CHAPTER 6: HEREDITY

IF YOU ONLY LEARN SIX THINGS IN THIS CHAPTER . . .

1. Meiosis refers to the process by which sexually reproducing organisms produce sex cells (gametes) with half the chromosomes (haploid) of the rest of the organism's cells (which are diploid). It has two stages, meiosis I and meiosis II, and results in the creation of four gametes. In sexual reproduction, the male and female gametes join to create a new organism with the normal number of chromosomes.
2. Different versions of a gene that code for the same trait are called alleles. In classical (Mendelian) genetics, an individual receives one allele from each parent. Individuals with matching alleles are homozygous for that trait while those with different alleles are heterozygous. Usually one version of the allele is dominant (e.g., brown eye color) and the other is recessive (e.g., blue eye color). Heterozygotes are "ruled" by the dominant allele.
3. Geneticists perform test crosses to determine the genetic makeup (genotype) of organisms displaying the dominant phenotype. A Punnett square is used to illustrate a test cross. Mendel's Law of Segregation states that an individual's alleles separate during meiosis, and either may be passed on to the offspring. Mendel's Law of Independent Assortment states that inheritance of a particular allele for one trait does not affect inheritance of other traits.
4. Non-Mendelian inheritance patterns include incomplete dominance, epistasis, polygenic inheritance, pleiotropy, genetic recombination, and gene transfer.
5. Sex-linked genes occur on the *X* or *Y* chromosome. Males who inherit recessive *X*-linked genes from their mothers always express the trait in question, as men have only one *X* chromosome.
6. Chi-squared analysis refers to a means of determining if experimental observations are significantly different from the expected result.

THE ROOTS OF THE FAMILY TREE: UNDERSTANDING INHERITANCE

The concepts in this chapter include classical genetics, the principles of inheritance through gamete formation, and sexual reproduction. Many of these concepts rely more on sequential logic and are less memory intensive, which is helpful to know when studying for the exam.

IMPORTANT

Even if you never took a biology class, it's hard not to notice that children look more like their parents than other adults. The passing on of characteristics or traits is controlled by the genetic information contained within the DNA in cell nuclei.

KEY POINT

The College Board suggests that 8 percent of the questions on the exam cover the concepts discussed in this chapter.

However, you are likely to find more questions on the information presented in this chapter than on cellular energetics. Be prepared!

Meiosis and Gametogenesis

In chapter 4, you reviewed how cells divide and duplicate through the process of mitosis. Mitosis results in the production of identical cells for growth or, in the case of single-celled organisms, **asexual reproduction**. In **sexual reproduction**, two parent organisms each contribute a cell, which combine to form an offspring that shares half of each parent's DNA. Sexual reproduction can also occur when a hermaphroditic organism fertilizes its own eggs to form offspring.

An organism that has two sets of **chromosomes** is said to be **diploid** (designated as 2N). A diploid organism has one set of chromosomes from each parent, for a total of two sets. When a cell has one set of chromosomes, which is half the number of chromosomes that a diploid cell contains, it is said to be **haploid** (1N or N). In some cases cells have several sets of chromosomes, and are called **polyploid** (3N, 4N, etc.). There are a few organisms that exist in a natural state with haploid or polyploid cells, but most organisms are diploid.

When diploid organisms sexually reproduce, one of their own cells is combined with a cell from the other parent. These cells, called **gametes**, are haploid before they combine. When gametes combine, they form a diploid offspring. The formation of these special haploid cells (gametes) is called **gametogenesis**. The process that forms cells with one set of chromosomes (haploid cells) is called **meiosis**.

Here is what you need to remember so far:

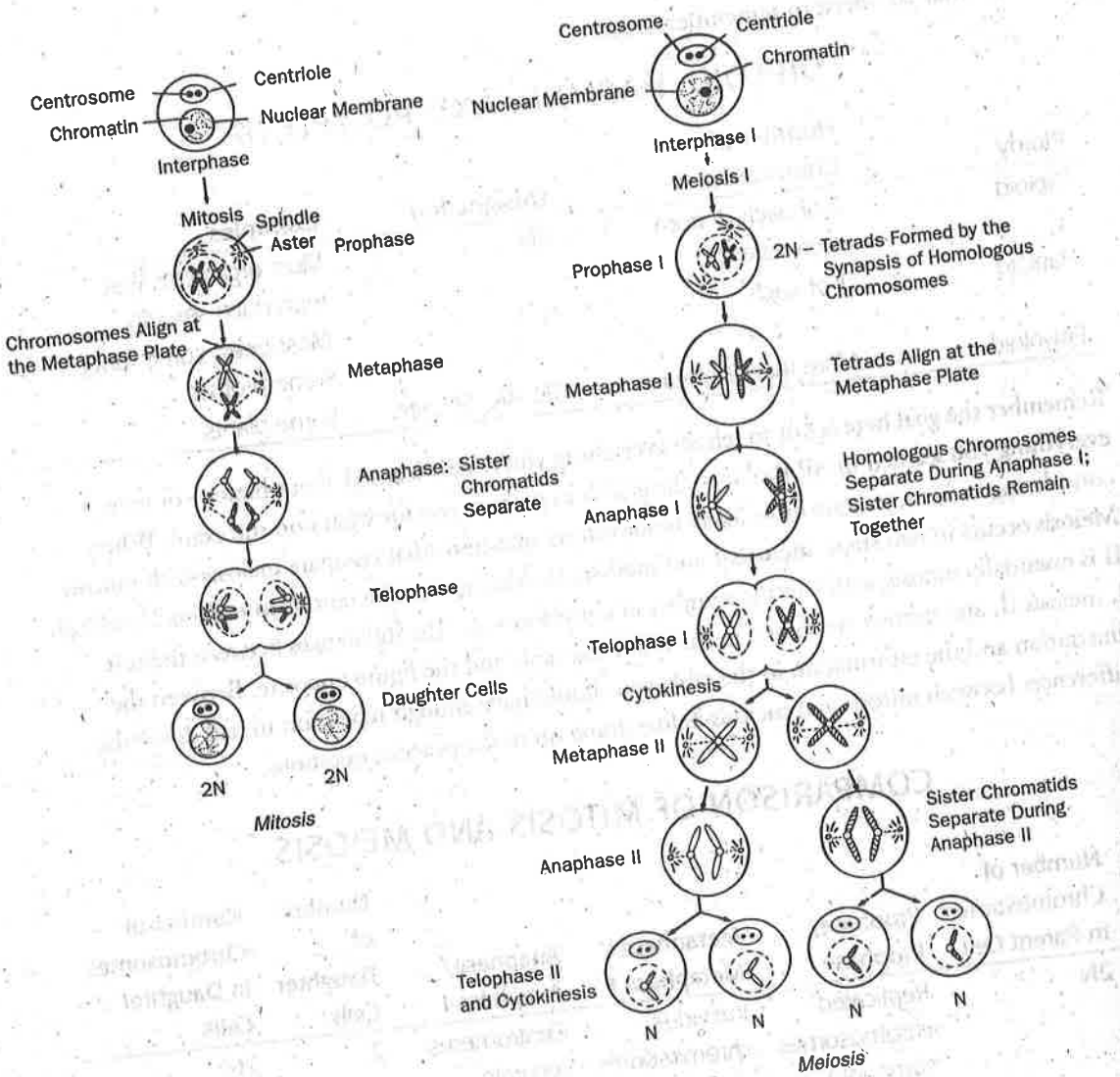
DIPLOID, HAPLOID, AND POLYPLOID

Ploidy	Number of Chromosomes	Designation	Examples
Diploid	2 of each, 1 from each parent	2N	Most organisms that reproduce sexually
Haploid	1 of each	N	Most bees, some fungi, some protists
Polyploid	More than 2 of each	3N, 4N, 5N, etc.	Some plants

Remember the goal here is not to rehash everything you've ever learned about biology or even everything you learned in AP Biology. The goal is to prepare you for what's on the exam. When considering meiosis, you are most likely to encounter questions that compare meiosis with mitosis. Meiosis occurs in two steps, **meiosis I** and **meiosis II**. Meiosis I is different from meiosis II; meiosis II is essentially mitosis with half the number of chromosomes. The differences between meiosis I, meiosis II, and mitosis are shown in the following table and the figure opposite. Between the illustration and the information in the table you should have enough repetition to nail down the differences between mitosis and meiosis before going on to the practice questions.

COMPARISON OF MITOSIS AND MEIOSIS

Division	Number of Chromosomes in Parent Cell	Prophase/ Prophase I	Metaphase/ Metaphase I	Anaphase/ Anaphase I	Number of Daughter Cells	Number of Chromosomes in Daughter Cells
Mitosis	2N	Replicated chromosomes come into view as sister chromatids	Individual chromosomes align at metaphase plate	Centromeres separate and sister chromatids travel to opposite poles	2	2N
Meiosis	2N	Chromosomes form tetrads by synapsis; crossing over at chiasmata	Pairs of homologous chromosomes align at metaphase plate	Synapsis ends and homologous chromosomes travel to opposite poles; sister chromatids travel to same pole	4	N



Comparison of Mitosis and Meiosis

The ultimate goal of meiosis is to produce cells with half the number of original chromosomes, so that two cells with half the number of chromosomes can combine to create offspring with a complete set of chromosomes. Later, you will review how chromosomes sort to provide genetic material from each parent to the offspring.

EUKARYOTIC CHROMOSOMES

Chromosomes are condensed bodies of DNA molecules that store codes for the translation of several different kinds of proteins. These proteins dictate how an organism is put together and functions. In the most simplified way of looking at the **chromosomal theory of inheritance**, each

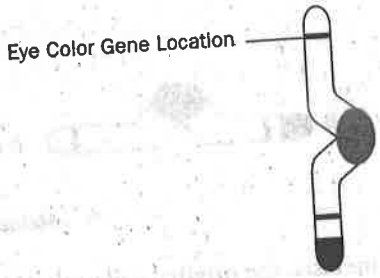
section of DNA that translates a different protein is called a **gene**. A region of DNA may code for a protein that controls eye color, so this region of DNA is called the gene for eye color.

GENES

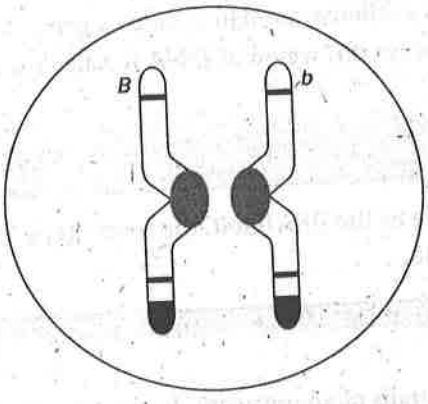
Instead of thinking of a gene as the DNA itself, the term "gene" can be associated with a location on the chromosome.

Alleles specifically code for the traits of an organism. In the case of eye color, there might be an allele for blue eye color and an allele for green eye color. The eye color that an organism ends up with (its **phenotype**) is dictated by which allele is placed in the gene location for eye color (its **genotype**). Sometimes an organism might have a genotype for one allele and express the phenotype of another.

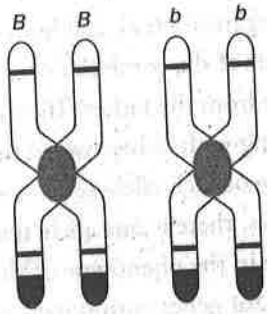
In a typical diploid (2N) eukaryotic organism, each cell has two copies of each chromosome. These pairs of chromosomes are a result of the combination of two haploid gametes formed by meiosis, one from the mother and one from the father. The mother and father each provided one allele for each gene, leaving the offspring with either two of the same allele (e.g., two for blue eye color or two for green eye color) or one of each allele (e.g., one allele for blue eye color and one allele for green eye color). In most genes, there is one allele that is **dominant** over the other and hides the expression of that other allele in the phenotype of the offspring. The other allele is called **recessive**. Geneticists record the alleles for genes with uppercase, italicized letters for dominant alleles (*B*) and lowercase, italicized letters for recessive alleles (*b*). If blue eye color is the dominant allele (indicated as *B*) and green eye color is the recessive allele (indicated as *b*) an offspring could have one of three different genotypes (*BB*, *Bb*, and *bb*) when its parents' gametes combine. The phenotype of an offspring with the genotype *BB* is blue eyes. An offspring with the genotype *Bb* will also have blue eyes because the blue eye color allele is dominant. Only offspring with the genotype *bb* will have a green eye phenotype. The following figures clarify this information.



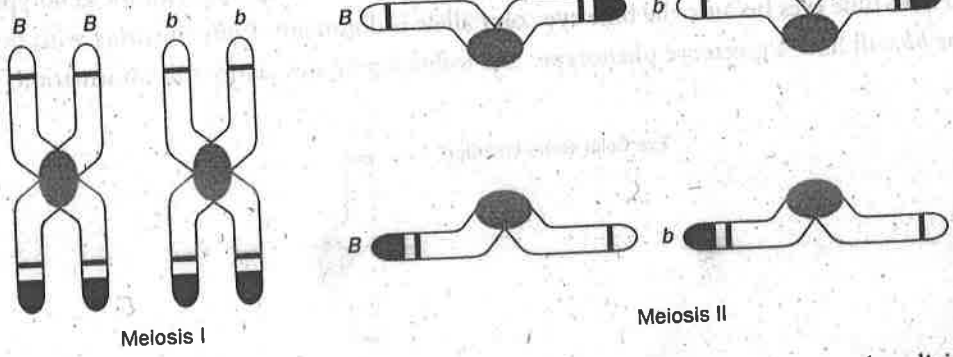
The model above indicates the location of the gene for eye color on an animal chromosome. Each animal has two of each of these chromosomes, one from its mother and one from its father.



The model above is a cell from an animal with one allele for blue eye color (*B*) and one allele for green eye color (*b*). This animal's genotype for eye color is *Bb* and its phenotype is blue eyes because the blue eye color allele is dominant. The chromosomes are essentially identical in appearance, but the alleles have a slightly different DNA code.



The figure above shows chromosomes after interphase I of meiosis. The DNA strands have been doubled so that there is an exact duplicate of each original chromosome linked by a centromere to sister chromatids. There are now two copies of the *B* allele and two copies of the *b* allele.



After the cell undergoes complete meiosis, the original cell with 2N chromosomes has divided into four daughter cells, each with 1N chromosomes. The daughter cells are four gametes, two with the *B* allele and two with the *b* allele. Since the original animal cell had both a *B* allele and a *b* allele, it can produce gametes with alleles for either blue or green eye color. This occurs in all

individuals of the same species. An individual with the recessive green eye color (its genotype is *bb*) can only produce gametes with *b* alleles. When an organism has two of the same alleles (i.e., *BB* or *bb*), it is called **homozygous** for that gene. If both alleles are the dominant allele, the gene is called **homozygous dominant**. If both alleles are recessive, the gene is called **homozygous recessive**. If the organism has different alleles (i.e., *Bb*), it is called **heterozygous** for the gene.

Since the green eye color allele is recessive, it can be discerned that an individual in the population with green eyes has the genotype *bb*. An individual with blue eyes can have either a *BB* genotype or a *Bb* genotype. The genotype of a blue-eyed individual can be determined by mating the blue-eyed individual with a green-eyed individual. This is called a **test cross**. If all of the offspring have blue eyes, then the blue-eyed individual was homozygous dominant. The genotypes of all of the offspring from a mating between a *BB* genotype and a *bb* genotype can only be *Bb*. If any of the offspring have green eyes, the blue-eyed adult must have been heterozygous. The genotypes of the offspring from a mating between a *Bb* genotype and a *bb* genotype are either *Bb* or *bb*. Mating between two heterozygous adults can produce offspring with three different genotypes: *BB*, *Bb*, and *bb*. In this case, two blue-eyed adults can produce a green-eyed offspring.

Inheritance Patterns

After learning the basics of genetics, it is important to review some specific information about inheritance that might appear on the exam. You need to be familiar with mathematical principles of simple probability. Relax; it's not as hard as it sounds. Even if there are more than two alleles for a gene, an individual can only have two alleles on each pair of chromosomes. Ending up with a combination of alleles is like tossing a coin.

Take an organism that is homozygous for a particular gene (*AA*). This organism produces four gametes, each with an *A* allele. Each of the gametes is one out of a possible four gametes, but because all of the gametes have an *A*, four out of four of the gametes have an *A*. Four out of four is a probability of one $\left(\frac{4}{4}=1\right)$. You can look at the question more intuitively by saying that since there are only *A* alleles in the parent cell, all of the daughter cells $\left(100 \text{ percent or } \frac{1}{1}\right)$ will have the *A* allele. If an organism is heterozygous (*Aa*) it produces two gametes with an *A* allele (2 out of 4) and two gametes with an *a* allele (2 out of 4). The probability of either the *A* or *a* allele in the gametes is $\frac{1}{2}\left(\frac{2}{4}=\frac{1}{2}\right)$. Or again, intuitively, if the parent cell has one *A* and one *a* allele, it will produce gametes that are half *A* $\left(50 \text{ percent or } \frac{1}{2}\right)$ and half *a* $\left(50 \text{ percent or } \frac{1}{2}\right)$. Got it?

Now consider two genes and an individual that is homozygous for both (*AABB*). Although there are two genes to consider, the organism still produces four gametes from a single parent cell. Each gamete gets an *A* allele and a *B* allele. What is the probability that a gamete will have BOTH an *A* and *B* allele?

Four out of four will have an $A\left(\frac{4}{4}\text{ or }1\right)$ and four out of four will have a $B\left(\frac{4}{4}\text{ or }1\right)$, so four out of four will have $AB\left(\frac{4}{4}\times\frac{4}{4}=\frac{16}{16}=1\right)$. If the individual is heterozygous for one of the genes (*AABb*), the probability of the *A* allele stays the same, but now only half of the gametes get a *B* allele $\left(\frac{2}{4}=\frac{1}{2}\right)$ and the other two get a *b* allele $\left(\frac{2}{4}=\frac{1}{2}\right)$.

What is the probability of producing a gamete with both an *A* and *B* allele from this individual?

Four out of four will have an $A\left(\frac{4}{4}\right)$ and two out of four will have a $B\left(\frac{2}{4}\right)$, so only two out of four will have $AB\left(\frac{4}{4}\times\frac{2}{4}=\frac{8}{16}=\frac{1}{2}\right)$. If the individual is heterozygous for both genes (*AaBb*), the probability of each allele in the gametes (*A*, *a*, *B*, or *b*) is $\frac{1}{2}$ so the probability of *AB* is $\frac{1}{4}$ $\left(\text{as }\frac{1}{2}\times\frac{1}{2}=\frac{1}{4}\right)$, the probability of *Ab* is $\frac{1}{4}\left(\text{as }\frac{1}{2}\times\frac{1}{2}=\frac{1}{4}\right)$, the probability of *Ba* is $\frac{1}{4}$ $\left(\text{as }\frac{1}{2}\times\frac{1}{2}=\frac{1}{4}\right)$, and the probability of *bb* is $\frac{1}{4}\left(\text{as }\frac{1}{2}\times\frac{1}{2}=\frac{1}{4}\right)$.

Once you can figure out what gametes will be produced from which individuals, you can figure out the genotypes of offspring from your **Punnett square**. The following is a simple cross between two heterozygous individuals for one gene. A mating of this kind is called a **monohybrid cross**.

	A	*a*
A	*AA*	*Aa*
a	*aA*	*aa*

Monohybrid Cross

Typically the gametes of the sperm are recorded along the left side of the Punnett square and those of the egg are recorded across the top. The allele from each sperm is paired with the allele from each egg where the column and rows meet, showing the genotype of the offspring. The different indications of *Aa* and *aA* are used only to demonstrate the source of the alleles; they are the same genotype.

The above cross results in three different genotypes (*AA*, *Aa*, and *aa*), but only two phenotypes because the dominant trait is expressed in both the *AA* and *Aa* individuals. The dominant trait shows up in the offspring in a ratio of 3 to 1.

You can put together a **dihybrid cross** (tracking two genes—*A* and *B*—rather than one) just as easily as a monohybrid cross. Just put the gametes of the male in the left column and the gametes of the female across the top.

	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AAbB	AAbb	AabB	Aabb
aB	aABB	aABb	aaBB	aaBb
ab	aABb	aAbb	aabB	aabb

Dihybrid Cross

The dihybrid cross produces the famous phenotypic ratio of 9:3:3:1. If you consider a large number of offspring, there will be on average 9 out of every 16 that express the dominant phenotypes for both genes, 3 out of every 16 that express the dominant phenotype of one gene, 3 out of every 16 that express the dominant phenotype of the other gene, and only one that expresses the recessive phenotypes for both genes.

These are the basics of inheritance. However, genes do not operate in isolation from one another; this makes genetics more complex than Mendel's experiments and the Punnett square might suggest.

More examples of inheritance patterns are shown in the review questions at the end of the chapter. Genetic terminology is further developed in the lab section of this chapter.

KEY STRATEGY

Notice patterns in the ratios that occur with specific types of crosses. For example, crossing a heterozygous dominant (Rr) with a homozygous recessive (rr) always gives a 1:1 (½ dominant phenotype; ½ recessive phenotype). This can speed up the process of figuring out phenotypic ratios and probabilities.

MITOSIS AND MEIOSIS LABORATORY

This lab is simple and straightforward. The mitosis portion of the lab involves observing commercial slides of some plants and/or animals fixed at various stages of mitosis. This portion of the lab will help you to recognize, through repetition, the different stages of mitosis. It will also give you some perspective on the sizes of cells and cell structures. The lab will provide you with some experience working with a microscope.

The meiosis portion of the lab is usually completed with cellular models that you can hold in your hand to study Mendel's Laws of Inheritance. The models are usually strings of beads or another kind of model linked to a chain.

Another observation to make in this lab is that Mendel's laws aren't true for all genes. There are some genes that do not sort independently from others. These genes may be linked to other genes on the same chromosome. It can also be observed that the probability ratios expected are often not quite what is expected based on Punnett squares. This is because some chromosomes exchange genetic information with each other by **crossing over** when the homologous pairs are

IMPORTANT

As you study, be sure you're aware of the distinct difference between **incomplete dominance** and **codominance.**

PHENOTYPE AND GENOTYPE—FACTORS TO CONSIDER

Many different factors can affect phenotype and genotype in offspring. You should be familiar with the following topics:

- **Incomplete dominance:** a form of inheritance in which heterozygous alleles are *both* expressed. This means that the offspring will display a combined phenotype that is distinct from both parent organisms. For example, a plant with purple flowers and a plant with white flowers might produce offspring that have pink flowers.
- **Epistasis:** two or more genes (that are not alleles) interact to control a single phenotype. For example, one gene might act to suppress the expression of another gene; this is what happens in albinism, when recessive albinism alleles governing production of the enzyme that catalyzes melanin production (tyrosinase) prevent expression of the genes that govern the amount of melanin production. In epistatic interactions, a gene's effect on phenotype depends on the presence or absence of other genes elsewhere on the chromosome.
- **Polygenic inheritance:** a type of inheritance in which several interacting genes control a single trait. Many traits result from the additive influences of multiple genes; skin color is one common example of a polygenic trait.
- **Pleiotropy:** a pleiotropic gene controls more than one characteristic in an organism, often controlling aspects of the phenotype that do not seem to be connected to one another. For example, white cats that get their fur color from certain alleles are also generally deaf. Many genes that cause diseases are pleiotropic, including the genes behind sickle cell anemia and phenylketonuria (PKU).
- **Genetic recombination:** molecular process by which an organism's genes are rearranged in its offspring. Through this process, two alleles can be separated and replaced by different alleles, thereby changing the genetic makeup but preserving the structure of the gene. Chromosomal crossing over is an example of a mechanism by which this process takes place.
- **Gene transfer:** vertical gene transfer occurs when an organism receives genetic material (i.e., DNA) from a parent organism or from a predecessor species. Horizontal gene transfer occurs when an organism transfers genetic material to cells that are not its offspring.

synapsed into **tetrads**. The farther away genes are from each other, the more likely they are to exchange DNA with another **chromatid**. The frequency of certain observed phenotypes can be used to estimate a relative rate of crossovers. The higher the frequency of occurrence of a certain phenotype, the more likely there is crossing over between genes, and the further away the genes are from each other.

TWO LAWS OF MENDELIAN INHERITANCE

The two laws of Mendelian inheritance are as follows:

Law of Segregation—This describes the separation of alleles in the parent genotype during the process of gametogenesis. There can only be a maximum of two different alleles in a single parent; half the gametes get one allele and the other half get the other allele.

Law of Independent Assortment—This suggests that different genes sort into different gametes, independently of each other. For example, the sorting of alleles for eye color is not affected by the sorting of alleles for hair color.

The laws above explain the 3:1 and 9:3:3:1 ratios of phenotypes observed in monohybrid and dihybrid crosses. By separating parent genotypes into gametes and recombining them into offspring with the model beads, you can get a hands-on perspective of chromosomal inheritance.

GENETICS OF ORGANISMS LABORATORY

In this lab you will manipulate several generations of a species of vinegar fly (distinctly different than the true fruit flies in the family Tephritidae) in the genus *Drosophila* (Drosophilidae). Vinegar flies are ideal study organisms, especially for genetics experiments, and have been used for decades to study genetics. The flies are easy to maintain, easy to work with, and have a very short life cycle.

Along with a review of genetics principles, this lab provides some hands-on experience working with diploid organisms; you learn about their life cycle, determine their sex, observe phenotypes directly from individual flies, etc. To some extent, you will be reproducing the work you did in the mitosis and meiosis laboratory, but instead of using plastic models you will be using actual organisms and observing phenotypes instead of genotypes. You will complete monohybrid and dihybrid crosses and look at the ratios of phenotypes in the **first filial (F1)** and **second filial (F2) generations** to see whether the chromosomes of the vinegar flies are following Mendel's two laws or not. At this point in your study of genetics, you should already know what to expect from the crosses, but you will be exposed to two new concepts in this lab.

First, you will perform a **sex-linked cross**. Up to this point you have been studying **autosomal genes**. These are genes that are on any chromosome in the organism that is not a sex-determining chromosome. Most diploid organisms have a chromosome system that determines the sex of an organism. In humans, the 23rd chromosome determines sex. If a human has an *X* and a *Y* pair of the 23rd chromosome, he is a male. If the human has a pair of *X*'s for the 23rd chromosome, she is a female. The presence of the *Y* chromosome is crucial for a human to be male. The system

is similar in the *Drosophila*, but not quite the same. Sex is actually determined by the ratio of *X* chromosomes to the number of autosomes (and even this is being questioned). If there are two *X* chromosomes, the fly will be a female, and if there is only one *X* chromosome, the fly will be a male. The presence of a *Y* chromosome is not crucial for determining sex in vinegar flies, although it is crucial for determining sex in humans.

What is important in understanding sex-linked genes is that *X* and *Y* chromosomes are not homologous. There are many more genes on an *X* chromosome than on a *Y* chromosome. If an *X* chromosome is not paired with another *X* chromosome (in other words, if the fly is male), all of the alleles on the *X* chromosome will be expressed as the phenotype. This includes all alleles on the *X* chromosome, whether they are dominant or recessive. Expression of only genes on the *X* chromosome could be lethal for male flies because many recessive lethal alleles occur in heterozygous parents. If a female is a carrier for a recessive lethal allele on one of her *X* chromosomes and the male that she mates with carries the same gene on the *X* chromosome, half of their male progeny will die!

You will most likely explore a sex-linked, recessive gene in this lab, since it will be easier to spot than a sex-linked, dominant gene. Regardless of which organisms you use in this lab or which genes, you can easily focus on the sex-linked gene by looking at the progeny of the cross between the mutant and wild type adults. In the cross of a wild type female with a mutant male, none of the offspring will show the mutant phenotype. All of the males will receive only a *Y* chromosome from the affected male parent and the females will have one wild type *X* chromosome from the mother to be dominant over the recessive *X* allele from the father. The cross between a mutant female and a wild type male will produce offspring with all females being heterozygous for the recessive allele and exhibiting the wild phenotype. All of the males will be mutant because they only possess one *X* chromosome, which they received from the mutant mother.

The second concept you need to know from this lab is the application of **chi-squared analysis.** The chi-squared analysis measures the difference between the number of observations you make that meet your expectations and the number that don't. You put these values into a formula and compare the answer to a table of standards. The formula is:

$$\chi^2 = \Sigma \frac{(Observed - Expected)^2}{Expected}$$

As an example, look at the frequencies observed if you toss a coin 100 times. You observe that the coin comes up heads 52 times and comes up tails 48 times. Since there is an equal likelihood that the coin will come up heads or tails, the expected values for heads and tails are 50 and 50. Putting these values into the formula:

$$\chi^2 = \frac{(52-50)^2}{50} + \frac{(48-50)^2}{50} = 0.16$$

Compare the value 0.16 to a chi-squared table at one **degree of freedom (d.f.)**. One degree of freedom is used because two observations were made and d.f. equals number of categories of observations minus one.

Probability (p)	Degrees of Freedom (d.f.)				
	1	2	3	4	5
0.05	3.84	5.99	7.82	9.49	11.1

At $p = 0.05$ in the chi-squared table, the value obtained from the above equation would have to be at least 3.84 in order for your observations to be statistically different from what is expected. The value calculated was only 0.16, so it can be concluded that although your observations weren't exactly $\frac{50}{50}$, they are not statistically significantly different from the expected outcome. There are many considerations and assumptions to make when performing statistical analyses, which would be covered in greater depth in a class on statistical analyses. For the chi-squared analysis, there are three key points to remember:

- The formula for the chi-squared statistic: $\chi^2 = \Sigma \frac{(Observed - Expected)^2}{Expected}$
- The formula for d.f., d.f. = # of categories of observations − 1
- Your calculated χ^2 must be greater than the corresponding p value in the table in order for the outcome to be significantly different than what you expected.