Genetics

Proteins are responsible for the color of your eyes, the texture of your hair, the color of your skin, and all the other traits that you possess. You resemble your parents because you inherited the genes for making these proteins from your parents. If a gene on your parent's chromosome that carries a code for making a specific protein is not functioning correctly, you may also inherit that gene for that defective protein.

The traits you inherit from your parents can be determined by a single pair of genes, known as **alleles** (two genes that may represent different forms of the same trait) or by several pairs of genes. A single pair of genes may also influence more than one trait. For example, Cystic Fibrosis and Huntington's disease are caused by a single pair of genes. Eye color and skin color are determined by multiple gene pairs. Sickle cell anemia is controlled by a single pair of genes that affects multiple traits.

Some traits you inherit are observable, such as hitchhiker's thumb, rolling of your tongue, or a widow's peak. Inheritance of other traits is not so obvious. Perhaps, due to inheritance, you may have a defective heart, a single kidney or defective enzyme(s) (ex: defective respiratory enzyme that does not permit your body to manufacture sufficient ATP) and/or hormone(s) (ex: impaired ability to manufacture insulin resulting in diabetes) or impaired functioning of receptors on cell membranes.

Gregor Mendel – the father of Modern Genetics

Many of the concepts of modern genetics were worked out in the mid-1800s by Gregor Mendel, an Austrian monk, who applied mathematical probabilities to the study of plant inheritance. Specifically working with pea plants, Mendel studied the inheritance of seven different contrasting traits: tall/short stems, round/wrinkled seeds, terminal/axial flower position on stems, green/yellow pea pods, yellow/green seed color, colored/white seed coat color, and inflated/constricted pod shape.

General Terms

- There are often two or more ways for a trait to be expressed, dependent upon the combination of alleles for that given trait.
- **Dominant:** the allele for a given trait is expressed. For example, in pea plants tall allele is dominant over the short stem allele and will be expressed when present. Dominant alleles are designated by a capital letter. In this case, TT or Tt alleles will be expressed as tall stemmed plants.
- **Recessive:** the contrasting form of a trait that is masked from being expressed by the dominant allele. Recessive alleles are designated by a lower case letter. The short stem trait (as discussed above) would only be expressed if both alleles were recessive – tt.
- **Pure-bred or homozygous**: when both the alleles for a given trait are in the same form. Ex. TT or tt
- **Heterozygous or hybrid:** when one allele is dominant and one allele is recessive
- **Phenotype:** physical appearance of the organism
- **Genotype:** the genetic makeup of an individual

Law of Dominance

- When Mendel crossed a pure dominant (TT) tall plant with a (tt) short plant, all resulting offspring were hybrid tall (Tt)
- Mendel concluded that when crossing two purebred plants what showed one pair of contrasting traits, the offspring expressed the dominant trait only. The recessive trait was masked by the presence of the dominant trait.

Punnett Squares

- The Punnett square is a convenient method for visualizing genetic crosses. The boxes that make up the square show all possible zygotes resulting from a cross of gametes bearing a specific trait. The letters representing each possible type of sperm are placed along the top of the square and those for each possible type of egg along the side.
- Look at the following example below of a cross between a pure tall (TT) plant and a pure short (tt) plant.

Parent (P) generation

TT X tt

Gametes

	t	t
	Tt (tall)	**Tt (tall)**
	Tt (tall)	**Tt (tall)**

Offspring: F_1 or first filial generation
Phenotype: 100 % tall
Genotype: 100% hybrid tall

Law of Segregation and Recombination (meiosis and fertilization)

- In another experiment, Mendel allowed the results of the F_1 generation to self-fertilize. 75% of the F_2 offspring were tall, 25% of the F_2 offspring were short.
- This result led Mendel to conclude that each trait he studied was controlled by a pair of alleles, one allele from each parent.
- Using the Punnett square below, determine the phenotype and genotype of a cross between two plants hybrid for round/wrinkled seeds (Rr x Rr)

Rr X Rr

F_2 generation

Phenotype: **Genotype:**

Law of Independent Assortment

- Mendel also crossed plants that showed two pairs of contrasting traits. For example, Mendel crossed a plant with long stems and round seeds (TTRR) with a plant that had short stems and wrinkled seeds (ttrr). The resulting F_1 offspring were all long stemmed with round seeds.
- Mendel let the F_1 generation self-fertilize. This is known as a **dihybrid cross** (TtRr X TtRr). The resulting offspring (F_2 generation) evidenced the phenotypic ratio of **9 tall, round; 3 tall, wrinkled; 3 short, round; 1 short, wrinkled plants**.
- The results indicated that the alleles for each trait segregated independently of any other traits during gamete formation.

Exceptions to Mendel's Laws

1. **Intermediate Inheritance**
   a. **Incomplete Dominance**
   - In such inheritance, the hybrid organism is different in appearance from either of the homozygous parents.
   - For example, in 4 o-clock flowers and snapdragons, where **R = red gene and W = white gene, RR=red flower, WW=white flower, and RW = pink flower**
   - Now, try to determine the phenotypes and genotypes when a red snapdragon is crossed with a pink snapdragon.

RR X RW

Phenotype: **Genotype:**

   b. **Co-dominance**
   - Both alleles in the hybrid organism are dominant and expressed.

- For example, in certain cattle and horses, the trait for coat color is either white hair or red hair. A horse with C^RC^R has a red coat color. A horse with C^WC^W shows a white coat color. For a heterozygous horse with a C^RC^W coat, (**roan colored coat**) where both white and red hairs are expressed.
- Practice: determine the offspring of a cross between a horse with a white coat and a horse with a roan coat.

Phenotype: **Genotype:**

2. Multiple Alleles

- There are certain traits, like skin color and blood type that are controlled by multiple alleles. However, no more than two alleles for a given trait can be present in a normal cell.
- This means that even though there are more than two alleles for a given trait, a given organisms can only have two of these alleles.
- Let's examine blood types. You already know that there are three different alleles for blood type – A, B, and O which are designated as I^A, I^B and i respectively.
  -- **both A and B alleles are dominant over the O allele**
  --**A and B alleles are codominant**
  --blood **type A** alleles can be **I^AI^A or I^Ai**
  --blood **type B** alleles can be **I^BI^B or I^Bi**
  --blood **type AB** alleles are **I^AI^B** only
  --blood **type O** alleles are **ii** only
- What are the possible blood types of the offspring from a heterozygous AB mother and an O
- blood type father.

Possible blood types of offspring:

3. Linkage

- When scientists discovered the events of meiosis, they realized that traits are inherited independently of one another only when these traits are one nonhomologous chromosomes.
- When the genes for two different traits are located on the same pair of homologous chromosomes, these traits are linked and tend to be inherited together.
- Mendel's Law of Independent Assortment had to be modified to include the concept of linked genes inheritance

4. Crossing Over

- During meiosis, homologous chromosomes undergo synapsis, forming tetrads. During this process, segments of chromosomes may be exchanged between homologous chromosomes
- The greater the distance between two genes on a chromosome, the more likely these genes are to be separated by crossing over.
- Genes that are closer together on a chromosome tend to be inherited together, such as freckles and red hair.

5. Sex Chromosomes

- Sex in humans and various other organisms (both plant and animal) is determined by a specific pair of chromosomes known as the sex chromosomes
- **Autosomes:** all chromosome pairs other than the sex chromosomes
- Humans have 22 pairs of autosomes and 1 pair of sex chromosomes
- In humans, sex chromosomes are designated X and Y
- **Females: XX Males: XY**
- In gamete formation, female eggs always contain one X chromosome. Male sperm can carry either an X or Y chromosome – therefore, **in humans, the sperm of the male determines the sex of the offspring.**

6. Sex-linked Traits

- Traits determined by genes located on the X or Y chromosome are said to be sex-linked traits.
- **Color-blindness and hemophilia are two recessive traits that are linked to the X chromosome.** These traits are more common in males than females because males only carry one X allele. The traits tied to the X chromosome will always be expressed in a male, regardless if the trait is dominant or recessive. Females must have two X of these defective genes to express the trait of color blindness or hemophilia.
- If a female has one defective gene, say for color blindness, she will have normal vision but is a carrier for the disorder and pass the gene to her offspring.
- Try to solve the following problem:

  Show the possible offspring produced by a normal father (XY) and a carrier mother for color blindness (XX^-) Note: X^- indicates a color blind gene

a. What percentage of their daughters will be colorblind?
b. What percentage of their daughters will be carriers of the color blind gene?
c. What percentage of their sons will be colorblind?