Basics of Mendelian Genetics.

The universal principles developed by Gregor Mendel a century and a half ago using garden peas have been shown to operate in other sexually reproducing species as well, including one descended from an African ape-like creature.

The study of genetics enables us to understand how biological characteristics encoded on the chromosomes can be transmitted from parent to offspring, and how variability in these characteristics occurs. In this exercise you will study the results of genetic crosses, examine some of the ways in which genes express themselves and solve a variety of genetics problems. In addition, each member of your group will prepare to present a solution to a unique problem generated by the Virtual Genetics Lab software at a later laboratory meeting.

Genes controlling a given trait occur at corresponding positions (gene loci) on homologous chromosomes (pairs of chromosomes—one maternal, one paternal—received from one's parents). There may exist two or more expressions (light, dark, etc.) for any characteristic. These alternate forms of a gene at a particular locus are called alleles (you have two alleles for most characteristics).

If the pair of alleles an individual received are identical, the individual is said to be homozygous for that trait. In a heterozygous individual, the alleles that code for a trait are different, which means that an individual developed from an embryo formed by the fusion of gametes bearing dissimilar alleles for that gene. In the heterozygous condition one allele may be dominant and totally mask the presence of the other (thus recessive) allele.

To determine what types of offspring may be produced, we can look at the parental genotypes for the particular traits we wish to observe. If we are looking at just one trait, we call it a **monohybrid cross**. For example, let us assume that curly hair (symbol C) is dominant over straight hair (c); and that two parents are both heterozygous (Cc) for curly hair. What kind of offspring and what proportion of each, would they be expected to have?

To solve this problem, it is best to set up a **Punnett Square** as seen below. The gametes formed by the female contain either gene C or c, in equal proportions. These are written across the top of the square:
In this case, the gametes of the male are the same and are written on the outside of the box and along the left hand column (**do this now**). To determine the offspring, we simply combine the gametes that contribute to form each square. For example, in the upper left square, C and C are combined to give CC offspring. In this particular cross, three out of four offspring would be expected to have curled toenails,

	C	c
_____		
_____		

and one out of four should have straight toenails. Of the ones with curled toenails, two out of three would be expected to be heterozygous for the trait. The proportion of offspring expected to exhibit a certain trait is usually written as a fraction, that is ¾ would be expected to have curly hair, and ¼ straight hair. Bear in mind that these are probabilities, based on **expected** results in a large population. Not every family has four children, and even if there were four children, they would not necessarily follow the expected probabilities.

In the same way we worked out the types of offspring in a monohybrid cross, we can tell What kind of the offspring are expected in a **dihybrid** cross, that is one in which we are working with **two** traits carried on **different** chromosomes. Since they are on different chromosomes they segregate independently from each other. Suppose we are dealing with two traits, for hairline and earlobes. Let P = widow's peak and p = straight hairline. E = detached lobes and e = attached lobes. The parents in this hypothetical case have the genotypes: PpEe x PpEe (that is both parents have a widow's peak and detached earlobes).

PE			

We must first determine all the possible combinations of genes in the gametes in order to solve this problem. In the case of the male parent, P may enter the same gamete as E (= PE) or e (= Pe). We therefore have four kinds of gametes which would be expected to form in equal proportions: PE, Pe, pE & pe. The same gametes would be formed by the female. We can now proceed, as before, by setting up a Punnett square as shown below, with the female gametes across the top and the male gametes at the left. Fill in the chart yourself and determine the phenotypes and proportions of the offspring formed.

Another type of inheritance pattern can be found with **sex-linked traits**. A sex-linked characteristic is encoded by a gene on a sex chromosome, usually the X chromosome. Note that there is no homologue for the X chromosome in the male; therefore, whatever genes are present on the X chromosome (whether they are dominant or recessive) are expressed in his phenotype.

Some humans are born without a critical protein, which results in their development of so-called male pattern baldness. The defective allele **b** is found on the X chromosome (coded as $X^{\mathbf{b}}$). Use a Punnett square to predict the possible offspring, male and female, of a couple in which the man has male pattern baldness and his wife has normal hair even though her father had male pattern baldness (the woman's genotype is written in for you). What is the probability that their children will have male pattern baldness? Why do you think this trait, like all human sex-linked traits, is more common in males than females?

X^B	X^b

Paper & Pencil Problems

Gain some skill with the genetic problems below. You'll need the practice, because afterwards you'll put these skills to work solving problems on the Virtual Genetics Lab software. In a later class meeting, each member of your group will present a solution to one of the VGL problems. Work with your group but be prepared to stand alone.

1. One version of a gene directs the assembly of an enzyme that breaks down a complex amino acid called phenylalanine into simpler components. Both Ken and Barbie have that version. But the hospital where Barbie has just had their new baby informs them that their daughter Scooter doesn't have it. She only has altered versions of the gene that are incapable of assembling the enzyme correctly. She won't be able to metabolize phenylalanine and will almost certainly become severely mentally retarded if she is not kept on a diet low in that amino acid for the first three years of her life. Write the probable genotypes for all three of them.
2. Achondroplasia, a form of dwarfism, is inherited as a dominant condition. Two dwarfs, both of whom had one parent of normal height, marry and plan to a have child. Their first child is born and is a dwarf. What is the probability that this child is heterozygous?
3. A pair of dominant alleles influence coat color in cattle wherein homozygotes for one allele have red coats, homozygotes for the other allele have white coats, and heterozygotes have roan coats that are produced by an intermix of both red and white hairs. When two or more different dominant alleles could occur at the same locus we call them "codominant" since each is fully expressed when present.

   Suppose that you start with a herd of 20 red cows, 40 roan cows, 20 white cows, one red bull, two roan bulls, and one white bull. Understanding that you can choose which bulls will mate with which cows and which will become hamburger, write a plan to manage your herd and achieve the following goals:

   a. Maximize the number of roan calves produced.
      (Could you achieve 100%?)

   b. Minimize the number of roan calves produced.
      (Could you achieve 0%?)

   c. Produce only roan and red calves.
4. Red-green colorblindness is a recessive sex-linked trait on the X chromosome. A woman, whose father had red-green colorblindness, marries a man who is colorblind. What is the probability that any son born will have red-green colorblindness?
5. In humans, normal skin color (A) is dominant over albino (a) and streptomicrodactyly (bent little fingers) is dominant to straight pinkies. A albino man with straight little fingers marries a normal woman with bent little fingers whose mother was an albino and whose father had straight little fingers. What are the genotypes of the man and the woman? What proportion of their children would be expected to be both non-diabetic and have normal color?

These instructions and problems are based on an earlier set formulated by Brad Paschal and Michael Bucher.