
- Individuals can easily bleed to death.
- Hemophilia prevalent in European royal families (late 1800's).
- One in 10,000 males are hemophiliacs.

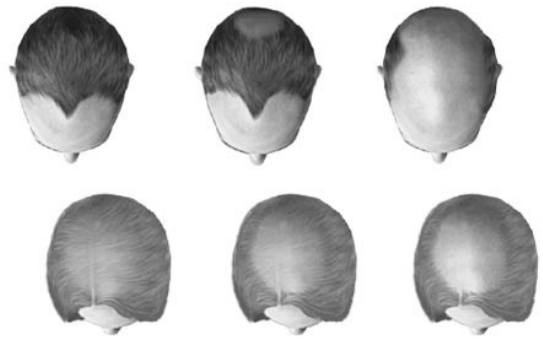
Sex Influence

- **Sex-influenced traits** = genes not on the sex chromosomes but phenotype dependent upon sex of the individual.
- Often the phenotype is determined by the levels of sex hormones.
- An allele may be dominant in females and recessive in males or vice versa.

Pattern Baldness

- No pattern baldness (H^N) = dominant in women, recessive in men
- Pattern baldness (H^n) = dominant in men, recessive in women
- H^NH^N = no p.b. woman or man
- H^NH^n = no p.b. woman
  p.b. man
- H^nH^n = p.b. woman or man

Pattern Baldness



Inbreeding

- **Inbreeding** = mating and producing offspring with relatives.
- **Incest Taboos** = present in virtually all human societies (don't mate within your nuclear family).
- Biologically what's the problem?
- Rare, recessive genes for genetic disorders are more likely to become homozygous and appear in the phenotype.

Inbreeding