

# Honors packet Instructions

The following are guidelines in order for you to receive FULL credit for this bio packet:

1. Read and take notes on the packet in full
2. Answer the multiple choice questions correctly and JUSTIFY your answer  
*Example Question: "Which of the following organelles is involved in photosynthesis?"*
  - a) *The mitochondria*
  - b) *The endoplasmic Reticulum*
  - c) *The liver*
  - d) *The chloroplast*

*Example answer: The correct answer is D because the chloroplast is an organelle that contains chlorophyll a pigment directly associated with Absorbing light from the sun. the other organelles listed have other functions throughout the cell, and the liver isn't an organelle at all.*

3. Complete the essay response

**\*\*you do not have to print out the packet in order to take notes and complete the quiz\*\***

See wynn for questions!

# Chapter 5: Classical Genetics

- Meiosis
- Mendelian Genetics
- Non-Mendelian Inheritance Patterns

*Classical genetics* is the study of the patterns and mechanisms of the transmission of inherited traits from one generation to another. The foundations for this field were laid by the monk Gregor Mendel, who in the mid-19th century performed a series of experiments to determine the rules of inheritance in garden pea plants.

The study of classical genetics requires an understanding of *meiosis*, the mechanism of gamete formation. Mendel knew that alleles were inherited from each parent, and that these alleles were somehow linked to the various characteristics he studied in his peas, but it was not until meiosis was truly elucidated that the mechanisms behind heredity were understood.

## MEIOSIS

In asexual reproduction, a single diploid cell (or cells) is used to create new identical copies of an organism. In sexual reproduction, two parents contribute to the genome of the offspring and the end result is genetically unique offspring. This requires that each parent contributes a cell with one copy of the genome. *Meiosis* is the process whereby these sex cells are produced.

As in mitosis, the gametocyte's chromosomes are replicated during the S phase of the cell cycle. The first round of division (*meiosis I*) produces two intermediate daughter cells. The second round of division (*meiosis II*) involves the separation of the sister chromatids, resulting in four genetically distinct haploid gametes. In this way, a diploid cell produces haploid daughter cells. Since meiosis reduces the number of chromosomes in each cell from  $2n$  to  $n$ , it is sometimes called *reductive division*.

### DON'T MIX THESE UP ON TEST DAY

#### Mitosis:

- Produces diploid cells from diploid cells
- Occurs in all somatic cells
- Does not involve the pairing up of homologous chromosomes
- Does not involve crossing over (recombination)

#### Meiosis:

- Produces haploid cells from diploid cells
- Occurs only in sex cells (gametocytes)
- Involves the pairing up of homologous chromosomes at the metaphase plate, forming tetrads
- Involves crossing over

Each meiotic division has the same four stages as mitosis, although it goes through each of them twice (except DNA replication, which only happens once). The stages of meiosis are detailed in the following paragraphs.

### Interphase I

Gametocyte chromosomes are replicated during the S phase of the cell cycle, while the centrioles replicate at some point during interphase.

### Prophase I

During this stage, the chromosomes condense, the spindle apparatus forms, and the nucleoli and nuclear membrane disappear. Homologous chromosomes (matching chromosomes that code for the same traits, one inherited from each parent), come together and intertwine in a process called *synapsis*. Since at this stage each chromosome consists of two sister chromatids, each synaptic pair of homologous chromosomes contains four chromatids, and is therefore often called a *tetrad*.

Sometimes chromatids of homologous chromosomes break at corresponding points and exchange equivalent pieces of DNA; this process is called *crossing over* or *recombination*. Note that crossing over occurs between homologous chromosomes and not between sister chromatids of the same chromosome. The chromatids involved are left with an altered but structurally complete set of genes.

The chromosomes remain joined at points called chiasmata where the crossing over occurred. Such genetic recombination can “unlink” genes that had been on the same chromosome in the parent, thereby increasing the variety of genetic combinations that can be produced via gametogenesis. Recombination among chromosomes results in increased genetic diversity within a species. Note that sister chromatids are no longer identical after recombination has occurred.

### Metaphase I

Homologous pairs (tetrads) align at the equatorial plane of the dividing cells, and each pair attaches to a separate spindle fiber by its kinetochore.

### Anaphase I

Homologous pairs separate and are pulled to opposite poles of the cell. This process is called *disjunction*, and it accounts for a fundamental Mendelian law. During disjunction, each chromosome of paternal origin separates (or disjoins) from its homologue of maternal origin, and either chromosome can end up in either daughter cell. Thus, the distribution of homologous chromosomes to the two intermediate daughter cells is random with respect to parental origin. Each daughter cell will have a unique pool of alleles provided by a random mixture of maternal and paternal origin. These different alleles may code for alternative forms of a given trait.

## Telophase I and Cytokinesis

A nuclear membrane forms around each new nucleus. At this point, each chromosome still consists of sister chromatids joined at the centromere. Next, the cell divides through cytokinesis into two daughter cells, each of which receives a nucleus containing the haploid number of chromosomes. Between cell divisions there may be a short rest period, or interkinesis, during which the chromosomes partially uncoil.

## Prophase II

The centrioles migrate to opposite poles and the spindle apparatus forms.

## Metaphase II

The chromosomes line up along the equatorial plane once again. The centromeres divide, separating the chromosomes into pairs of sister chromatids.

## Anaphase II

The sister chromatids are pulled to opposite poles by the spindle fibers.

## Telophase II and Cytokinesis

Finally, a nuclear membrane forms around each new haploid nucleus. Cytokinesis follows and two daughter cells are formed. Thus, by the time meiosis is complete, four haploid daughter cells are produced per gametocyte. In females, only one of these becomes a functional gamete.

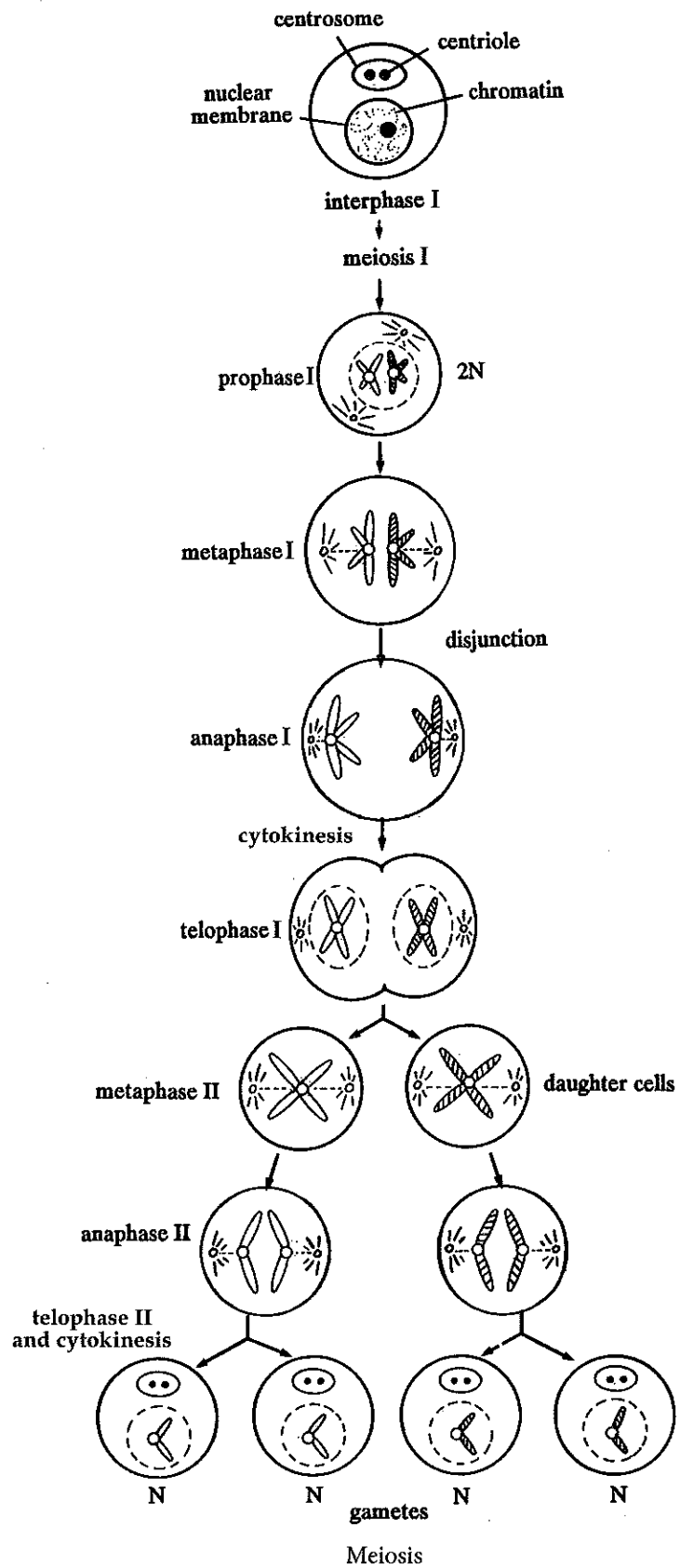
The diagram on the following page summarizes the various stages of meiosis I and meiosis II. Notice that the random distribution of homologous chromosomes in meiosis, coupled with crossing over in prophase I, enables an individual to produce gametes with many different genetic combinations. Every gamete gets one copy of each chromosome, but the copy of each chromosome found in a gamete is random. For example, each gamete has a chromosome #9, but this chromosome can be either of the two copies of this chromosome. With 22 autosomal chromosomes, this factor alone allows for  $2^{22}$  possible gametes, not including the additional genetic diversity created by recombination. This is why sexual reproduction produces genetic variability in offspring, as opposed to asexual reproduction, which produces identical offspring. The possibility of so many different genetic combinations is believed to increase the capability of a species to evolve and adapt to a changing environment.

## MENDELIAN GENETICS

Around 1865, based on his observations of seven characteristics of the garden pea, Gregor Mendel developed the basic principles of genetics—*dominance*, *segregation*, and *independent assortment*. Although Mendel formulated these principles, he was

### THE PROBLEM WITH NONDISJUNCTION

If, during anaphase I or II of meiosis, homologous chromosomes or sister chromatids fail to separate (in what is termed nondisjunction), one of the resulting gametes will have two copies of a particular chromosome and another gamete will have none. Subsequently, during fertilization, the resulting zygote may have one too many or one too few copies of the chromosome in question. Few of these mutated zygotes survive. Those that do survive encounter difficulties associated with conditions like Down syndrome (Trisomy 21).



unable to propose any mechanism for hereditary patterns, since he knew nothing about chromosomes or genes. Hence his work was largely ignored until the early 1900s.

After Mendel's work was rediscovered, Thomas H. Morgan tied the principles of genetics to the chromosome theory. He linked specific traits to regions of specific chromosomes visible in the salivary glands of *Drosophila melanogaster*, the fruit fly. Morgan brought to light the giant chromosomes, at least 100 times the size of normal chromosomes, that are found in the fruit fly's salivary glands. These chromosomes are banded, and the bands coincide with gene locations, allowing geneticists to visibly follow major changes in the fly genome. Morgan also described sex-linked genes.

The fruit fly is a highly suitable organism for genetic research. With its short life cycle, it reproduces often and in large numbers, providing large sample sizes. It is easy to grow in the laboratory, but has a fairly complex body structure. Its chromosomes are large and easily recognizable in size and shape. They are also few in number (eight chromosomes/four pairs of chromosomes). Finally, mutations occur relatively frequently in this organism, allowing genes for the affected traits to be studied.

Some of the basic rules of gene transmission and expression are:

- Genes are elements of DNA that are responsible for observed traits.
- In eukaryotes, genes are found in large linear chromosomes, and each chromosome is a very long continuous DNA double-helix. Humans have 23 different chromosomes, with two copies of each chromosome in somatic cells.
- Each chromosome contains a specific sequence of genes arranged along its length.
- Each gene has a specific location on a chromosome.
- Diploid organisms have two copies of each chromosome and therefore two copies of each gene (except for the X and Y chromosomes in males).
- The two copies of each gene can have a different sequence in an organism and a gene can have several different sequences in a population. These different versions of a gene are called *alleles*.
- The type of alleles an organism has (its genetic composition) is called the *genotype*.
- The appearance and physical expression of genes in an organism is called the *phenotype*.
- Types of alleles include dominant and recessive alleles. A dominant allele is expressed in an organism regardless of the second allele in the organism. The trait encoded by a recessive allele will not be observed as the phenotype if the other allele for the gene an organism carries is a dominant one.
- A *homozygous* individual has two copies (two alleles) of a gene that are identical and a *heterozygous* individual has two different alleles for a gene.
- The phenotype of an individual is determined by the genotype.

### PERFECT PEAS

It is suspected that Mendel might have doctored the results of his experiments, because the data from his pea crosses is almost too perfect.

### FLASHBACK

As discussed in the chapter on cellular and molecular biology, DNA sequence is responsible for genotype, while the protein produced during the translation of mRNA transcribed from the DNA sequence is responsible for phenotype.

### DOMINANT VERSUS RECESSIVE

Dominant alleles are always seen in the phenotype of heterozygotes, while recessive alleles are not.

## Dominance of Phenotypic Traits

If two members of a pure-breeding strain are mated, their offspring will always have the same phenotype as the parents since they are all homozygous for the same allele. What happens if two different pure-breeding strains that are homozygous for two different alleles are crossed? In an example such as two different alleles for flower color, what often occurs is that all of the offspring of the cross match the phenotype of one parent and not the other. For example, if a pure-breeding red strain is crossed with a pure-breeding white one, perhaps all of the offspring are red. Where did the allele coding for the white trait go? Did it disappear from the offspring?

If it is true that both parents contribute one copy of a gene to each of their offspring, then the allele cannot disappear. The offspring must all contain both a white allele and a red allele. Despite having both alleles, however, they exhibit only one—the red allele. Red is then a dominant allele and white a recessive allele, since it is not observed as the phenotype in heterozygotes such as the offspring in this cross of two pure-breeding strains.

Every human has two copies of each of their 23 chromosomes, with the exception of the X and Y chromosome in men. Thus, each gene is present in two copies that can either be the same or different. For example, a gene for eye color could have two alleles: *B* or *b*. *B* is a *dominant allele* for brown eye color and *b* is a *recessive allele* for blue eye color. There are three potential genotypes: *BB*, *Bb*, or *bb*. *BB* individuals and *Bb* individuals have brown eyes, and only *bb* homozygous people have blue eyes. *Bb* people have brown eyes since the *B* allele is dominant and the recessive *b* allele is not observed as the phenotype in the heterozygote.

### DON'T MIX THESE UP ON TEST DAY

#### Test cross:

The breeding of two individuals of the same species to determine, based on the offspring, which of two alleles is dominant for a given trait

#### Punnett square:

A tool used to predict the results of a test cross, by mapping out all the possible combinations of parental alleles that could occur in the offspring

## Test Crosses

Often, a geneticist will study the transmission of a trait in a species such as flies or pea plants by performing crosses (matings) between organisms with defined traits. For example, an investigator may identify two possible phenotypes for flower color in pea plants: pink and white. Pink plants bred together always produce pink offspring and white plants bred together always produce offspring with white flowers. It is likely that the differences in flower color are caused by different alleles in a gene that controls flower color. But which of these traits is determined by a recessive or dominant allele? You cannot tell based on the color alone which trait will be dominant or recessive. Either pink or white could be dominant, or neither.

The way to determine the dominant or recessive nature of each allele is by performing a test cross. Since the pink plants always produce pink plants and the white plants always produce white plants, these are both termed “pure-breeding” plants and are each homozygous for either the *P* allele (*PP* genotype has a pink phenotype) or for the *p* allele (*pp* genotype has a white phenotype). What will be the phenotype of a plant with the *Pp* genotype?

When performing a test cross, a useful tool is called a *Punnett square*. To perform a Punnett square, first determine the possible gametes each parent in the cross can produce. In the example below, a  $PP$  parent can make gametes with either of the two  $P$  alleles and the  $pp$  parent can only make gametes with the  $p$  allele:

$PP$  parent: Gametes have either one  $P$  allele or the other  $P$  allele.

$pp$  parent: Gametes have either one  $p$  allele or the other  $p$  allele.

The next step is to examine all of the ways that these gametes could combine if these two parents were mated together in a test cross. This is where the Punnett square comes in. On one side of the square, align the gametes from one parent, and on the other side of the square align the gametes from the other parent. At the intersection of each potential gamete pairing, fill in the square with the diploid zygote produced by matching the alleles. In this example, all of the offspring of this cross are going to be heterozygous.

If all of the offspring are pink, what does this reveal about the nature of these alleles? If the heterozygous  $Pp$  plant has the same phenotype as the homozygous  $PP$  plant, then the  $P$  allele is dominant over the  $p$  allele. If the  $p$  allele is not exhibited as the phenotype in the heterozygote, the  $p$  allele is recessive and the  $P$  allele is dominant. The offspring of this cross (shown within the box) can be called the  $F_1$  generation.

A cross between two pure-breeding strains ( $F_1$  generation):

	$P$	$P$
$p$	$Pp$	$Pp$
$p$	$Pp$	$Pp$

The  $F_1$  offspring all have the  $Pp$  genotype and the pink phenotype. What will occur if two of these  $F_1$  plants are crossed? A Punnett square can be used again to predict the genotypes in the  $F_2$  generation.

Parent 1:  $P$  and  $p$  gametes are produced.

Parent 2:  $P$  and  $p$  gametes are produced.

$F_2$  generation Punnett square:

	$P$	$p$
$P$	$PP$	$Pp$
$p$	$Pp$	$pp$

#### PUNNETT SQUARE

A Punnett square, as shown in the text at left, is a useful tool. It provides a quick way to determine the probable traits of offspring produced from particular crosses.



Since we know that the  $P$  allele for pink is dominant, we can use the genotypes to predict phenotypes of the  $F_2$  generation.  $PP$  homozygotes will be pink, and  $Pp$  heterozygotes will also be pink since  $P$  is dominant.  $pp$  plants will be white like the original pure-breeding white plants. Filling in the square above with these phenotypes:

	$P$	$p$
$P$	$PP$ (pink)	$Pp$ (pink)
$p$	$Pp$ (pink)	$pp$ (white)

The ratios of the different genotypes and phenotypes in the Punnett square should match the statistical probability of producing these in real life by a cross of this type. For example, if two heterozygous  $Pp$  plants are crossed, 75 percent of the offspring will be pink and 25 percent white. This is predicted from the Punnett square based on the ratio of 3:1 for phenotypes that will produce pink (3) to white (1).

The behavior of different pea plant traits helped Mendel to formulate two fundamental rules of Mendelian genetics, the Law of Segregation and the Law of Independent Assortment. Mendel derived these rules based purely on his knowledge of the transmission of traits, without knowing anything about the molecular basis for his observations in the mechanisms of meiosis.

### MENDEL'S LAWS

Mendel's laws are:

- Law of Segregation
- Law of Independent Assortment

The dominance of phenotypic traits is also sometimes referred to as one of Mendel's laws, the Law of Dominance.

### Law of Segregation

The Law of Segregation states that if there are two alleles in an individual that determine a trait, these two alleles will separate during gamete formation and can act independently. For example, when a heterozygous  $Pp$  plant is forming gametes, the  $P$  and the  $p$  alleles can separate into different gametes and act independently during a cross. If this was not the case, and the  $P$  and  $p$  alleles could not separate, then all of the offspring would remain  $Pp$  and all of the  $F_2$  would be pink. The fact that white offspring are produced indicates that alleles do indeed segregate into gametes independently. The molecular basis for this observation is that during meiosis, each homologous chromosome carrying the two different alleles will end up in a different haploid gamete.

### Law of Independent Assortment

The Law of Independent Assortment describes the relation between different genes. If the gene that determines plant height is on a different chromosome than the gene for flower color, then these traits will act independently during test crosses. In this example, the two alleles for tallness are the dominant  $T$  allele for tall plants and the recessive  $t$  allele for short plants. The two alleles for color are the dominant  $Y$  allele for yellow and the recessive  $y$  allele for green. When plants are crossed, the alleles for the tall gene act independently of the alleles for the color gene.

Example of a dihybrid cross in which tall and yellow are both hybrids (heterozygotes):

	<i>TY</i>	<i>Ty</i>	<i>tY</i>	<i>ty</i>
<i>TY</i>	<i>TTY</i> <i>Y</i>	<i>TTY</i> <i>y</i>	<i>TtY</i> <i>Y</i>	<i>TtY</i> <i>y</i>
<i>Ty</i>	<i>TTY</i> <i>y</i>	<i>TTy</i> <i>y</i>	<i>TtY</i> <i>y</i>	<i>Tty</i> <i>y</i>
<i>tY</i>	<i>TtY</i> <i>Y</i>	<i>TtY</i> <i>y</i>	<i>ttY</i> <i>Y</i>	<i>ttY</i> <i>y</i>
<i>ty</i>	<i>TtY</i> <i>y</i>	<i>Tty</i> <i>y</i>	<i>ttY</i> <i>y</i>	<i>tty</i> <i>y</i>

Results of the cross:

Phenotype ratio:

9 tall yellow ( $\frac{9}{16}$ ): 3 tall green ( $\frac{3}{16}$ ): 3 short yellow ( $\frac{3}{16}$ ): 1 short green: ( $\frac{1}{16}$ ).

The simplest approach to an independent assortment problem is to consider each of the genes separately, determine the predicted Mendelian ratios for each of the traits alone, and then use the laws of probability to combine these. For example, in the cross above, the predicted Mendelian phenotype ratios are  $\frac{3}{4}$  for tall and  $\frac{1}{4}$  for green. The probability of observing these phenotypes together is the product of their independent probabilities—that is,  $\frac{3}{4} \times \frac{1}{4}$ , or  $\frac{3}{16}$ . A significant variation from this ratio indicates linkage and a failure to assort independently.

## Linkage

There is a significant exception to the Law of Independent Assortment. For genes to assort independently into gametes during meiosis, they must be on different chromosomes. If two genes are located near each other on the same chromosome, then the alleles for these genes will stay together during meiosis. This phenomenon in which alleles fail to assort independently because they are on the same chromosome is called *linkage*.

Another factor that affects linkage is the recombination between homologous chromosomes that occurs during meiosis. Even if two genes are on the same chromosome, they will not necessarily be linked. If recombination occurs in the DNA between the two genes in the chromosome, then this will tend to reduce the linkage between genes. The further apart the genes, the more recombination that will occur between them and the less linkage that will be observed. If the genes are far enough apart on the chromosome, then recombination between the genes may be so frequent that they will display almost no linkage and will assort independently despite being located on the same chromosome.

The fact that the linkage between genes is related to their distance from each other can be used to map the position of genes relative to each other on a chromosome. By performing a test cross and counting the number of offspring with those different phenotypes encoded by the two genes, a geneticist can determine how many recombination events occurred between the genes and from this can estimate the distance of two genes from each other.

### THREE TO ONE

Note that in the dihybrid cross under discussion here, each trait assorts individually in a 3:1 ratio, as is generally the case in a monohybrid cross. There are 9 tall yellow and 3 tall green for a total of 12 tall. There are also 3 short yellow and 1 short green, which amounts to a total of 4 short. Hence both the tall:short ratio (12:4) and the yellow:green ratio are 3:1.

### SEGREGATION VERSUS ASSORTMENT

*Law of Segregation*—two alleles of a gene separate in meiosis

*Law of Independent Assortment*—alleles of different genes are independent during meiosis

**STUDY TIP**

When faced with a difficult pedigree on your Biology E/M exam, remember that recessive phenotypes can only have one possible genotype—homozygous recessive.

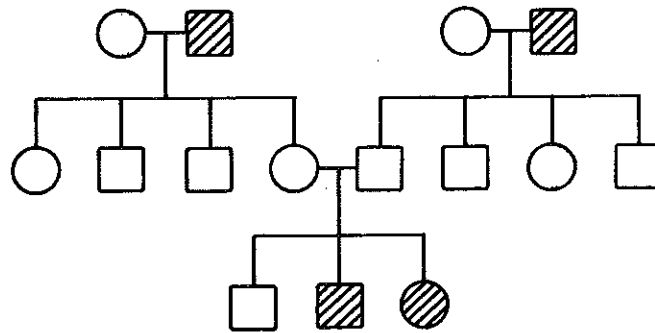
**Inheritance Patterns**

Ethical restraints forbid geneticists to perform test crosses in human populations. Instead, they must rely on examining matings that have already occurred, using tools such as pedigrees. A *pedigree* is a family tree depicting the inheritance of a particular genetic trait over several generations. By convention, males are indicated by squares and females by circles. Matings are indicated by horizontal lines, and descendants are listed below matings, connected by a vertical line. Individuals affected by the trait are generally shaded, while unaffected individuals are unshaded. When carriers of sex-linked traits have been identified (typically, female heterozygotes), they are usually half-shaded in family trees.

The following pedigrees illustrate two types of heritable traits: recessive disorders and sex-linked disorders. When analyzing a pedigree, look for individuals with the recessive phenotype. Such individuals have only one possible genotype—homozygous recessive. Matings between them and the dominant phenotype behave as test crosses; the ratio of phenotypes among the offspring allows deduction of the dominant genotype. In any case in which only males are affected, sex linkage should be suspected.

**Recessive Disorders**

Note how the trait skips a generation in the autosomal recessive disorder depicted in the figure. Albinism is an example of this form of disorder.



Recessive Disorder

**Sex-Linked Disorders**

Gender skewing is evident in this type of disorder, which includes traits such as hemophilia. Sex-linked recessive alleles are most often expressed only by males and transmitted from one generation to another by female carriers.

**QUICK QUIZ**

Match each of the following numbered traits with the correct lettered definition.

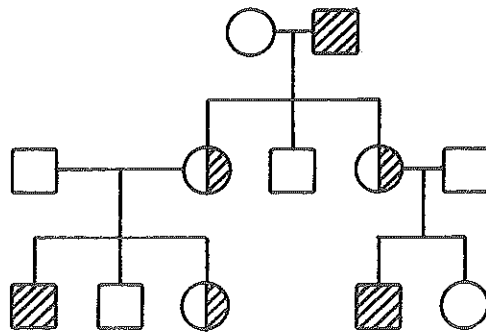
1. Recessive trait
  2. Sex-linked recessive trait
  3. Dominant trait
- (A) Appears in every generation
- (B) Skips generations
- (C) Is almost always found in males

3. (A)

2. (C)

1. (B)

Answers:



Sex-linked Disorder

### MATERNAL INHERITANCE

Genes carried on mitochondria always pass in ova from a mother to all of her children, male or female.

## NON-MENDELIAN INHERITANCE PATTERNS

While Mendel's laws hold true in many cases, these laws cannot explain the results of certain crosses. Sometimes an allele is only incompletely dominant or, perhaps, codominant. The genetics that enable the human species to have two genders would also not be possible under Mendel's laws.

### Incomplete Dominance

Incomplete dominance is a blending of the effects of contrasting alleles. Both alleles are expressed partially, neither dominating the other.

An example of incomplete dominance is found in the four-o'clock plant and in the snapdragon flower. When a red flower ( $F_R F_R$ ) is crossed with a white flower ( $F_W F_W$ ), a pink blend ( $F_R F_W$ ) is created. When two pink flowers are crossed, the yield is 25 percent red, 50 percent pink, and 25 percent white (phenotypic and genotypic ratio 1:2:1).

### Codominance

In codominance, both alleles are fully expressed without one allele dominant over the other. An example is blood types. Blood type is determined by the expression of antigen proteins on the surface of red blood cells. The  $A$  allele and the  $B$  allele are codominant if both are present, and combine to produce  $AB$  blood.

The allele for blood type  $A$ ,  $I_A$ , and the allele for blood type  $B$ ,  $I_B$ , are both dominant to the third allele,  $i$ .  $I_A$  and  $I_B$  may appear together to form blood type  $AB$ ; however, when both are absent, blood type  $O$  results.

To summarize:

- $I_A$  = gene for producing antigen A on the red blood cell
- $I_B$  = gene for producing antigen B on the red blood cell
- $i$  = recessive gene; does not produce either antigen

### DON'T MIX THESE UP ON TEST DAY

In *incomplete dominance*, two traits are blended together; both are partially expressed, and neither dominates.

In *codominance*, both traits are fully expressed; neither dominates.

And these genes combine in various ways to form the following possible genotypes and blood types (phenotypes):

- $I_A I_A$  or  $I_A i$  = type A blood
- $I_B I_B$  or  $I_B i$  = type B blood
- $I_A I_B$  = type AB blood, with A and B alleles codominant
- $ii$  = type O blood

## Sex Determination

Most organisms have two types of chromosomes: *autosomes*, which determine most of the organism's body characteristics; and *sex chromosomes*, which determine the sex of the organism. Humans have 22 pairs of autosomes and one pair of sex chromosomes. The sex chromosomes are known as X or Y. In humans, XX is present in females and XY in males. The Y chromosome carries very few genes. Sex is determined at the time of fertilization by the type of sperm fertilizing the egg, since all eggs contain X chromosomes only. If the sperm carries an X chromosome, the offspring will be female (XX); if the sperm carries a Y chromosome, the offspring will be male (XY).

This process is illustrated in the Punnett square below:

		X	Y	(From the father)
	X	XX	XY	
(From the mother)	X	XX	XY	

The ratio of the sex of the offspring is 1:1.

## Sex Linkage

Genes for certain traits, such as color blindness or hemophilia, are located on the X chromosomes. Hence these genes are linked with the genes controlling sex determination. These genes seem to have no corresponding allele on the Y chromosome, with the result that the X chromosome contributed by the mother is the sole determinant of these traits in males. Genes determining hemophilia and red-green color blindness are sex-linked (on the X chromosome). They are recessive, implying that they can be hidden by a dominant normal allele on the other X chromosome in a female. For this reason, the female with two X chromosomes may carry, but will rarely exhibit, these afflictions. The male, on the other hand, with his Y chromosome, has no dominant allele to mask the recessive gene on his X chromosome. As a consequence of having a single copy of X-linked genes, males exhibit sex-linked traits much more frequently than females do.

**Cross 1:** Let's see what happens when we cross a hemophilia-carrying female and a normal male:

$XX_h \times XY$ :

	X	$X_h$
X	XX	$XX_h$
Y	XY	$X_hY$

*Results of the cross:*

XX = healthy female

$XX_h$  = carrier but healthy female

XY = healthy male

$X_hY$  = hemophiliac male

There are no male carriers of this trait, since all males that have the hemophilia allele express it.

**Cross 2:** Here's a cross between a carrier female and a male hemophiliac:

$XX_h \times X_hY$ :

	X	$X_h$
$X_h$	$XX_h$	$X_hX_h$
Y	XY	$X_hY$

*Results of the cross:*

$X_hX_h$  = hemophiliac female (very rare)

$XX_h$  = carrier but healthy female

XY = healthy male

$X_hY$  = hemophiliac male

## Mutations

Mutations can create new alleles, the raw material that drives evolution via natural selection. Mutations are changes in the genes that are inherited. To be transmitted to the succeeding generation, mutations must occur in sex cells—eggs and sperm—rather than somatic cells (body cells). Mutations in nonsex cells are called somatic cell mutations and affect only the individual involved, not subsequent generations. A somatic mutation can cause cancer, but will have no effect on offspring since it is not present in gametes. Most mutations are recessive. Because they are recessive, these mutations can be masked or hidden by the dominant normal genes.

### FLASHBACK

DNA mutations can occur in two ways:

- Some point mutations involve changes in single nucleotide bases in the DNA sequence
- Other mutations are called *frame-shift mutations* and involve the insertion or deletion of nucleotides, changing the reading frame of the protein

## Chromosomal Mutations

These mutations result in changes in chromosome structure or abnormal chromosome duplication. In crossing over, segments of chromosomes switch positions during meiotic synapsis. This process breaks linkage patterns normally observed when the genes are on the same chromosome. A translocation is an event in which a piece of a chromosome breaks off and rejoins a different chromosome.

*Nondisjunction* is the failure of some homologous pairs of chromosomes to separate following meiotic synapsis. The result is an extra chromosome or a missing chromosome for a given pair. For example, Down syndrome is due to an extra chromosome #21 (Trisomy 21). The number of chromosomes in a case of single nondisjunction is  $2n + 1$  or  $2n - 1$ . In Trisomy 21, the individual has 47 chromosomes instead of the usual 46.

### THE TWO FACES OF MUTATIONS

Mutations may be beneficial (enabling a population to develop new adaptations to the changing environment and leading to evolution of a species). Yet they can also damage the organisms in which they occur, sometimes fatally. Mutations are the root cause of diseases such as cancer. The vast majority of mutations are probably neutral, in terms of an organism's fitness.

*Polyploidy* ( $3n$  or  $4n$ ) involves a failure of meiosis during the formation of the gametes. The resulting gametes are  $2n$ . Fertilization can then be either  $n + 2n = 3n$  or  $2n + 2n = 4n$ . Polyploidy is always lethal in humans, although it is often found in fish and plants. Finally, *chromosome breakage* might be induced by environmental factors or mutagenic drugs.

## Gene Mutations

As discussed in the chapter on cellular and molecular biology, there might be changes in the base sequence of DNA that result in changes in single genes, changing one or more base pairs and the protein produced by transcribing and translating the gene.

## Mutagenic Agents

Mutagenic agents, which induce mutations, include ultraviolet (UV) light, X-rays, radioactivity, and some chemicals that cause mutations by damaging DNA. Such agents are also typically carcinogenic, causing cancer.

This concludes the chapter on classical genetics. With the knowledge you have gained here, you should be able to look at your own family and study the heritability of certain traits. For example, if you have blue eyes, a recessive trait, you must be homozygous for that trait; each of your parents gave you an allele for blue eyes. If one of your parents has brown eyes, he or she must be a heterozygote, possessing alleles for both blue and brown eyes.

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## THINGS TO REMEMBER

- Stages of meiosis
  - Homologous chromosomes
  - DNA recombination
- Genotype and phenotype
- Alleles
- Mendel's laws of genetics
  - Dominance
  - Segregation
  - Independent Assortment
- Non-Mendelian inheritance patterns
  - Incomplete dominance
  - Codominance
  - Sex-linked traits
  - Mutation



## CLASSICAL GENETICS QUIZ

- Breeding animals closely related in a pedigree is known as
  - inbreeding.
  - codominance.
  - linkage analysis.
  - selective breeding.
  - test crossing.
- A process that CANNOT take place in haploid cells is
  - mitosis.
  - meiosis.
  - cell division.
  - growth.
  - digestion.
- |   |   |   |   |
|---|---|---|---|
| A | B | C | D |
|   |   |   |   |

If the diagram above represents genes on a chromosome, which genes would have the highest frequency of recombination between them?

  - A and B
  - A and D
  - B and C
  - B and D
  - The frequencies are the same for all crossovers
- Laboratory mice are to be classified based on genes *A*, *B*, and *C*. How many genetically different gametes can be formed by a mouse that is genotypically *AaBbCc*? (Assume that none of these is a lethal gene.)
  - 3
  - 6
  - 8
  - 9
  - 12
- An individual with type O blood must have which of the following?
  - Multiple alleles at the blood type gene
  - Several different genes that control blood type
  - Homozygous recessive alleles
  - Selection against other types that delete the gene
  - Spontaneous mutations
- The gene for red-green color blindness is located on the X chromosome. The offspring of a man suffering from red-green color blindness would have which of the following characteristics if he married a normal homozygous female?
  - 50% of the females would be carriers; 100% of the males would be affected
  - 100% of the females would be normal; 50% of the males would be affected
  - 100% of the females would be carriers; 100% of the males would be normal
  - 50% of the females would be affected; 100% of the males would be affected
  - 100% of the females would be normal; 50% of the males would be carriers

7. A mutation in a gene in a somatic cell is deleterious because
- (A) it will affect gamete formation.
  - (B) it will be dominant.
  - (C) it may be passed on to subsequent generations.
  - (D) it may lead to a tumor in that tissue.
  - (E) None of the above
8. Which of the following genetic mutations will NEVER affect the protein produced?
- (A) Point
  - (B) Silent
  - (C) Insertion
  - (D) Frame shift
  - (E) All of the above
9. Tall is dominant over short in a certain plant. A tall plant was crossed with a short plant, and both tall and short offspring were produced. This demonstrates
- (A) the Law of Segregation.
  - (B) incomplete dominance.
  - (C) linkage.
  - (D) mutation.
  - (E) the Law of Independent Assortment.
10. A typical human gamete
- (A) contains a haploid number of genes.
  - (B) always contains an X or Y chromosome.
  - (C) is a result of the meiotic process.
  - (D) has genetic material that has undergone recombination.
  - (E) All of the above
11. Spermatogenesis and oogenesis differ in that
- (A) spermatogenesis is mitotic while oogenesis is meiotic.
  - (B) oogenesis is mitotic while spermatogenesis is meiotic.
  - (C) spermatogenesis produces gametes while oogenesis does not.
  - (D) spermatogenesis produces four haploid sperm cells while oogenesis produces one egg cell and more than one polar body.
  - (E) spermatogenesis involves unequal division of cytoplasm.
12. If a male with blood type A marries a female with blood type B, which of the following types would be impossible for a first-generation child?
- (A) Type B
  - (B) Type A
  - (C) Type O
  - (D) Type AB
  - (E) All types are possible
13. Polar bodies are formed during
- (A) male mitosis.
  - (B) female mitosis.
  - (C) male meiosis.
  - (D) female meiosis.
  - (E) Two of the above

14. Red is dominant over white in a certain flower. To test whether a red offspring is homozygous or heterozygous in this flower, one would
- (A) cross it with a red plant that had a white parent.
  - (B) cross it with a red plant that had two red parents.
  - (C) cross it with a white plant.
  - (D) Two of the above will work
  - (E) None of the above will work
15. Green ( $Y$ ) is dominant over yellow ( $y$ ) in peas, and the smooth allele ( $W$ ) is dominant over wrinkled ( $w$ ). Which cross must produce all green, smooth peas?
- (A)  $YyWw \times YyWw$
  - (B)  $Yyww \times YYWw$
  - (C)  $YyWW \times yyWW$
  - (D)  $YyWw \times YYWW$
  - (E) None of the above
16. Unequal division of the cytoplasm occurs in the
- (A) production of sperm cells.
  - (B) production of egg cells.
  - (C) mitosis of an epidermal cell.
  - (D) binary fission in bacteria.
  - (E) None of the above
17. Disjunction is the process whereby
- (A) homologous chromosomes separate into two cells.
  - (B) homologous pairs of chromosomes recombine.
  - (C) the spindle apparatus is formed from the centrioles.
  - (D) the cell membrane invaginates to form two daughter cells.
  - (E) None of the above

## ANSWERS AND EXPLANATIONS

### 1. (A)

Inbreeding occurs when animals that are closely related are bred to create progeny. Codominance (B) is a pattern of allelic expression. Linkage analysis (C) is the study of recombination between genes to map their positions. Selective breeding (D) is defined as the creation of certain strains of specific traits through controlled breeding, while test crossing (E) is the mating of an organism with a homozygous recessive in order to determine whether that organism is homozygous dominant or heterozygous dominant for a given trait.

### 2. (B)

A cell that is  $n$  (haploid) cannot undergo meiosis to become  $\frac{1}{2}n$ . (A), (C), and (D) are incorrect because there are a number of organisms that are haploid. These organisms undergo mitosis, divide, and grow. Meanwhile, (E) is incorrect because an organism, whether it is diploid or haploid, must be able to digest to maintain life.

### 3. (B)

Homologous recombination occurs during metaphase I tetrad formation. The farther apart two genes are, the more likely it is that a homologous recombination will occur between them. Therefore, the genes that are farthest apart are also those most likely to have recombination occur between them. It is also important to note that the farther away from the centromere the genes are, the more likely they are to recombine.

### 4. (C)

The gametes that can be formed by the mouse in this case are  $ABC$ ,  $ABc$ ,  $AbC$ ,  $Abc$ ,  $aBC$ ,  $aBc$ ,  $abC$ , and  $abc$ . To calculate, multiply the number of alleles at each gene:  $2 \times 2 \times 2 = 8$  possible gametes.

### 5. (C)

Type O blood is found in people with two recessive alleles that express neither the A or B antigen.

### 6. (C)

A male affected with red-green color blindness would have a genotype of  $X_{cb}Y$ . If he mated with a normal female,  $XX$ , all their female offspring would be  $X_{cb}X$ , receiving one good copy of the X chromosome from their mother and the color blindness gene from their father. Since color blindness is a recessive trait, all the female offspring would be carriers. All the male offspring of this mating would be  $XY$ , receiving one good copy of the X chromosome from their mother and the Y chromosome from their father.

### 7. (D)

Mutations in somatic cells (cells of the body, not germ tissue) affect only the individuals involved. They cannot be passed on to the next generation and will not affect gamete formation. These mutations are typically recessive, although there are some instances of dominant negative mutations. The major concern about mutations in somatic cells is that they are linked to the development of tumors; these are produced by proteins that have lost their functions or gained cancerous functions due to somatic mutation.

### 8. (B)

A silent mutation is a point mutation that either occurs in a noncoding region or does not change the amino acid sequence, due to degeneracy of the genetic code. Therefore, silent mutations by definition do not affect the protein produced. Point mutations (A), meanwhile, occur when a single nucleotide base is substituted for another nucleotide base which can change a codon to a different amino acid. A frame shift mutation (D) is either an insertion or deletion of a number of nucleotides. These mutations have serious effects on the encoded protein, since nucleotides are read as series of triplets. The addition or loss of nucleotides (except in multiples of three) will change the reading frame of the mRNA.

### 9. (A)

The Law of Segregation states that when gametes are formed, the two alleles for a particular trait will separate or segregate into the gametes, so that each of

the gametes contains only one of the alleles for a given trait. So if tall is dominant over short, and both tall and short offspring were produced, then the tall plant is a hybrid or heterozygous plant. This means that the genotype of the tall plant contains both one tall allele and one short allele. The short plant contains two short alleles. When the gametes are formed for this mating, the two alleles in the tall plant, the tall and short, will segregate into the gametes, forming tall-containing gametes and short-containing gametes. When these meet and fertilize the short-containing gametes from the other plant, half the offspring produced will be tall because they are the result of the tall gamete's fertilization of a short gamete, and the other half will be the result of the short gamete's fertilization of the other short gamete.

In (B), incomplete dominance, or blending, occurs when two individuals mate and the resulting offspring is a phenotype that appears to be midway between the two phenotypes of the two parents. For example, if a tall and short plant were crossed in this case and they produced an offspring of medium height, the result would be incomplete dominance. As for (C), linkage refers to genes or alleles that travel with each other on the same chromosome. Hence linked genes cannot segregate into two separate gametes because they are near each other on the same chromosome. (D) is not correct either; mutation refers to changes in the DNA sequence of a chromosome, and there is no evidence of mutation occurring in this question. Finally, in (E), the Law of Independent Assortment states that when we are dealing with more than one trait at a time and these traits are not linked, they are carried on different chromosomes or are far from each other on the same chromosome, and the inheritance of these traits is not connected.

**10. (E)**

During meiosis, the gamete reduces its genetic component from  $2n$  to  $n$ , resulting in a haploid cell with half the normal chromosome number. When a haploid egg and sperm unite, they form a diploid organism known as a zygote. All ova will contain an X chromosome, and all sperm will contain either an X or a Y chromosome. These gametes are formed during the two reductional divisions of meiosis. During metaphase I

of meiosis I, tetrads form and sister chromatids undergo a homologous recombination known as crossing over.

**11. (D)**

Spermatogenesis and oogenesis are both forms of gametogenesis in which haploid gametes are produced through reductional division (meiosis) of diploid cells. Both processes occur in the gonads. They differ, however, in that in spermatogenesis, the cytoplasm is equally divided during meiosis and four viable sperm are produced from one diploid cell. In oogenesis, the cytoplasm is divided unequally and only one ovum, containing the bulk of the cytoplasm, is produced, along with two or three inert polar bodies.

**12. (E)**

The type A man can be either  $AA$  or  $Ai$  and the type B woman can be either  $BB$  or  $Bi$ . A and B blood groups are codominant over blood antigen O. Therefore, if a man heterozygous for blood type A ( $Ai$ ) married a woman heterozygous for blood type B ( $Bi$ ), they could have children with the possible genotypes  $Ai$ ,  $Bi$ ,  $ii$ , or  $AB$ . Therefore, all blood types are possible in this mating.

**13. (D)**

Polar bodies are nonfunctional, gamete-like cells that are formed during female meiosis. Recall that meiosis is a two-stage process. There are a total of four haploid cells formed from each original diploid germ cell. In the case of sperm cells, four functional haploid gamete sperm cells are formed. In the case of egg cells, the first meiotic division involves an unequal division of cytoplasm, resulting in the formation of one large cell and one small cell. The large cell will go on to divide again, while the smaller cell is known as the first polar body. This small-cell polar body may then divide again to form two other polar bodies. The large cell, meanwhile, undergoes a second meiotic division, again unequally dividing the cytoplasm into one larger resulting cell, the final haploid ovum, and another small cell, which again becomes a polar body. Hence it is possible that during a female meiotic division, one large ovum or functional egg cell and three polar bodies may be formed. Polar bodies are not formed in mitosis, which is how all other cell divisions occur in the body.

**14. (D)**

This question illustrates two test crosses that will determine whether the red plant is homozygous or heterozygous. A test cross is performed to determine if a particular dominant individual's phenotype is caused by a homozygous or heterozygous genotype. In this case, there are two possible red genotypes:  $RR$ , the pure homozygous red, and  $Rr$ , the hybrid heterozygous red. These two individuals would have the same phenotype. In order to determine the genotype, the unknown red plant would be mated with a recessive white plant (C) or a known heterozygous red plant (A). If the red plant and the white plant produce only red offspring, then it can be assumed that the original red plant is homozygous. Similarly, if we mate the unknown red plant with a known heterozygous one and produce only red offspring, then the original plant is homozygous. If the two matings of the unknown red organism produce any white offspring at all, then we know that the original unknown red was heterozygous. This is because white offspring can be produced only through the production of one white gamete by each parent.

**15. (D)**

Both green and smooth are dominant phenotypes. The goal in this question is to produce only green, smooth peas, so we want only dominant phenotype offspring. Therefore, we must avoid any crossing that may result in the mating or combining of two recessive alleles. In (A), crossing  $Yy$  and  $Yy$  could result in approximately one-quarter of the offspring turning out yellow. Similarly, in (B),  $ww$  crossed with  $Ww$  would produce offspring of which approximately half would possess a wrinkled phenotype. In (C),  $Yy$  crossed with  $yy$  is likely to produce offspring which are approximately half yellow. (D) is correct because one of the parents is a double dominant, meaning that all offspring will have

the dominant phenotype, regardless of the genotype of the other parent.

**16. (B)**

Unequal cytoplasm division occurs when egg cells are produced during the meiotic process of oogenesis. In the first stage of egg production, the precursor diploid cell produces two daughter cells, but one of the daughter cells receives almost the entire amount of cytoplasm while the other becomes a nonfunctioning polar body. In the second division of oogenesis, the large daughter cell divides again, and once again one of the new daughter cells receives almost all of the cytoplasm and the other becomes a small nonfunctional polar body. The original first polar body may also divide to form two nonfunctional polar bodies. The final result is a potential four haploid cells, but only one of them—the one that received a greater amount of cytoplasm during each meiotic division—becomes a functional egg cell. (A) is incorrect; during spermatogenesis, one diploid precursor cell forms four functional haploid sperm cells. In this case, both divisions are equal and all sperm cells are equal in amount of cytoplasm. As for (C), during mitosis of epidermal cells, cytoplasm is distributed equally. Likewise, in binary fission of bacteria (D), in which bacterial cells are dividing as a means of reproduction, cytoplasmic division will be equal.

**17. (A)**

Disjunction is the separation of homologous chromosomes during meiosis. Each tetrad is separated into two halves. One of each pair of chromosomes (each containing two chromatids) is pulled to opposite ends of the cell. Note that some of the maternal chromosomes can go to one end and some to the other end of the cell. The distribution of homologous chromosomes between the two resultant nuclei is random.



# Chapter 7: Evolution and Diversity

- Evidence of Evolution
- Mechanisms of Evolution
- Classification and Taxonomy

On your SAT Subject Test, you'll probably be faced with quite a few questions dealing with the evolution and classification of the millions of species on Earth. This chapter gives the background you need to ace these questions. It covers topics ranging from types of evidence for evolution to the taxonomic classification of various common species.

## EVIDENCE OF EVOLUTION

Evolution provides a sweeping framework for the understanding of the diversity of life on earth. Living systems, from the cell to the organism to the ecosystem, arose through a long process through geologic time, selecting solutions out of diverse possibilities. What is the evidence that supports the evolutionary view of life? The evidence takes several forms.

### The Fossil Record

Fossils are preserved impressions or remains in rocks of living organisms from the past. Fossils provide some of the most direct and compelling evidence of evolutionary change and are generally found in sedimentary rock. When animals settle in sediments after death, their remains can be embedded in the sediment. These sediments then might be covered over with additional layers of sediment that turn to rock through heat and pressure over many millions of years. The embedded remains turn to stone, replaced with minerals that preserve an impression of the form of the organism, often in a quite detailed state. Most fossils are of the hard, bony parts of animals, since these are preserved the most easily. Fossils of soft body parts or of invertebrates are much more unusual, probably since these parts usually decay before fossil formation can



occur. In some cases, however, it appears that animals died in anaerobic sediments that prevented decay, resulting in soft-body fossils.

One question that must be answered about fossils when they are discovered is their age, to place the fossil correctly in the timeline of life on earth. One way to determine the age is to compare the location of the fossil sediment to other sedimentary rock formations in which the age is already known. Dating using *radioactive decay* is also very useful. Carbon dating is frequently used for material that is only a few thousand years old, but cannot be used for older material since the decay rate of carbon is too rapid.

The conditions for fossil formation are relatively particular, especially for the preservation of invertebrates or soft body parts. Scientists locate fossils by luck, and overall can only look at a tiny percentage of possible fossil locations. They have, over time, located a great variety of fossils, including fossils that provide a clear story for the evolution of modern species. *Archaeopteryx*, for instance, is a primitive bird that represents an intermediate step in the evolution of birds from dinosaurs. It has birdlike features such as feathers and wings, but also dinosaurian features such as a tail and teeth. Changes in fossils over time have revealed a great deal about evolution and provided insight into the evolutionary paths that resulted in modern species, including horses, whales, and humans. Any of the so-called "gaps" in the fossil record are probably the result of scarcity of fossils and difficulty in finding them, and is not evidence that evolution did not occur.

## Comparative Anatomy

One way to find the evolutionary relationship between organisms is by examining their external and internal anatomy. Animals that evolved from a common ancestor might be expected to have anatomical features in common that they share with their common ancestor. Alternatively, two organisms might share features that look the same, but they evolved from different ancestors and acquired in similar structures as a result of similar functions. When we compare the anatomies of two or more living organisms, we can not only form hypotheses about their common ancestors, but we can also glean clues that shed light upon the selective pressures that led to the development of certain adaptations, such as the ability to fly. Comparative anatomists study *homologous* and *analogous structures* in organisms.

**Homologous structures.** Homologous structures have the same basic anatomical features and evolutionary origins but may perform different functions. They demonstrate similar evolutionary patterns with relatively late divergence of form due to differences in exposure to evolutionary forces. Examples of homologous structures include the wings of a bat, the flippers of a whale, the forelegs of a horse, and the arms of a human. These structures were all derived from a common ancestor but diverged to perform different functions in what is termed *divergent evolution*.

**Analogous Structures.** Analogous structures have similar functions but may have different evolutionary origins and entirely different patterns of development. The wings of a fly (membranous) and the wings of a bird (bony and covered in feathers) are analogous structures that have evolved to perform a similar function—to fly. The wings

### DON'T MIX THESE UP ON TEST DAY

*Homologous* structures share a common ancestry.

*Analogous* structures are not inherited from a common ancestor but perform similar functions.

of flies and birds might look similar but this does not indicate that they share a winged ancestor. The evolution of structures that look the same for a common function but are not derived from a common ancestor is called *convergent evolution*. Analogous organs demonstrate superficial resemblances that cannot be used as a basis for classification.

## Comparative Embryology

Comparison of embryonic structures and routes of embryo development is another way to determine evolutionary relationships. The development of the human embryo is very similar to the development of other vertebrate embryos. Adult tunicates (sea squirts) and amphibians lack a notochord, one of the key traits of the chordate phylum, but their embryos both possess notochords during development, indicating that these animals are in fact vertebrates with a common evolutionary ancestor, even though the adults do not resemble each other. The earlier that embryonic development diverges, the more dissimilar the mature organisms are. Thus, it is difficult to differentiate between the embryo of a human and that of an ape until relatively late in the development of each embryo, while human and sea urchin embryos can be differentiated much earlier.

Other embryonic evidence of evolution includes such characteristics as teeth in an avian embryo (recalling its reptile ancestry); the resemblance of the larvae of some mollusks (shellfish) to annelids (segmented worms), and the tail of the human embryo (indicating relationships to other mammals).

## Molecular Evolution

If organisms are derived from a common ancestor, this should be evident not just at the anatomical level but also at the molecular level. The traits that distinguish one organism from another are ultimately derived from differences in genes. With the advent of molecular biology, the genes and proteins of organisms can be compared to determine their evolutionary relationship. The closer the genetic sequences of organisms are to each other, the more closely related they are and the more recently they diverged from a common ancestor.

Some genes change rapidly in evolution while others have changed extremely slowly. The rate of change in a gene over time is called the *molecular clock*. The rate of change in a gene's sequence is probably a function of the tolerance of the gene to changes without disrupting its function. Genes that change very slowly over extremely long periods of time probably do not tolerate change very well and play key roles in the life of cells and organisms. The large ribosomal RNA has changed slowly enough that it can be used to compare organisms all the way back to the divergence of eukaryotes, bacteria, and archaeobacteria. The enzymes of glycolysis play an essential role in energy production for all life and also evolve very slowly, allowing comparison of their gene sequences to illuminate evolutionary relationships over billions of years. Fossil genes are not available, but using computers to compare the gene sequences of many organisms allows researchers to determine how long ago organisms evolved from a common ancestor. More recently evolved genes and genes that evolve more rapidly can be used to compare more recent evolutionary events.

### SYSTEMATICS

Creating phylogenetic trees is part of systematics, the field of biology that investigates the diversity of life.

### WHEN THE APPENDIX MAKES ITS PRESENCE KNOWN

Although useless in present-day humans, the appendix can cause a lot of trouble if left unattended when it becomes inflamed. If it bursts, it can cause illness and even death.

## Vestigial Structures

*Vestigial structures* are structures that appear to be useless in the context of a particular modern-day organism's behavior and environment. It is apparent, however, that these structures used to have some function in an earlier stage of a particular organism's evolution. They serve as evidence of an organism's evolution over time, and can help scientists to trace its evolutionary path.

There are many examples of vestigial structures in humans, other animals, and plants. The appendix—small and useless in humans—assists digestion of cellulose in herbivores, indicating humanity's vegetarian ancestry; the tail—long and functional in most mammals—is reduced to a few useless bones (coccyx) at the base of the spine in humans. The splints on the legs of a horse are vestigial remains of the two side toes of *Hyracotherium*, the earliest known ancestor of the modern horse.

## MECHANISMS OF EVOLUTION

### The Population as the Basic Unit of Evolution

Evolution is the change in a population of a species over time. These changes are the result of changes in the gene pool of a population of organisms. Evolution does not happen in one individual but in a population of organisms. What is a population? A *population* is a group of individuals of a species that interbreed. A *species* is a group of individuals capable of interbreeding to produce offspring that can reproduce. In classical genetics, it is observed that the genotype of organisms produces their phenotype, the physical expression of inherited traits. A population of organisms includes individuals with a range of phenotypes and genotypes. It is possible, however, to describe a population not by their individual characteristics but by certain traits of the group as a whole, including the abundance of alleles within the whole population. The sum total of all alleles in a population is called the *gene pool*, and the frequency of a specific allele in the gene pool is called the *allele frequency*. Each individual receives its specific set of alleles from the gene pool, and not every individual receives the same alleles, leading to individual variation in genotypes and phenotypes.

How is the allele frequency calculated and used? For a diploid organism, the total number of alleles for a specific gene in a population is the number of individuals multiplied by 2. If there are 1,000 rabbits in a population and they are diploid, with two copies of every gene, then the gene pool of the rabbit population will include 2,000 alleles for each gene. If the genotype of every rabbit is known, then the allele frequency in the population can be determined by adding up how many copies of each allele are found in the population. If 100 rabbits are homozygous for an allele (both gene copies are the same) and another 200 rabbits in the population are heterozygous, then the allele frequency =  $[2(100) + 1(200)]/2,000$ . Allele frequency is the decimal fraction representing the relative presence of a particular allele for all members of a population. The letter  $p$  is used for the frequency of the dominant allele of a particular gene locus. The letter  $q$  represents the frequency of the recessive allele. For a given gene locus,  $p + q = 1$ . The total allele frequency for a gene must always equal 1.

Sexual reproduction constantly shuffles alleles around in a population, mixing and remixing them in new combinations through meiotic recombination, independent segregation of chromosomes during meiosis, and random matching of alleles from parents during mating and fertilization. All of these allow for mixing of alleles in a population to create variation in individual genotypes and phenotypes. Mutation in a population can create new alleles. Evolution is caused by changes in the gene pool of a population over time, as a result of changes that occur to individuals in the population caused by the alleles they carry.

## Hardy-Weinberg and Population Changes

The allele frequencies in the gene pool of a population determine how many individuals in a population get each allele and this in turn determines the phenotypes of individuals. If nothing changes the allele frequencies, then every generation will get the same alleles in the same proportions, and the population will not change over time. This idea is the foundation of population genetics and the central idea of *Hardy-Weinberg equilibrium* in population genetics.

According to the Hardy-Weinberg principle, allele frequencies in a population remain constant from generation to generation and a population is maintained in equilibrium as long as certain assumptions are met. If the assumptions are met, and the allele frequencies in the gene pool of a population are constant over time, the population does not change and evolution does not occur. If the assumptions are not true, then the allele frequencies of the population will change and the population will evolve.

The assumptions for Hardy-Weinberg equilibrium to be maintained are:

- Random mating (no isolation) must occur, so that no particular trait is favored. There can be no assortative mating (in other words, no organisms may select mating partners that resemble themselves).
- Immigration or emigration cannot take place.
- There must be no mutations.
- Large populations are required. As in all cases of probability, large samples are needed to provide an accurate approximation of the expected occurrence.
- Natural selection does not occur.

Under the above conditions, there is a free flow of alleles between members of the same species, while the total content of the gene pool is continually being shuffled. A constant gene pool is nevertheless maintained for the entire population. The constancy of the gene pool is always threatened by changes in the environment (which would favor certain genes), mutations, immigration and emigration (new genes introduced), or reproductive isolation (lack of random mating favors certain genes).

**Mathematical Demonstration of the Hardy-Weinberg Equilibrium Principle.** We can cross two individuals to demonstrate mathematically that the gene-pool frequencies remain constant generation after generation. Let us assume for the original gene pool

that the gene frequency of the dominant allele for tallness,  $p$ , is 0.80, and the gene frequency of the recessive allele,  $q$ , is 0.20. Thus  $p = 0.80$  and  $q = 0.20$ . The parents are crossed and their offspring frequencies are shown with a Punnett square.

The resulting gene frequency in the  $F_1$  generation is: 64 percent  $TT$ , 16 percent  $Tt$ , and 4 percent  $tt$ .

		Possible Sperm	
		0.80 $T$	0.20 $t$
Possible Eggs	0.80 $T$	0.64 $TT$ ( $p^2 = 0.64$ )	0.16 $Tt$ ( $pq = 0.16$ )
	0.20 $t$	0.16 $Tt$ ( $pq = 0.16$ )	0.04 $tt$ ( $q^2 = 0.04$ )

If there are no mutations, no migrations, and no decrease in population size, the frequencies of the above  $F_1$  generation are applicable in calculating the frequencies for the  $F_2$  generation. The alleles are reshuffled between individuals between generations but they are not lost.

**Some Mathematical Applications of the Hardy-Weinberg Principle.** In working out population genetics problems, we must recall that  $p$  = gene frequency of the dominant allele,  $q$  = frequency of the recessive allele, and  $p + q = 1$ , since frequencies of the dominant and recessive alleles total 100 percent. In a population that includes alleles  $p$  and  $q$ , the alleles will produce offspring with the following types and frequencies:

		Possible Sperm	
		$p$	$q$
Possible Eggs	$p$	$pp$	$pq$
	$q$	$pq$	$qq$

Thus, when we cross  $p + q$ , we obtain  $p^2 + 2pq + q^2 = 1$  (the "1" indicates that the total is 100 percent of the offspring). Note that the result is scientific confirmation of an obvious algebraic identity:  $(p + q)(p + q) = p^2 + 2pq + q^2$ . The official key for working out the following problem is as follows:

$p$  = frequency of dominant allele

$q$  = frequency of recessive allele

$p^2$  = frequency of homozygous dominant individuals

$2pq$  = frequency of heterozygous individuals

$q^2$  = frequency of homozygous recessive individuals

Another mathematical application of the Hardy-Weinberg principle is explained below:

**Problem:** In a certain population, the frequency of homozygous curly hair ( $CC$ ) is 64 percent. What percentage of the population has curly hair?

**TEST STRATEGY**

All you need to know to solve any Hardy-Weinberg problem is the value of  $p$  (or  $p^2$ ) and  $q$  (or  $q^2$ ). Once you know these, you can calculate everything else using the formulas  $p + q = 1$  and  $p^2 + 2pq + q^2 = 1$ .

**Solution:** According to the key,  $p$  represents the frequency of the dominant allele ( $C$ ), while  $q$  represents the frequency of the recessive allele ( $c$ ). We are told that the  $CC$  frequency is 64 percent. This means that  $p^2 = 0.64$  or  $p = 0.8$ . Since  $p + q = 1$ ,  $q = 1 - 0.8$ , or 0.2. An individual with curly hair may be either  $CC$  or  $Cc$ . The frequency of each genotype =  $p^2 + 2pq + q^2$ .

$$p^2 = 0.64 \text{ or } 64 \text{ percent homozygous curly hair}$$

$$2pq = 2(0.8)(0.2) \text{ or } 32 \text{ percent heterozygous curly hair}$$

$$q^2 = 0.04 \text{ or } 4 \text{ percent homozygous straight hair}$$

Therefore, the percentage of the population possessing curly hair is as follows:

$$p^2 + 2pq = 64 \text{ percent} + 32 \text{ percent} = 96 \text{ percent}$$

### Disruption of Hardy-Weinberg Equilibrium in Evolution

The Hardy-Weinberg principle describes the stability of the gene pool. However, no population stays in Hardy-Weinberg equilibrium for very long, because the stable, ideal conditions needed to maintain it do not exist. The assumptions required for equilibrium cannot be met in the real world.

As conditions change, the gene pool changes and the population changes. Changes in the gene pool caused by breaking the assumptions are the basis of evolution.

#### Mutation

If the gene pool is not going to change, and there is no immigration or emigration, then there can be no new alleles that appear in the population. Mutations may be infrequent in a population as a result of the great accuracy of DNA replication, but DNA replication is never perfect and some mutations will occur at least infrequently. Radiation from the environment and environmental mutagens also contribute a low but inescapable level of mutation in any population. The mutations will be a very small percentage of alleles in the gene pool of the population, but they do form an important component, acting as a source of variation for that population. Many mutations are harmful, but a small minority may confer a selective advantage in some way. Phenotypes are the material that evolution acts on in a population and mutations are the only source of truly new alleles that will result in truly new phenotypes.

#### Gene Flow

If two populations are separated from each other and do not interbreed, then the allele frequencies in their gene pools may be different from each other. If individuals move between the populations, however, carrying their alleles with them, this creates gene flow and will alter the frequency of alleles in both of the populations involved.

#### ADAPTING TO POISON

DDT is an insecticide used to kill insects. In the past, when it has been introduced into insect populations, a favorable environmental change has been created for DDT-resistant mutants and a fatal environmental change has been created for the rest of the insect population. Conditions have selected for the survival of DDT-resistant organisms.

### INSTABILITY IN THE GENE POOL

The following factors cause instability in the gene pool, resulting in the loss of Hardy-Weinberg equilibrium:

- Mutations
- Genetic drift
- Immigration/emigration
- Natural selection
- Nonrandom mating

### Population Size

One of the assumptions for the maintenance of Hardy-Weinberg equilibrium is that a population is large. Small populations are subject to random events that can statistically alter the gene pool. Changes in the gene pool caused by random events in a small population are called *genetic drift*. One example is a *population bottleneck*. If an event like a flood suddenly and dramatically reduces the size of a population, the allele frequencies of the survivors are not necessarily the same as the allele frequencies in the original population. When the population grows in size again, the allele frequencies in the new gene pool will represent the frequencies in the small bottleneck population, not the population before the reduction in size. A similar phenomenon called the *founder effect* occurs in the colonization of a new habitat. When a new island forms, it might be colonized by a very small number of individuals from another population. Since the new population is founded by a small number of individuals, it is statistically unlikely that the island population will represent the same allele frequencies as the population from which it was derived.

### Nonrandom Mating/"Sexual Selection"

If a population is going to maintain constant allele frequencies, then alleles must be matched randomly in each new generation. This requires that individuals mate with each other without any preference for specific traits or individuals. If the phenotype of individuals influences mating, this will change allele frequencies and disrupt Hardy-Weinberg equilibrium. Most species are quite discerning in mate selection, however, blocking maintenance of Hardy-Weinberg equilibrium.

### Natural Selection

Within a population of organisms, individuals are non-identical. Mutation is a source of new alleles, and sexual reproduction leads to constant shuffling of alleles in new genotypes. The variety of genotypes created in a population in this way creates a variety of phenotypes. If individuals have different phenotypes, then these individuals probably interact with their environment with differing degrees of success in escaping predators, finding food, avoiding disease, and reproducing. The differential survival and reproduction of individuals based on inherited traits is *natural selection*, as described first by Charles Darwin.

*Fitness* is a quantitative measure of the ability to contribute alleles and traits to offspring and future generations. The key to fitness is reproduction and survival of offspring. Traits that allow organisms to avoid predators, find food, resist disease, and improve survival are likely to improve fitness but only to the extent that they lead to more offspring and to more of the alleles being involved in the future gene pool. Finding a mate, mating, successful fertilization, and caring for offspring are factors that can improve fitness as well. There are different strategies for improving fitness. For example, some animals have lots of offspring but provide little parental care, while other animals have few offspring but provide lots of care for each of them.

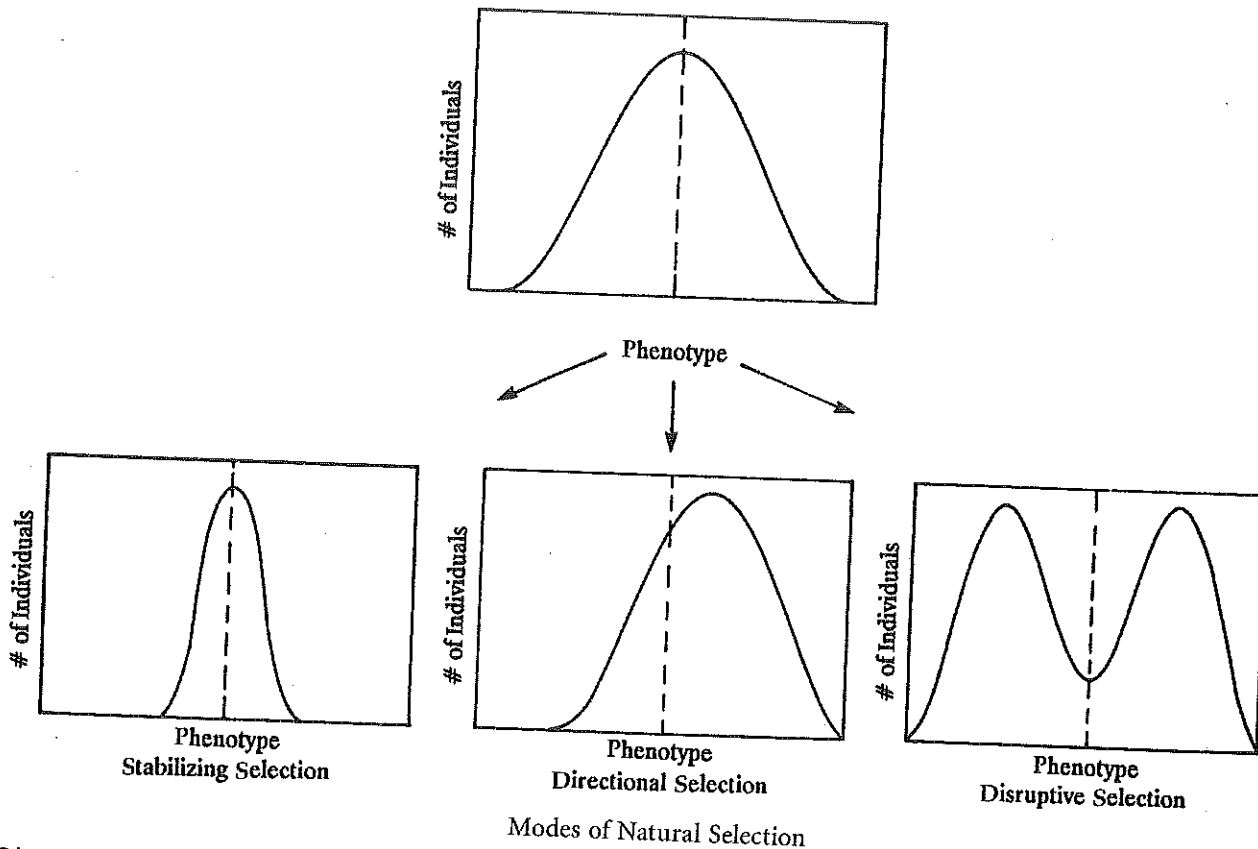
### FITNESS

*Fitness* is defined as the ability of an organism to contribute its alleles and therefore its phenotypic traits to future generations.

None of the other factors that alter Hardy-Weinberg equilibrium alter it in a directed fashion. Genetic drift, mutation, and migration are all random in their effects on the gene pool. Natural selection, however, increases the prevalence of alleles in a population that increase survival and reproduction. Alleles that increase fitness will over time increase in their allele frequency in the gene pool, and increase the abundance of the associated phenotype as well. This effect will change the population in a directed manner over many generations, creating a population that is better adapted to its environment.

Different types of natural selection can occur, including *stabilizing selection*, *disruptive selection*, and *directional selection*. Traits in a population such as the height of humans are often distributed according to a bell-shaped curve. The type of selection that occurs can affect the average value for the trait or it can alter the shape of the curve around the average.

*Stabilizing selection* does not change the average but makes the curve around the average sharper, so that values in the population lie closer to the average. For example, if both very small fish and very large fish tend to get eaten, then stabilizing selection may not alter the average fish size but is likely to cause future generations to be closer to average.



*Disruptive selection* is the opposite, in which the peak value is selected against, selecting for either extreme in a trait, so that a single peak for a trait in a population tends to be split into two peaks. *Directional selection* alters the average value for a trait, such as selecting for darker wings in a population of moths in an industrial area.



Natural selection acts on an individual and its direct descendants. In some cases natural selection can also act on closely related organisms that share many of the same alleles. This type of natural selection, called *kin selection*, occurs in organisms that display social behavior. The key to fitness is that an organism's alleles are contributed to the next generation. Contribution of alleles can happen by an individual or by close relatives like siblings, aunts, uncles, etc., who share many of the same alleles. The evolution of social organisms is the result of the increased fitness that social behavior provides. Cases of altruistic behavior in animals are probably the result of kin selection at work, in which an animal might sacrifice its own safety to allow relatives to survive, thereby increasing the fitness of itself and the whole social group with whom it shares alleles.

#### WHEN DIFFERENT SPECIES INTERBREED

Members of different species can interbreed under certain circumstances, but the offspring they produce is frequently infertile. Hence the mule, which is the offspring of two different species, the horse and the donkey, is sterile.

### Speciation

As mentioned previously, a species is a group of organisms that are able to interbreed and produce viable, fertile offspring. The key to defining a species is not external appearance. Within a species, there can be great phenotypic variation, as in the domestic dog. What defines a species is reproductive isolation, an inability to interbreed and create fertile offspring. Actual interbreeding is not necessary to make organisms the same species. Two groups of animals may live in different locations and never contact each other to interbreed, but if a researcher transports some of the animals and they create fertile offspring, they are part of the same species. The horse and the donkey can interbreed and create offspring, the mule. The mule, however, is sterile, meaning horses and donkeys are two different species.

*Speciation*, the creation of a new species, occurs when the gene pool for a group of organisms becomes reproductively isolated. At this point, evolution can act on a group that shares a gene pool separately from others. Two species can be derived from a single common ancestor species when two populations of a species are separated geographically or by a physical barrier, through a process known as *allopatric speciation*.

Separation of a widely distributed population by emerging geographic barriers causes each population to evolve specific adaptations for the environment in which it lives, in addition to the accumulation of neutral (random, nonadaptive) changes. These adaptations will remain unique to the population in which they evolve, provided that interbreeding is prevented by the barrier. In time, genetic differences will reach the point where interbreeding becomes impossible and reproductive isolation would be maintained if the barrier were removed. In this manner, geographic barriers promote evolution.

*Adaptive radiation* is the production of a number of different species from a single ancestral species. Radiation refers to a branching out; adaptive refers to the hereditary change that allows a species to be more successful in its environment or to be successful in a new environment. Whenever two or more closely related populations occupy the same niche, natural selection favors evolution of different living habits. This eventually results in the occupation of different niches by each population (this process is discussed in detail in the chapter on ecology). This divergent evolution through adaptive radiation has been an extremely frequent occurrence, as demonstrated by the famous example of Darwin's finches.

Speciation by populations that occupy the same region is called *sympatric speciation*. For organisms to become a new species through sympatric speciation they must be reproductively isolated while still living in the same region. The most common way for this to occur is through a sudden dramatic genetic change, and usually happens as a result of polyploidy of the genome. Polyploidy can occur within a species if an individual spontaneously (through an accident of meiosis) produces offspring with twice the normal chromosomal number. Polyploidy can also result when a cross between two related species produces a hybrid with the chromosomal complement of both parents. Since the chromosomes don't match, the hybrid cannot go through meiosis to reproduce sexually; but if it can reproduce asexually, then it can propagate.

## Lamarckian Evolution

Until it was supplanted by Darwin's ideas, French naturalist Jean-Baptiste Lamarck's theory was one of the more widely accepted explanations of the mechanisms of evolution. The cornerstone of Lamarck's hypothesis was the principle of use and disuse. He asserted that organisms developed new organs, or changed their existing ones, in order to meet their changing needs. The amount of change that occurred was thought to be based on how much or little the organ in question was actually used.

Unfortunately for Lamarck, this theory of use and disuse was based upon a fallacious understanding of genetics. Any useful characteristic acquired in one generation was thought to be transmitted to the next. An oft-cited example was that of early giraffes, who stretched their necks to reach for leaves on higher branches. The offspring were believed to inherit the valuable trait of longer necks as a result of their parents' excessive use of their necks. Modern genetics has disproved this concept of acquired characteristics.

It has now been established that changes in the DNA of sex cells are the only types of changes that can be inherited; because acquired changes are changes in the characteristics and organization of somatic cells, they cannot be inherited.

## CLASSIFICATION AND TAXONOMY

Evolution has created a great diversity of organisms on earth, but these organisms are related to each other through common ancestors they shared in the history of life. By examining organisms for common features and common ancestors, it should be possible to make sense of the diversity of life by grouping organisms into categories. The science of classifying living things and using a system of nomenclature to name them is called *taxonomy*. Carolus Linnaeus developed modern taxonomy in the 1700s, grouping organisms and naming them according to a hierarchical system.

A modern classification system seeks to group organisms on the basis of evolutionary relationships. The bat, whale, horse, and human are placed in the same class of animals

### NOT COMPLETELY WRONG

Although Lamarck's theory of evolution has been disproved, he did correctly observe that evolution should not be envisioned as a ladder, but instead as a pathway with many branches.

(mammals) because they are believed to have descended from a common ancestor. The taxonomist classifies all species known to have descended from the same common ancestor within the same taxonomic group.

Since much about early evolutionary history is not understood, there is some disagreement among biologists as to the best classification to employ, particularly with regard to groups of unicellular organisms. Taxonomic organization proceeds from the largest, broadest group to the smaller, more specific subgroups. The largest group, or kingdom, is broken down into smaller and smaller subdivisions. Each smaller group has more specific characteristics in common. Furthermore, each subgroup is distinguishable from the next. The naming system is subject to discussion and revision as research yields new insights over time into the relationship between organisms. Some classifications are clearer than others.

Viruses are obligate intracellular parasites that cannot conduct metabolic activities or replicate on their own. As such, they are not generally considered living, although they are certainly important to living systems. They are not classified within this taxonomy.

### Classification and Subdivisions

In the Linnean system, each *kingdom* has several major phyla. A *phylum*, or division, has several *classes*. Each class consists of many *orders*, and these orders are subdivided into *families*. Each family is made up of many *genera*. Finally, the *species* is the smallest subdivision.

Hence the order of classificatory divisions is as follows:

**KINGDOM → PHYLUM → [SUBPHYLUM] → CLASS →  
ORDER → FAMILY → GENUS → SPECIES**

The complete classification of humans is:

<i>Kingdom:</i>	Animalia
<i>Phylum:</i>	Chordata
<i>[Subphylum:]</i>	Vertebrata]
<i>Class:</i>	Mammalia
<i>Order:</i>	Primates
<i>Family:</i>	Hominidae
<i>Genus:</i>	<i>Homo</i>
<i>Species:</i>	<i>sapiens</i>

### Assignment of Scientific Names

All organisms are assigned a scientific name consisting of the genus and species names of that organism. Thus, humans are *Homo sapiens*, and the common housecat is *Felis domestica*.

#### MNEMONIC

The following sentence will help you remember the order of classificatory divisions:

**King Phillip Came Over For Good Sushi** (kingdom, phylum, class, order, family, genus, species).

One of the primary groupings of all living organisms separates *prokaryotes* from *eukaryotic* organisms. The *prokaryotes* include bacteria and another type of organism called archaeobacteria, or archae. Like the bacteria, archae have no organelles and have a simple circular DNA genome. Archae were relatively unknown until recently, and tend to inhabit harsh environments like hot springs that might resemble the early earth. Archae are distinct from bacteria in many ways such as the composition of their membrane lipids, and in some ways appear to be related more closely to eukaryotes than *prokaryotes*. For this reason, more recent classification schemes break all living things into three *domains*, groups at an even higher level than kingdom: bacteria, archaeobacteria, and eukaryotes.

## Bacteria

The ubiquitous bacteria are single-celled, lack true nuclei, lack a cytoskeleton, and contain double-stranded circular chromosomal DNA that is not enclosed by a nuclear membrane. These creatures nourish themselves heterotrophically—either saprophytically or parasitically—or autotrophically, depending upon the species. Bacteria are classified by their morphological appearance: cocci (round), bacilli (rods), and spirilla (spiral). Some forms are duplexes (diplococci), clusters (staphylococci), or chains (streptococci).

Bacteria have cell walls made of peptidoglycan, a specialized matrix of carbohydrates and peptides. A method of staining bacteria called Gram staining separates them into two groups according to the strength of the staining of their cell wall: gram-positive and gram-negative. Gram-positive cells have a thick peptidoglycan cell wall that stains strongly, while gram-negative cells have a thin cell wall and an outer membrane that stains poorly.

## The Protist Kingdom

The simplest eukaryotic organisms are the *protists*. Protists probably represent the first step in the evolution between *prokaryotes* and the rest of the eukaryotic kingdoms, including fungi, plants, and animals. Most, but not all, protists are unicellular eukaryotes. One way to define the protists is that this group includes organisms that are eukaryotes but are not plants, animals, or fungi. The protists include heterotrophs like *amoeba* and *paramecium*, photosynthetic autotrophs like *euglena* and *algae*, and fungilike organisms like *slime molds*. Some protists are mobile through the use of flagella, cilia, or amoeboid motion. Protists use sexual reproduction in some cases and asexual reproduction in others.

One of the best known types of protists are the *amoebas*. Amoebas are large, single-celled organisms that do not have a specific body shape. They move and change their shape through changes in their cytoskeleton and streaming of cytoplasm within the cell into extensions called pseudopods. Amoebas are heterotrophs that feed by engulfing a food source through phagocytosis, internalizing the food to digest it in vacuoles in the cytoplasm.

### STUDY TIP

Know the following broad categories of organisms by heart when the time comes to take your Biology E/M exam:

- Heterotrophic aerobes (amoebae, earthworms, humans)
- Heterotrophic anaerobes (yeast)
- Autotrophic aerobes (green plants)
- Autotrophic anaerobes (chemosynthetic bacteria)

*Ciliates* are another well-known group of protists and are complex single-celled organisms, including paramecium. The surface of ciliates is covered with cilia (short, hairlike processes) that beat in a coordinated fashion to move the cell through water. Ciliates have a defined shape and contractile vacuoles; under a microscope, these vacuoles can be seen beating to remove from the cell the excess water that enters the cell via osmosis. Food (yeast cells, in the case of paramecia) is internalized through an oral groove where it is digested in vacuoles. Paramecia reproduce mitotically, but also have a mechanism called conjugation for exchange of genetic material between cells.

*Slime molds* are an interesting group of organisms that in some cases are grouped with the fungi kingdom. They are heterotrophs, with some slime molds spending some of their time as independent cells but at other times gathering together to form multicellular forms that produce spores.

*Algae* are an important group of photosynthetic protists, mostly unicellular. Algae include diatoms, single-celled organisms with intricate silica shells; dinoflagellates, with flagella; and brown and red algae. Algae can reproduce sexually and with alternation of generations between diploid sporophytes and haploid gametophytes, as occurs in plants. The algae include large multicellular forms like giant kelp that might be grouped with the protists since they are an algae, but are also grouped with plants by others. It is likely that the plants evolved from one group of algae, the green algae.

### The Fungi Kingdom

*Fungi* are heterotrophs that absorb nutrients from the environment. Fungi are often saprophytic, feeding off of dead material as their nutrition source, and are important along with bacteria in the decay of material in ecosystems. Absorptive nutrition involves the secretion of enzymes that digest material in the extracellular environment, followed by absorption of the digested material back into the cell. One of the distinguishing features of fungi is their cell wall made of chitin, unlike the cellulose found in plants. Fungi often form long, slender filaments called hyphae. Mushrooms, molds, and yeasts are all examples of fungi.

#### WHAT CAME FIRST?

Heterotrophs were the first form of life to develop. As their need for nutrients surpassed the rate at which these nutrients were being spontaneously formed, autotrophs developed.

Most fungi reproduce both sexually and asexually. Asexual reproduction can occur through the production of haploid spores or through splitting of a piece of fungus that grows mitotically into a new organism. Sexual reproduction in fungi does not involve distinct male or female sexes, but multiple mating types that are not distinct in their morphology. Fungi spend most of their life cycle in a haploid form. Fertilization of haploid gametes forms a diploid zygote that usually quickly enters meiosis to produce haploid spores that can grow mitotically into mature haploid fungi.

### The Plant Kingdom

Plants are multicellular eukaryotes that produce energy through photosynthesis in chloroplasts, using the energy of the sun to drive the production of glucose. A cell wall of cellulose is a common feature of all plants, along with a life cycle featuring alternation of generations between haploid gametophytes and diploid sporophytes. The sporophyte

is a diploid form that makes haploid spores that grow into a complete haploid form, the gametophyte. The gametophyte in turn is a haploid form that produces haploid gametes that unite through fertilization during sexual reproduction. The resulting diploid zygote grows into the mature sporophyte once again.

Plants are distinct from animals in that plants are usually nonmotile while animals are heterotrophic and do move. Plant structure is adapted for maximum exposure to light, air, and soil by extensive branching; animals, on the other hand, are usually adapted for minimum surface exposure and maximum motility with compact structures. Animals have much more centralization in their physiology, while plants often exhibit delocalized control of processes and growth.

The evolution of plants has included the ongoing increase in the ability to conquer land through resistance to gravity and ability to tolerate drier conditions. The first plants probably evolved from green algae in or near shallow water. These first plants were nontracheophytes, without water transport systems (called vascular systems). Nontracheophytes include mosses. They also lack woody stems. Their lack of a vascular system restricts their size and generally restricts their range to very moist environments. In non-tracheophytes, the sporophyte is larger than the gametophyte.

The evolution of vascular systems was a major adaptation in plants. The first vascular plants, tracheophytes that did not produce seeds, included ferns and horsetails, plants with cells called tracheids that form tubes for the movement of fluid in the plant tissue called xylem. This vascular system also helps to provide rigid stems that plants need in order to live on land. These plants colonized land about 400 million years ago, making it possible for animals like arthropods to colonize land soon after. The non-seed tracheophytes dominated the land for 200 million years before plants with seeds appeared. Ferns form large sporophytes, which release haploid spores. These spores grow into gametophytes that produce haploid gametes. Fertilization in ferns requires sperm to swim through water, restricting most ferns to moist environments.

The evolution of the seed was the next major event in plant evolution, found first in the gymnosperms and later in the flowering plants, the angiosperms. The seed is a young sporophyte that becomes dormant early in development. The embryo is usually well-protected in the seed and able to survive unfavorable conditions by remaining dormant until conditions become more favorable again and the embryo begins to grow again, sprouting. In some cases seeds can remain viable for many years, waiting for the right conditions for the sporophyte to grow. This increases the ability of plants to deal with the variable conditions found on land. Seeds are produced as the result of fertilization of male and female gametes produced by male and female gametophytes. In the seed plants, the male and female gametophytes are small.

The conifers are the most abundant gymnosperms today. About 200 million years ago gymnosperms replaced seedless vascular plants, such as ferns, as the predominant plant forms on land. Pines and other conifers are large diploid sporophytes. Gymnosperms such as conifers produce male and female spores in separate cones. The male cones make the male gametophytes, which are pollen grains. In conifers, pollen grains are usually dispersed

by the wind to find female cones. Unlike ferns, conifers do not require male gametes to swim in water to find the female gametes to fertilize, an adaptation that allows these plants to live in drier environments. When the pollen grain finds the female cone, pollen tubes grow from the pollen grain to the female gametophyte that contains the eggs of the plant. Male gametes swim through the pollen tube to the eggs to fertilize them and create the diploid zygote that will grow into a seed and eventually another mature plant sporophyte.

Following the evolution of the seed, the next big innovation in plant evolution was the flower. The angiosperms represent the flowering plants and are today the predominant plant group in many ecosystems. Like the gymnosperms, angiosperms produce seeds. The seeds of gymnosperms are "naked," growing without a large amount of nutritional material or protective tissues. Angiosperms produce flowers as the sites for fertilization and produce seeds that are surrounded by nutritional tissues. Angiosperm development involves a double fertilization. One of the fertilizations involves the fertilization of an egg by one sperm that grows into the embryo. The other fertilization involves the fertilization of two female nuclei by one sperm to form a triploid tissue that grows to form the nutritive component of the seed, the endosperm. When the seed embryo germinates, it first gains nutrition from the endosperm.

### DON'T MIX THESE UP ON TEST DAY

Most people can tell the difference between plants and animals. But you will need to keep in mind some less obvious differences when taking your biology test:

*Animals* have a larval stage, are heterotrophic and mobile, are adapted for minimum surface exposure, and have no cell walls.

*Plants* have no larval stage, are photosynthetic and sessile, have extended branching, and have cell walls made of cellulose.

## The Animal Kingdom

Animals are fairly easy to recognize: animals are all multicellular heterotrophs. The evolution of animals has included the evolution of a variety of body plans to solve problems such as getting food, avoiding predators, and reproducing. Over time, animals have tended to become larger in size and more complex, with greater specialization of tissues. Another trend in the animal kingdom has been the evolution of increasingly complex nervous systems to enable complex behaviors in response to the environment.

Different groups of animals have evolved different body shapes, reflecting their different lifestyles. Animals with *radial symmetry* are organized with their body in a circular shape radiating outward. The echinoderms (like sea stars) and the cnidarians (like jellies) are examples of animals with radial symmetry. Another common body plan is *bilateral symmetry*, in which the body has a left side and a right side that are mirror images of each other. Humans are a good example of bilateral symmetry, in which a plane drawn vertically through the body splits the body into left and right sides that look the same.

In animals, the front of the body, where the head is located, is the *anterior*, and the rear of the animal is the *posterior*. The back of the animal, where the backbone is located in vertebrates, is the *dorsal* side (like the dorsal fin) and the opposite side of the animal (which, depending on the posture of the animal, could be the underside, belly, or front) is the *ventral* side.

The method used to capture food is intimately related to the animal's body shape. Some animals that do not move are called *sessile*. Most of these animals gather food by filtering it from the environment. Examples of sessile filter feeders include sponges and cnidarians. This is a highly successful lifestyle that requires little energy to gather food, waiting instead for the food to come to you, but animals with this lifestyle are

t the mercy of their environment and must compete for space and resources. Animals with more active lifestyles have evolved increasingly complex nervous systems and motor systems to enable them to navigate their environment.

During the early stages of animal development, immediately after fertilization, the embryo enters into several rapid cycles of cell division that split the zygote into increasingly smaller cells. In some animals called *protostomes* the cells in the early embryo divide in a spiral pattern, while in *deuterostome* animals, the cells are cleaved in a radial pattern. The protostomes include annelids, arthropods, mollusks, and roundworms, while the deuterostomes include the echinoderms and chordates. These divisions reflect one of the major evolutionary divisions in the animal kingdom.

The body cavity in animals has evolved over the history of animals. The body cavity is the area between the gut and the exterior of the animal. Early animals like flatworms have only solid tissue between the gut wall and the exterior surface. Other animals like annelids and chordates have evolved a cavity called the *coelom* between the gut and the exterior wall. The coelom is lined with muscle both around the gut and the interior wall. The organs of the chest and abdomen in chordates reside within the coelom. The coelom in annelids makes coordinated motion with a hydrostatic skeleton possible.

The phyla that are described here do not include all of the animal phyla, but most of the more abundant and important phyla.

**Phylum Porifera (sponges).** Animals probably evolved from simple colonial heterotrophic protists with groups of cells living together and starting to specialize for different functions. These simple animals, probably representing the first evolutionary step between protists and animals, are the *sponges*, *phylum Porifera*. Sponges resemble a colonial organism, with only a small amount of specialization of cells within the animal, no organs, and distributed function. Sponges usually have only a few different types of cells and no nervous system, and if broken apart can reassemble into new sponges. With a saclike structure, sponges use flagellated cells that move water into the animal through pores into a central cavity and back out again. Cells lining the cavity capture food from water as it moves past.



Sponge

**THE SESSILE SPONGE**

Unlike most animals, sponges are sessile—they do not move. Like many sessile organisms, sponges gather food by filtering water through their bodies.

**Phylum Cnidaria (hydra, sea anemone, jellies).** *Cnidarians*, also called coelenterates, have radial symmetry, with tentacles arranged around a simple gut opening. Their gut has only one opening to the environment, through which food passes in and wastes pass out. They are aquatic animals and represent one of the earliest phyla of animals. With only two cell layers, the endoderm and ectoderm. These animals have a decentralized system for simple response to the environment, a nerve net to respond to the environment, a decentralized system for simple

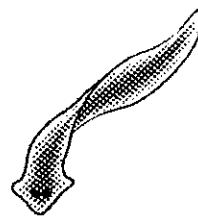


responses to the environment such as retraction of tentacles or swimming the body. The tentacles contain one of the trademarks of cnidarians, stinging *nematocysts* that have a harpoon-like structure with toxins to capture prey. Life cycles of cnidarians can include a polyp stage and a medusa stage. The polyp is a sessile form, with the mouth opening pointed upward, while the medusa is a swimming form, with the mouth opening pointed downward. Polyps are asexual. Sea anemones in a cluster on a rock are often clones of each other that have reproduced by budding, competing for space with other clones. Sexual reproduction occurs in the medusa, where sperm and eggs are produced and released into the environment for fertilization.



Cnidarian

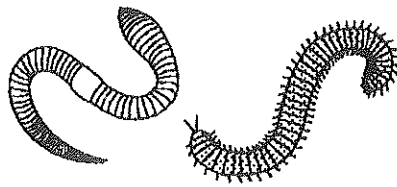
**Phylum Platyhelminthes (flatworms).** Flatworms are ribbonlike with bilateral symmetry. They possess three layers of cells, including a solid mesoderm, but lack a circulatory system. Their nervous system consists of simple light detection organs, an anterior brain ganglion, and a pair of longitudinal nerve cords. Their digestive system is a cavity with a single opening, and they lack a coelom. These animals are not swift-moving, using cilia to move over surfaces. A common flatworm is the planaria, famous for its regeneration. The shape of the worm, elongated and without appendages, has evolved in many phyla as a compact structure that is well designed for movement. Planaria are free-living but many flatworms are internal parasites, including flukes and tapeworms, deriving their nutrition by direct absorption into their cells from the host.



Flatworm

**Phylum Nematoda (roundworms).** Nematodes are roundworms, with three layers, including mesoderm, a complete digestive tract with two openings, a mouth and an anus, and a body cavity called a *pseudocoelom* around the gut. The pseudocoelom has muscle lining the interior body wall but not around the gut. This allows for active movement, but it is more of a wiggling motion than movement in a specific direction. Nematodes do not have respiratory or circulatory systems, exchanging gases directly with the environment. Roundworms are one of the most abundant animal groups, including huge numbers of free-living, scavenging species as well as parasitic species. The species *Caenorhabditis elegans* is a simple organism with only 950 cells, which has made it popular in modern biology for studies of cell differentiation and genetics.

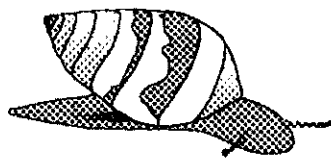
**Phylum Annelida (segmented worms).** The earthworm and leeches are examples of annelid worms, commonly called *segmented worms*. The annelids are worms with segmented bodies and a coelom body cavity. The division of the body into segments and the presence of the coelom body cavity filled with water creates a hydrostatic skeleton that allows annelids complex, sophisticated movement, which is coordinated by a nervous system. Each segment has local control by a ganglion of nerves but these nerves are coordinated by a ventral nerve cord and a larger nerve collection that might be called a brain in the front of the worm. Annelids exchange gases directly with their environment through their skin, an important reason why they have moist skin and live in moist environments. Each segment has a twin set of excretory organs called *nephridia*. Annelids have a complete digestive tract with some specialization into organs along the tract and they also have a closed circulatory system with five pairs of hearts.



Annelids

**Phylum Arthropoda.** Arthropods have jointed appendages, exoskeletons of chitin, and open circulatory systems. With an exoskeleton, the coelom of arthropods is reduced and less important in movement. The three most important classes of arthropods are insects, arachnids, and crustaceans. The exoskeleton of arthropods has muscles attached to its interior for movement. The exoskeleton provides protection and has a variety of specialized appendages. The exoskeleton prevents gas exchange between the skin and exterior, however, as occurs in annelids, requiring the evolution of a respiratory system. Insects possess three pairs of legs, spiracles, and tracheal tubes designed for breathing outside of an aquatic environment. Arachnids have four pairs of legs and "book lungs" (layers of tissue, arranged like the pages in a book, over which air circulates for respiration); spiders and scorpions are types of arachnids. Most arthropods have complex sensory organs, including compound eyes, that provide information about the environment to their increasingly complex nervous systems. Crustaceans have segmented bodies with a variable number of appendages. Crustaceans such as the lobster, crayfish, and shrimp possess gills for gas exchange. The exoskeleton of arthropods allowed them to colonize land and become the first winged organisms as well. Arthropods, particularly insects, remain one of the most abundant and varied groups of organisms on earth.

**Phylum Mollusca.** The *mollusks* include animals like clams, squid, and snails. In body shape these animals do not resemble each other very much, but they do share a few basic traits that lead biologists to classify them together as mollusks. These shared mollusk traits include a muscular foot, a mantle that secretes a shell, and a rasping tongue called the *radula*. Most mollusks are covered by a hard protective shell secreted by the mantle. Some mollusks such as squid and octopi have a reduced, internal shell. Mollusks are mostly aquatic and for respiration use gills that are enclosed in a space created by the mantle, the mantle cavity. The gills are also involved in feeding and move water over their surface with the beating of cilia.



Mollusk

**Phylum Echinodermata.** The *echinoderms*, which include sea stars and sea urchins, are spiny, have radial symmetry, contain a water-vascular system, and possess the capacity for regeneration. The echinoderms may not resemble the vertebrates but they share with chordates that they are deuterostomes. The water-vascular system is a unique adaptation of the echinoderms, with a network of vessels that carries water to extensions called tube feet. The tube feet are the small suckerlike extensions in sea stars, sea urchins, and sand dollars that allow the animals to adhere to surfaces and to move. Echinoderms also have a hard internal skeleton, formed from calcium deposits, that assists in protection and locomotion.

**Phylum Chordata.** The chordates have a stiff, solid dorsal rod called the *notochord* at some stage of their embryologic development, as well as paired gill slits. Chordata have dorsal hollow nerve cords, tails extending beyond the anus at some point in their development, and a ventral heart. These adaptations may not sound impressive but they paved the way for the evolution of the vertebrates, a major subphylum of chordates.

The chordates probably originated from animals like tunicates, commonly called sea squirts. Adult tunicates are sessile filter feeders that do not resemble vertebrates at all. Tunicate larvae, however, are free-swimming, with a notochord and a dorsal nerve cord, and resemble tadpoles.

#### EVOLUTIONARY PATHWAY

The evolutionary pathway to humans is as follows:

Porifera  
 Radiata  
 Acoelomates  
 Pseudocoelomates  
 Protostomes  
 Annelida  
 Arthropoda  
 Deuterostomes  
 Chordata  
 Vertebrata  
 Mammalia  
*Homo sapiens*

The *vertebrates* are a subphylum (smaller than a phylum, larger than a class) of the chordates that includes fish, amphibians, reptiles, birds, and mammals. In vertebrates the notochord is present during embryogenesis but is replaced during development by a bony, segmented vertebral column that protects the dorsal spinal cord and provides anchorage for muscles. Vertebrates have bony or cartilaginous endoskeletons, chambered hearts for circulation, and increasingly complex nervous systems. The vertebrate internal organs are contained in a coelom body cavity.

The first vertebrates were probably filter-feeding organisms that evolved into swimming jawless fishes that were still filter feeders. Jawless fish such as lampreys and hagfish still exist today. The evolution of fish with jaws led to the development of the cartilaginous and bony fishes that are dominant today. These fish use gills for respiration, and move water over the gills through paired gill slits. The jaw allows fish to adopt new lifestyles other than filter feeding, grabbing food with their jaws. Cartilaginous fish (class *Chondrichthyes*) like sharks and rays have an endoskeleton that is made entirely of cartilage rather than hard, calcified bone. They have large, oil-producing livers for buoyancy regulation in water. Bony fishes (class *Osteichthyes*) have swim bladders for the regulation of buoyancy.

Two adaptations were important to set the stage for vertebrates to colonize the land. One was the presence of air sacs that allowed some fish in shallow water to absorb oxygen from air for brief periods. The other adaptation was a change in the structure of fins to have lobes that allowed some degree of movement on land. Fish with these features evolved into the *amphibians* about 350 million years ago. Most amphibians such as frogs and salamanders still live in close association with water and have only simple lungs or gills supplemented by oxygen absorbed through the skin. Another reason that amphibians are mostly associated with water is that amphibian eggs lack hard shells and will dry out on land. Amphibian larvae often live in water and then metamorphose into the adult form.

*Reptiles* became independent of water for reproduction through the evolution of hard-shelled eggs that do not dry out on land. The eggshell protects the developing embryo but still allows gas exchange with the environment. Reptiles also evolved more effective lungs and hearts and thicker dry skins to allow them a greater metabolic activity than amphibians and the ability to survive on land.

*Birds* evolved from reptilian dinosaurs with the development of wings, feathers, and light bones for flight. Birds also have four-chambered hearts and uniquely adapted lungs to supply the intense metabolic needs of flight. Birds have hard-shelled eggs and usually provide a great deal of parental care during embryonic development and maturation after hatching. A famous evolutionary intermediate from the fossil record is *Archaeopteryx*, which is dinosaurlike in some respects, but had feathers and wings.

*Mammals* are the remaining major class of vertebrates. Mammals have hair, sweat glands, mammary glands, and four-chambered hearts. The fossil record indicates that mammals evolved 200 million years ago and coexisted with the dinosaurs up until the major extinction of the dinosaurs 65 million years ago. At that time, mammals diversified to occupy many environmental niches and become the dominant terrestrial vertebrates in many ecosystems. Mammals are highly effective in regulating body temperature, and most mammals provide extensive care for their young. One small group of mammals, the *Monotremes* (for example, the duck-billed platypus), lay eggs. Other mammals gestate their embryos internally and give birth to live young. *Marsupial* mammals give birth after a short time and complete the development of their young in an external pouch. *Placental* mammals gestate their young to a more mature state, providing nutrition to the embryo with the exchange of material in the placenta. Marsupial mammals were once widespread across the globe but were replaced in most cases by placental mammals. Australia, being isolated, was and is a haven for marsupial mammals to the present day.

Among the mammals, the primates have opposable thumbs and stereoscopic vision for depth perception, adaptations for life in the trees, and additional traits that have been important factors in the evolution of humans. Many primates have complex social structures. The ancestors of humans included *australopithecines*. Fossils indicate these ancestors were able to walk upright on two legs on the ground. Fossil remains of hominids such as *Homo habilis* from 2 to 3 million years ago display an increasing size of the brain. *Homo habilis* probably used tools, setting the stage for modern humans, *Homo sapiens*.

#### THE POUCH HOLDS ITS GROUND

Pouched animals are primitive, and have generally died out in areas where they were put in direct competition with more evolutionarily advanced placental animals. Opossums, however, did manage to cross the Isthmus of Panama land bridge and compete successfully with placental mammals.

**THINGS TO REMEMBER**

- Evolution and natural selection
- Homologous, analogous, and vestigial anatomic structures
- The Hardy-Weinberg principle and equation
- Gene flow
- The order of Linnean classification (kingdom, phylum, [subphylum], class, order, family, **genus**, species)

**EVOLUTION AND DIVERSITY QUIZ**

1. Nematocysts are characteristic of
  - (A) Porifera.
  - (B) Protozoa.
  - (C) Cnidaria.
  - (D) Annelida.
  - (E) Echinodermata.
2. According to the modern theory of evolution, which of the following evolved first?
  - (A) The Krebs cycle
  - (B) Anaerobic respiration
  - (C) Autotrophic nutrition
  - (D) Photosynthesis
  - (E) Chemosynthesis
3. Which of the following is an INCORRECT association?
  - (A) Porifera: sessile
  - (B) Echinodermata: radial symmetry
  - (C) Annelida: coelom
  - (D) Platyhelminthes: anus
  - (E) insects: tracheal tubes
4. Intestinal nematodes evolved from a free-living to a parasitic form through developing
  - (A) special reproductive segments.
  - (B) a symbiotic relationship with intestinal bacteria.
  - (C) an external cuticle resistant to digestive enzymes.
  - (D) a long digestive tube.
  - (E) an intricate nervous system.
5. Which statement about the phylum Echinodermata is FALSE?
  - (A) The phylum includes sea stars and sea urchins.
  - (B) Echinoderms reproduce sexually.
  - (C) The phylum includes crayfish.
  - (D) Echinoderms are heterotrophs.
  - (E) Echinoderms are invertebrates.
6. Echinoderms are regarded as being closely related to chordates due to their
  - (A) bilateral symmetry.
  - (B) form of circulatory system.
  - (C) endoskeleton.
  - (D) early embryonic development.
  - (E) possession of a notochord.
7. Which of the following is NOT a distinction between plants and animals?
  - (A) Plants have cellulose cell walls.
  - (B) Plants have intermediate larval stages.
  - (C) Plants are extensively branched.
  - (D) Animals are heterotrophic.
  - (E) All of the above.
8. Which of the following organisms is a chordate but NOT a vertebrate?
  - (A) Shark
  - (B) Lamprey eel
  - (C) Turtle
  - (D) Tunicate
  - (E) None of the above

9. The notochord is
- (A) present in all adult chordates.
  - (B) present in all echinoderms.
  - (C) present in chordates during embryological development.
  - (D) always a vestigial organ in chordates.
  - (E) part of the nervous system of all vertebrates.
10. Which order of classificatory divisions is correct?
- (A) Kingdom, class, genus, family
  - (B) Phylum, class, order, genus
  - (C) Species, order, family, phylum
  - (D) Class, kingdom, family, order
  - (E) Genus, order, species, phylum
11. The hypothesis that chloroplasts and mitochondria were originally prokaryotic organisms living within eukaryotic hosts is supported by the fact that mitochondria and chloroplasts
- (A) possess protein synthetic capability.
  - (B) possess genetic material.
  - (C) possess a lipid bilayer membrane.
  - (D) possess characteristic ribosomes.
  - (E) All of the above
12. Which of the following is a member of the protist kingdom?
- (A) Archaeobacteria
  - (B) Unicellular green algae
  - (C) Anaerobic bacteria
  - (D) Fungi
  - (E) Mosses
13. Which of the following has a chitinous exoskeleton?
- (A) Oriole
  - (B) Sea stars
  - (C) Clam
  - (D) Honeybee
  - (E) Earthworm
14. Compound eyes are found in
- (A) Porifera.
  - (B) Coelenterata.
  - (C) Mollusca.
  - (D) Arthropoda.
  - (E) Annelida.
15. Reptiles
- (A) are homeothermic.
  - (B) respire with gills.
  - (C) must live in water at some stage of their life cycle.
  - (D) possess notochords as adults.
  - (E) lay leathery eggs.
16. Which of the following men might have explained the auk's loss of the ability to fly with the following hypothesis?
- "Since the auk stopped using its wings, the wings became smaller, and this acquired trait was passed on to the offspring."*
- (A) Darwin
  - (B) Mendel
  - (C) De Vries
  - (D) Lamarck
  - (E) Morgan

17. All arthropods
- (A) have a body consisting of three parts (the head, the thorax, and the abdomen).
  - (B) breathe through spiracles that lead to the tracheal tubes.
  - (C) have a hard, calcium-rich shell with a soft mantle.
  - (D) have radial symmetry.
  - (E) have jointed appendages and exoskeletons.
18. Which of the following will NOT affect the frequency of a gene in an ideal population?
- (A) Environmental selective pressure
  - (B) Mutation
  - (C) Random breeding
  - (D) Nonrandom matings
  - (E) Selective emigration



# Review Chapter 3: DNA, RNA, and Proteins

**T**wo of the most important types of molecules in the cell are *proteins* and *nucleic acids*. Proteins are large, diverse molecules with many roles in cell structure and function. Proteins allow molecules to cross the cell membrane, make up the cytoskeleton, associate with and organize DNA, and compose substances surrounding cells and tissues, such as collagen. Protein enzymes help carry out biochemical reactions. Proteins depend on nucleic acids, DNA and RNA. DNA makes up the chromosomes and contains the cell's genetic information—the instructions for making proteins. RNA plays other crucial roles in protein synthesis.

## Proteins and Amino Acids

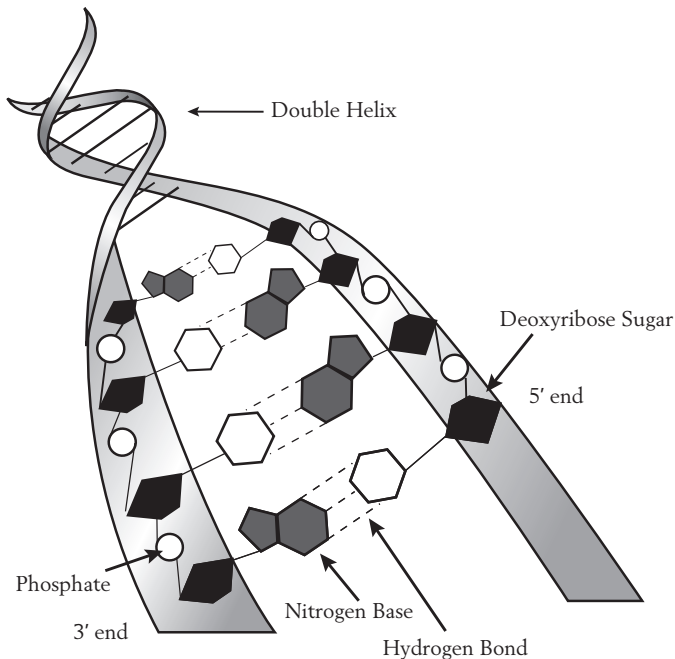
Proteins are polymers or macromolecules (large molecules) made up of repeating monomers (subunits) called *amino acids*. Just 20 different amino acids make up all the proteins of nearly every living thing. Each amino acid shares the same basic molecular structure. However, they differ in the side group, the one or more atoms attached to the central carbon atom that confer different chemical properties (for example, hydrophilic vs. hydrophobic) to each type of amino acid.

Amino acids are joined end to end in linear chains. Once synthesized, these chains twist and fold to take on their functional, three-dimensional

shapes. Four levels of protein structure are considered. The primary structure of a protein refers to the sequence of amino acids making up the chain. The secondary structure refers to shapes or motifs (such as an alpha helix) that may occur in parts of the chain. The tertiary structure refers to the three-dimensional shape of the folded polypeptide chain. Finally, the quaternary structure describes how the polypeptide associates with other polypeptides (for example, if it forms a protein with multiple parts).

## Nucleic Acids and Nucleotides

Nucleic acids, DNA and RNA, are polymers made up of nucleotide monomers. The molecular structure of DNA is shown in the figure below. DNA stands for *deoxyribonucleic acid*; each monomer consists of (1) a five-carbon sugar, deoxyribose; (2) a phosphate group, and (3) a nitrogen base (A, C, G, or T). If you consider a double DNA strand as a ladder, the sugar and phosphate groups make up the “sides,” while the paired nitrogenous bases make up the “rungs.”



The two strands are held together by hydrogen bonding interactions between complementary bases: adenine bonds with thymine, and guanine bonds with cytosine. The double-stranded DNA twists to form a double helix.

Each DNA strand in a double strand has a direction. One strand is oriented in the 3'-to-5' direction; the other strand is oriented in a 5'-to-3' direction. This means that the strands are antiparallel. (NOTE: The numbers 3 and 5 refer to carbons in the sugar.) Eukaryotic chromosomes are made up of extremely long double strands of DNA wound around proteins called *histones*.

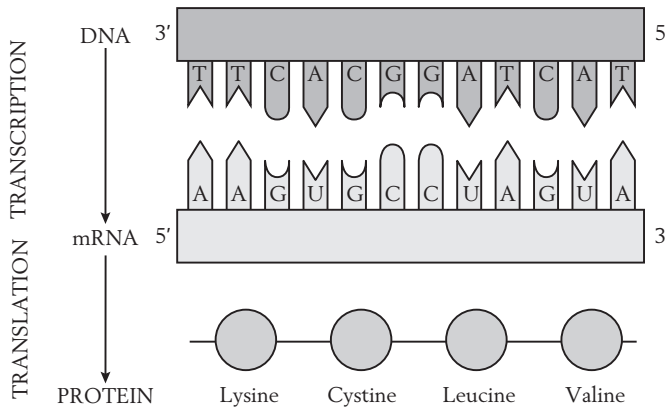
## Replication of DNA

When a chromosome replicates its DNA, the DNA double strands “unzip” as the hydrogen bonds between them are broken. Then, free nucleotides pair with the nucleotides of the separated strands, which act as templates. The nucleotides are joined by the enzyme DNA polymerase, creating a new, complementary strand for each original DNA strand. Each new DNA double strand consists of one old (template) strand and one newly formed strand. For this reason, DNA replication is called *semiconservative*.

## RNA

RNA (for *ribonucleic acid*), like, DNA, is made up of nucleotide monomers linked by their sugar and phosphate subunits. Unlike DNA, the five-carbon sugar in RNA is ribose. In addition, the nucleotide uracil replaces thymine, and RNA does not form a double-stranded helix.

Three important forms of RNA are mRNA, tRNA, and rRNA. Ribosomal RNA (rRNA), along with protein, makes up the ribosome. The others (mRNA and tRNA) are also involved in protein synthesis, as described in the following sections. The relationship between the information in DNA and mRNA, and between mRNA and polypeptide sequence, is known as the central dogma of molecular genetics.



## Transcription of mRNA

Genetic information specifies the proteins that a cell may produce. The sequence of nucleotides making up a gene corresponds to the sequence of amino acids in the polypeptide chain that will eventually be assembled. This information is contained in the chromosomes, which reside in the nucleus. Protein assembly, however, takes place in the ribosomes of the ER and cytoplasm. Therefore, the information in DNA must be copied and transported out of the nucleus; this is the function of messenger RNA, or mRNA.

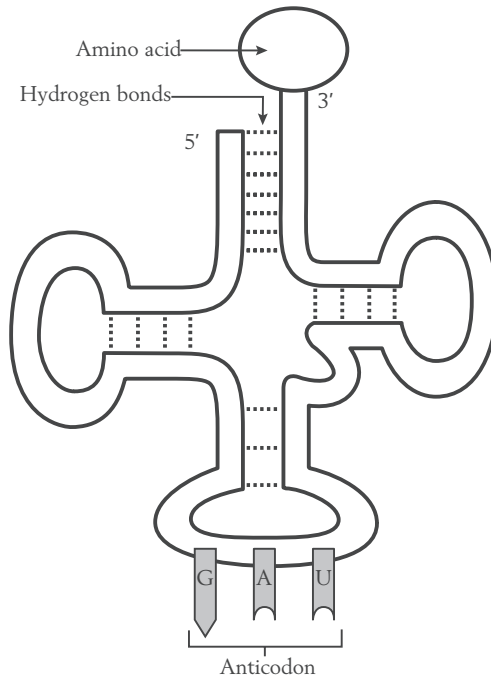
In the location of the gene, the DNA uncoils, and its two strands “unzip,” exposing the nitrogen bases. As RNA nucleotides base-pair with the exposed DNA nucleotides, a complementary, antiparallel RNA chain forms. The free nucleotides are joined together by the RNA polymerase enzyme. Once the gene has been transcribed (or copied to RNA), the newly formed mRNA is released and processed in the nucleus. It then exits through a nuclear pore and travels to a free or ER-bound ribosome in the cytoplasm.

## Translation of mRNA to Protein

Translation of the genetic information in mRNA to a polypeptide strand is carried out by the ribosome. A nucleotide sequence is read in triplets called *codons*. Each codon corresponds to an amino acid; this is the

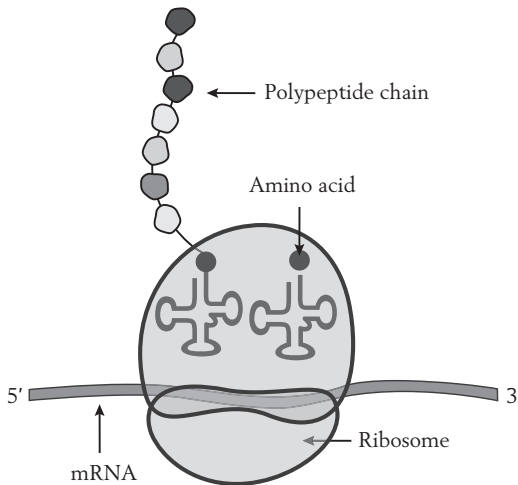
nearly universal genetic code of life. Because there are four nucleotides making up the codons, there are 64 ( $4 \times 4 \times 4$ ) codons. However, there are only 20 amino acids. Most amino acids are specified by more than one codon. There is one codon, a stop codon, that does not specify any amino acid but acts as a signal for translation to stop. The codon signal for translation to start codes for the amino acid methionine. The mRNA strand attaches to a ribosome, and translation initiates at the start codon.

Transfer RNA (tRNA) interprets the genetic sequence into an amino acid sequence. The cell contains different forms of tRNA, each of which has a specific anticodon and carries a specific amino acid. A tRNA molecule is a looped structure with an anticodon at one end and a site that attaches to an amino acid at the opposite end.



As the mRNA binds to the ribosome, one codon is exposed at a time. The tRNA with the complementary anticodon binds to the mRNA codon in the "hot seat." Once bound, the amino acid it carries is transferred to the ribosome, and the tRNA molecule is released. Then, the mRNA transcript

shifts along the ribosome so that the next codon is in the “hot seat.” A complementary tRNA binds, its amino acid is joined to the previous amino acid, the tRNA is released, and the mRNA shifts again. As this process is repeated, the polypeptide chain elongates. Eventually, the stop codon is reached and translation is completed. The polypeptide chain is released and allowed to take on its three-dimensional protein conformation.



## Gene Expression

The transcription of a gene, or *gene expression*, is regulated by DNA sequences (called *promoters* and *enhancers*) located upstream and downstream of the gene. Proteins called *transcription factors* bind to these DNA sequences and either activate or repress the activity of RNA polymerase. Different sets of transcription factors are present in different cells of multicellular organisms, driving the differences in gene expression among the millions of genetically identical cells that make up a plant or animal body. Gene expression differences lead to the differentiation and specialization of cells in multicellular organisms.

## Review Questions

1. The genetic code table shows the amino acids that correspond to mRNA codons. Each codon is read from 3' (first nucleotide) to 5' (third nucleotide). A DNA template strand is shown. Which amino acid sequence will be assembled from the mRNA associated with this strand?

		Second mRNA Nucleotide					
		U	C	A	G		
First mRNA Nucleotide (5' end)	U	UUU (Phe) UUC UUA (Leu) UUG	UCU UCC (Ser) UCA UCG	UAU (Tyr) UAC UAA STOP UAG STOP	UGU (Cys) UUC UGA STOP UGG (Trp)	U C A G	Last mRNA Nucleotide (3' end)
	C	CUU CUC (Leu) CUA CUG	CCU CCC (Pro) CCA CCG	CAU (His) CAC CAA (Gln) CAG	CGU CGC (Arg) CGA CGG	U C A G	
	A	AUU (Ile) AUC AUA (Met) AUG	ACU ACC (Thr) ACA ACG	AAU (Asn) AAC AAA (Lys) AAG	AGU (Ser) AGC AGA (Arg) AGG	U C A G	
	G	GUU GUC (Val) GUA GUG	GCU GCC (Ala) GCA GCG	GAU (Asp) GAC GAA (Glu) GAG	GGU GGC (Gly) GGA GGG	U C A G	

DNA template strand: 5'-GCG ACA TAC ACT-3'

- A. Ala—Cys—Met
- B. Arg—Cys—Met
- C. Ala—Thr—Tyr—Thr
- D. Ala—Thr—Tyr—Trp
- E. Arg—Thr—Tyr—Thr

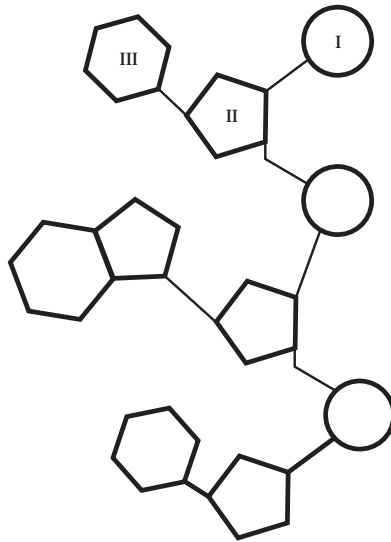
2. Which answer choice matches the functions listed below to the correct RNA types?
- I. Interprets a codon as an amino acid
  - II. Binds to a gene transcript
  - III. Contains information for assembling a protein
- A. I. mRNA; II. rRNA; III. tRNA
  - B. I. mRNA; II. tRNA; III. rRNA
  - C. I. rRNA; II. mRNA; III. tRNA
  - D. I. tRNA; II. mRNA; III. rRNA
  - E. I. tRNA; II. rRNA; III. mRNA
3. An original cell replicates its DNA. The cell undergoes mitosis and cell division, and one of the daughter cells replicates its DNA. Which of these BEST describes the DNA of the second-generation daughter cells?
- A. One of the cells will contain DNA consisting of one strand from the original cell.
  - B. One of the cells will contain DNA consisting of two strands from the original cell.
  - C. Both of the cells will contain DNA consisting of one strand from the original cell.
  - D. Both of the cells will contain DNA consisting of two strands from the original cell.
  - E. Neither of the cells will contain DNA consisting of strands from the original cell.



4. The partial sequence of a single DNA strand is shown. What will be the sequence of the complementary strand produced during DNA replication?

3'-ATGCTGAACT-5'

- A. 5'-ATGCTGAACT-3'  
 B. 5'-CGTAGTCCAG-3'  
 C. 5'-GACCTGATGC-3'  
 D. 5'-GCATCAGGTC-3'  
 E. 5'-TACGACTTGA-3'
5. Which of these labels matches the structure of the partial RNA strand shown?



- A. I. phosphate; II. ribose; III. nitrogen base  
 B. I. phosphate; II. nitrogen base; III. ribose  
 C. I. nitrogen base; II. phosphate; III. ribose  
 D. I. ribose; II. nitrogen base; III. phosphate  
 E. I. ribose; II. phosphate; III. nitrogen base

## Answer Explanations

1. **B.** The DNA template is used to produce a complementary RNA strand with the sequence 3'-CGC UGU AUG UGA-5'. The first three codons specify the amino acids alanine, cystine, and methionine, respectively. The last codon is a stop codon, which tells the ribosome to end translation.
2. **E.** Transfer RNA (tRNA) molecules carry amino acids and bind to specific mRNA codons. Ribosomal RNA (rRNA) makes up the ribosome and binds to the mRNA transcript. Messenger RNA (mRNA) carries genetic information from the nucleus to the cytoplasm.
3. **A.** DNA replication is semiconservative. During the first round of replication, each original single strand pairs with a new complementary strand. The first-generation daughter cells each have DNA consisting of one original and one new strand. During replication, the single original strand will pair with a new strand. The other double strand will consist of nonoriginal DNA.
4. **E.** The complementary base-pairing rule for DNA is that adenine (A) pairs with thymine (T), and cytosine (C) pairs with guanine (G). DNA strands run antiparallel, so the complementary strand will be in the 5'-to-3' direction. The complementary sequence is, therefore, 5'-TACGACTTGA-3'.
5. **A.** The five-carbon ribose sugar (II) and the phosphate group (I) make up the backbone of the RNA strand. The nitrogen bases (III) are attached to the ribose sugars.

# Honors Bio Essay Response

Respond to the following prompt using complete sentences. Images, Diagrams or flow charts should only be used to enhance responses.

Evolution is the change in allele frequencies in a population over time. We tend to think of evolution and natural selection as being synonymous, but in fact evolution can also occur through a variety of mechanisms, four of which are listed below:

Natural Selection

Genetic Drift

Mutation

Migration

- A. Define three of the four forces of evolution listed above, and give an explanation of each.
- B. You are studying a population of field mice that includes individuals with light and dark brown coats. Every six months you perform capture/recapture experiments to census the proportion of light and dark individuals. The following numbers indicate the percentage of dark coat mice caught in each successive census over the course of 5 years:

96

94

95

91

93

95

74

73

77

76

Give a hypothetical explanation based on ONE of the four forces of evolution which is having the biggest effect on the mice population.