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CHAPTER 3

Transmission of Heritable Information from Generation to Generation and Processes That Increase Genetic Diversity

Read This Chapter to Learn About

- DNA Is the Genetic Material
- Basic Mendelian Concepts
- Predicting Genotypes
- Exceptions to Mendel's Laws
- Pedigree Analysis
- Environmental Influences on Genes and Epigenetics
- Genetic Variability During Cell Division
- Mutations
- Evolution
- Genetic Basis for Evolution and the Hardy–Weinberg Equation
- Types of Natural Selection
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- Types of Evolution

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DNA IS THE GENETIC MATERIAL

A series of historic experiments collectively provided evidence that DNA is the genetic material of living things. A brief summary of those experiments follows.

Frederick Griffith worked with *Streptococcus pneumoniae* that exist in two forms: a virulent form (termed **smooth**) and a nonvirulent form (termed **rough**). He injected various combinations of smooth and rough *S. pneumoniae* into mice and observed the consequences, seen in Figure 3-1. Of note was that the injection of rough *S. pneumoniae* (which should not be virulent) and dead, smooth *S. pneumoniae* (which also should not be virulent) caused death in mice. The conclusion was that the dead, smooth bacteria were passing to the rough bacteria something that caused virulence, which the rough bacteria took up and expressed. Griffith called this the **transforming principle**.

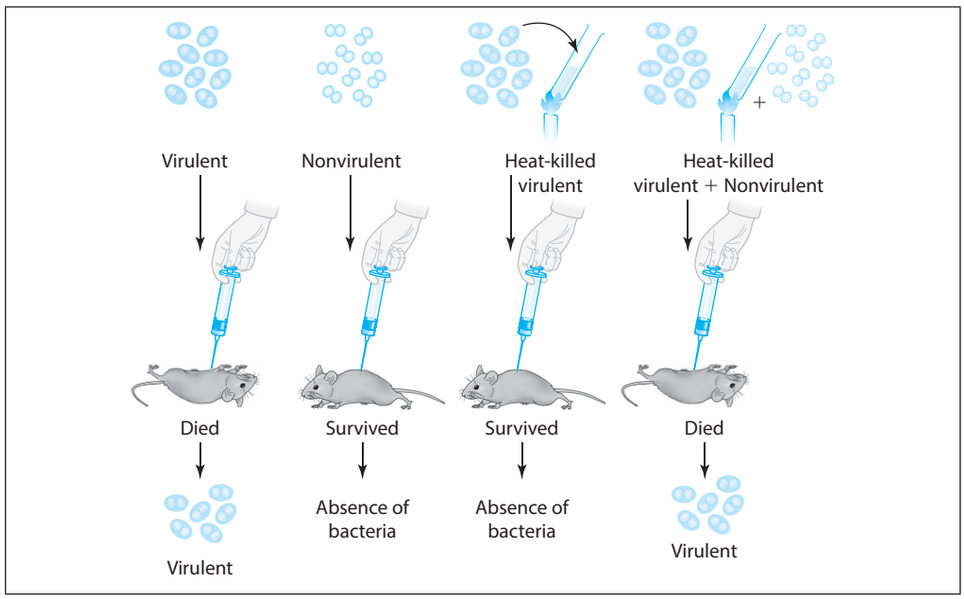

FIGURE 3-1 Griffith's experiments on the transforming principle.

Oswald Avery, Colin MacLeod, and Maclyn McCarthy followed up on Griffith's work by determining what the transforming principle was. Their work relied on the elimination of specific components in *S. pneumoniae* in order to determine which was responsible for transformation. Their results made it clear that DNA was the transforming factor.

Alfred Hershey and Martha Chase provided conclusive evidence that DNA is the genetic material of cells. They utilized the T2 bacteriophage and were able to radioactively label the DNA (using ^{32}P) and proteins (using ^{35}S) of T2 so that they could be tracked as T2-infected *Escherichia coli*, as seen in Figure 3-2. Following infection, they were able to track which material (DNA or proteins) entered the *E. coli*. In this way it was determined that DNA was the genetic material.

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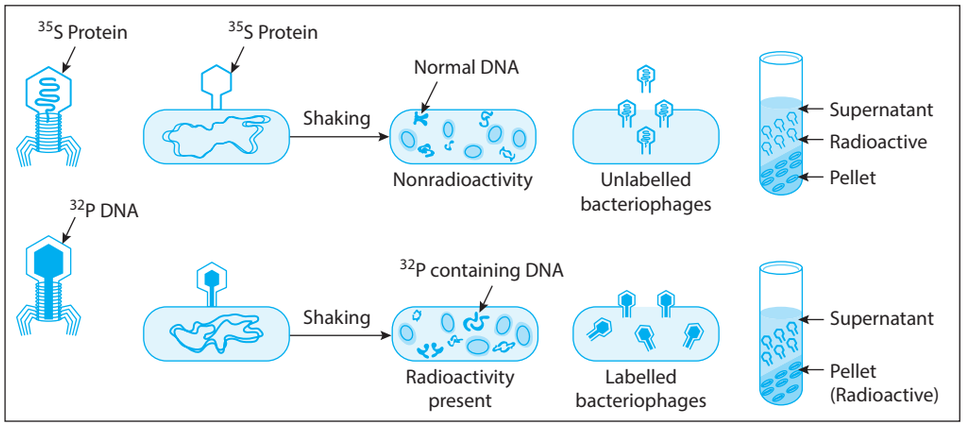

FIGURE 3-2 Hershey and Chase experiments.

BASIC MENDELIAN CONCEPTS

The basic principles of genetics were proposed by Gregor Mendel in the 1860s. His work with traits in pea plants led him to propose several theories of inheritance. Mendel did all his work and postulated his theories at a time when the genetic material had not even been discovered, so the fact that his theories hold true today could be considered quite a stroke of luck.

An understanding of some basic terminology is essential to discuss genetics. The exact genetic makeup of an individual for a specific trait is referred to as the **genotype**, while the physical manifestation of the genetic makeup is referred to as a **phenotype** for a specific trait. A **gene** has information to produce a single protein or enzyme. However, genes can exist in different forms termed **alleles**. In some cases, mutations can cause the production of alleles that produce faulty enzymes needed for metabolism. This leads to a class of genetic disorders known as inborn errors of metabolism.

Through his studies of pea plants, Mendel formulated several laws to explain how particular traits were inherited. These laws addressed issues concerning how specific traits were sorted and passed on to progeny and how some traits exerted dominance over others.

Mendel's Law of Segregation

One of Mendel's most important contributions was the **law of segregation**. There are several important ideas in this law. These ideas can be summarized as follows:

- For every given trait, an individual inherits two alleles for the trait (one from each parent).
- As an individual produces gametes, the two alleles segregate so that each gamete contains only a single allele per trait. During fertilization, each gamete contributes one allele per trait, providing the offspring with two alleles per trait.

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There are exceptions to the law of segregation. These include the alleles carried on sex chromosomes in males. Because males contain one X chromosome and one Y chromosome, the male does not have two alleles per trait for genes on the sex chromosomes. Another exception is that mitochondria contain their own DNA (in single copy) that is inherited separately from chromosomal DNA. Occasionally, alleles from the mitochondrial DNA may incorporate into the chromosomal DNA in a process termed **genetic leakage**.

Complete Dominance

Mendel also proposed the concept of dominance to explain how some traits are expressed, whereas others are hidden. Individuals can inherit two of the same allele (homozygous) or two different alleles (heterozygous) for any given trait. In the heterozygous individual, only one allele is normally expressed, while the other allele is hidden. The **dominant allele** is the one expressed, whereas the **recessive allele** is hidden in the presence of a dominant allele. When individuals are heterozygous for a particular trait, their phenotype appears dominant, yet they still carry and can pass on the recessive allele via their gametes. A recessive phenotype is only observed when the individual is homozygous for the recessive allele. Keep in mind that dominant traits are not necessarily more common or more advantageous than recessive traits. Those labels only refer to the pattern of inheritance that the allele follows and say nothing about the frequency of advantageousness of the allele. The most common allele in the population is usually referred to as wild type.

By convention, a single letter is selected to represent a particular trait. The dominant allele is always notated with a capital letter, and the recessive allele is notated with a lowercase letter. An example of possible allelic combinations can be seen in the following table.

TABLE 3-1 Possible Allelic Combinations

Alleles Inherited	Genotype	Phenotype
AA	Homozygous dominant	Dominant
Aa	Heterozygous	Dominant
aa	Homozygous recessive	Recessive

PREDICTING GENOTYPES

When the genotypes of both parents are known for a specific trait, the genotypes of the potential offspring can be determined. The tool used for this is known as the **Punnett square**.

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Punnett Squares

A **monohybrid cross** is a breeding between two parents (the P generation) in which a single trait is studied. The offspring of this cross are called the F_1 (first filial) generation. A breeding between two F_1 offspring produces the next generation, F_2, and so on. The potential gametes of each parent are determined and every possible combination of gametes is matched up on a matrix (the Punnett square) to determine every possible genotype of the potential offspring. A ratio of the offspring is expressed as dominant:recessive.

Mendel worked with many traits in the pea plant. He found that when he crossed a true breeding (homozygous) plant of a dominant phenotype to a true breeding plant of a recessive phenotype, 100% of the F_1 offspring had the dominant phenotype. However, when he bred two of the F_1 offspring, he found that 75% of the F_2 offspring had the dominant phenotype, yet 25% had the recessive phenotype. Although the recessive phenotype disappeared in the F_1 generation, it reappeared in the F_2 generation. The F_1 offspring were all heterozygous. When two heterozygotes are bred, the offspring will always show Mendel's observed 3:1 phenotypic ratio. A cross between two heterozygotes that results in a 3:1 phenotypic ratio can be seen in Figure 3-3.

TEST CROSSING

The genotype of a parent with a dominant phenotype can be determined using a method known as a **testcross** (also called a backcross). An organism with the dominant phenotype may be either homozygous or heterozygous. In the testcross, the parent with the dominant phenotype is always crossed to a homozygous recessive mate. The outcome of the phenotypic ratio of the offspring reveals the genotype of the unknown parent. If 100% of the offspring have the dominant phenotype, then the unknown parent is homozygous dominant. If the offspring display a 1:1 ratio, the genotype of the unknown parent is heterozygous. The possible outcomes of a testcross can be seen in Figure 3-4.

Mendel's Law of Independent Assortment

A **dihybrid cross** considers the inheritance of two different traits at the same time. The same rules of the monohybrid cross apply as long as the traits involved meet certain criteria. Those criteria are developed from **Mendel's law of independent assortment**, which states the following:

- The alleles must assort independently during gamete formation, meaning that the distribution of alleles for one trait has no influence on the distribution of alleles for the other trait.

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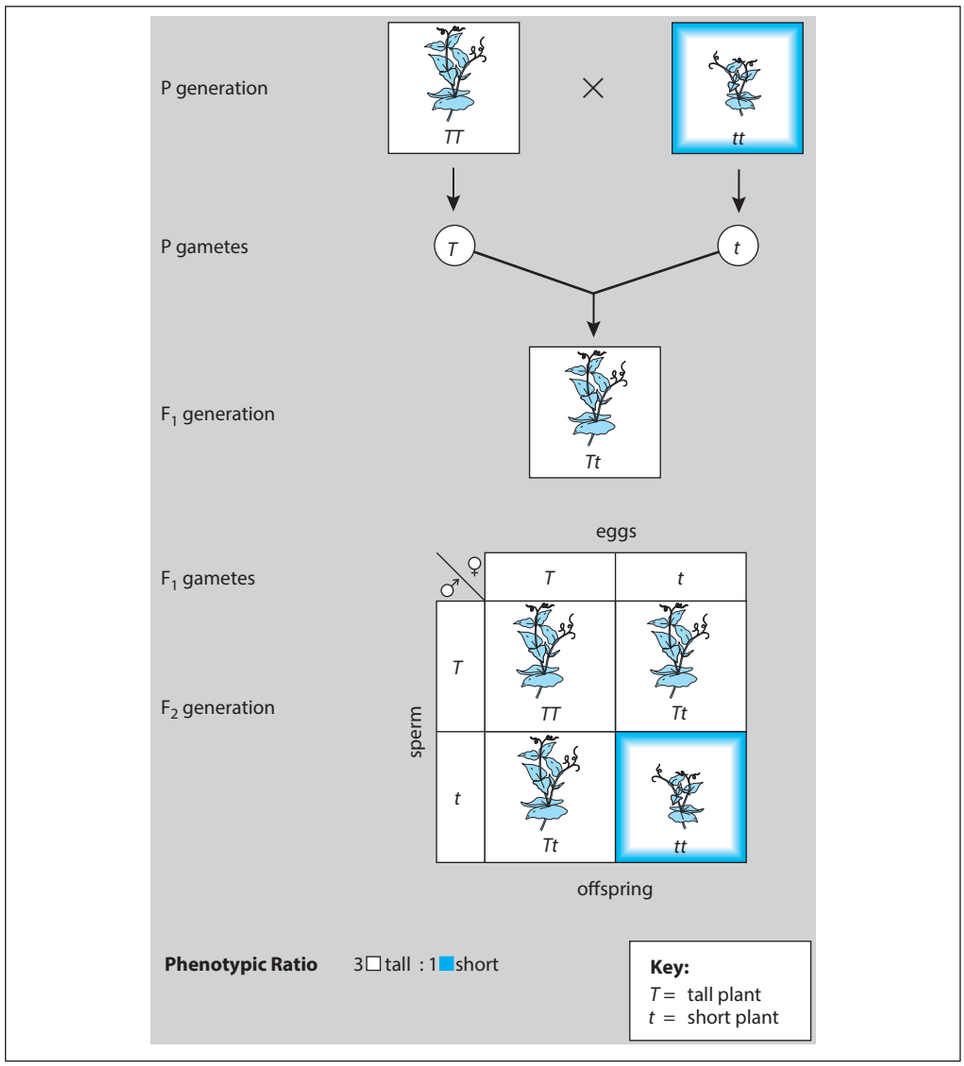

FIGURE 3-3 Monohybrid cross. The crossing of two heterozygous individuals leads to the typical 3:1 phenotypic ratio observed by Mendel. *Source:* From Sylvia S. Mader, *Biology*, 8th ed., McGraw-Hill, 2004; reproduced with permission of The McGraw-Hill Companies.

➤ If two genes are **linked**, meaning they occur on the same chromosome, they do not assort independently and thus are inherited together, changing the expected outcomes in the offspring.

Two unlinked traits can be considered together in a Punnett square. When two traits are involved in a dihybrid cross, each trait is assigned a different letter. To predict the possible offspring, all possible gamete combinations of each trait for the parents must be considered. Suppose two parents have the genotypes *AABB* and *aabb*. All F_1 offspring will be *AaBb*. If two F_1 offspring are bred, a 9:3:3:1 ratio will be seen in the F_2 generation. See Figure 3-5 for an example of a dihybrid cross.

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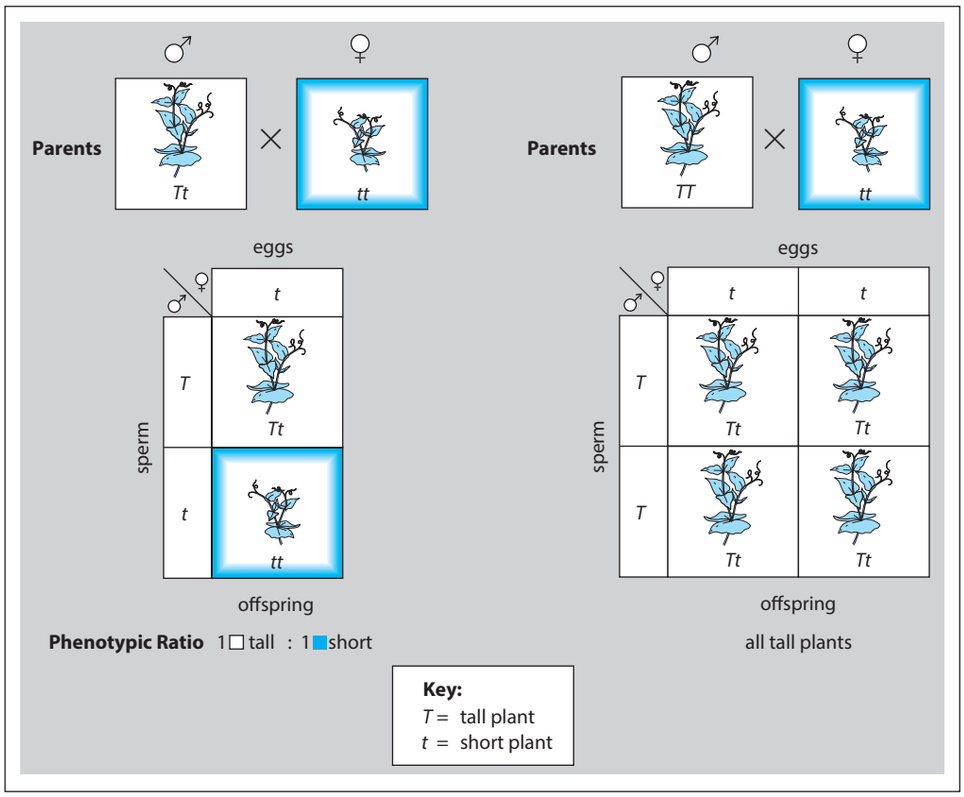

FIGURE 3-4 Testcross outcomes. (a) If the dominant phenotype parent is heterozygous, the ratio observed in the testcross is 1:1. (b) If the dominant phenotype parent is homozygous, all of the offspring exhibit the dominant phenotype. *Source:* From Sylvia S. Mader, *Biology*, 8th ed., McGraw-Hill, 2004; reproduced with permission of The McGraw-Hill Companies.

EXCEPTIONS TO MENDEL'S LAWS

Although Mendel's laws tend to be good predictors of inheritance for some genetic situations, sometimes these laws do not apply. Not every trait operates according to a simple dominant/recessive pattern or in a completely predictable manner. A summary of the genetic situations that are not predicted by Mendel's models can be found in the table on page 49.

Linked Genes

The location of a gene on a chromosome is referred to as the **locus** of the gene. Genes that are linked occur on the same chromosome, which means that if one allele is found in a gamete, the other is too since they are on the same chromosome. In the case of linkage, the combination of gametes produced is not as diverse as would be the case with nonlinked alleles. In some cases, the loci of the alleles are so close together that

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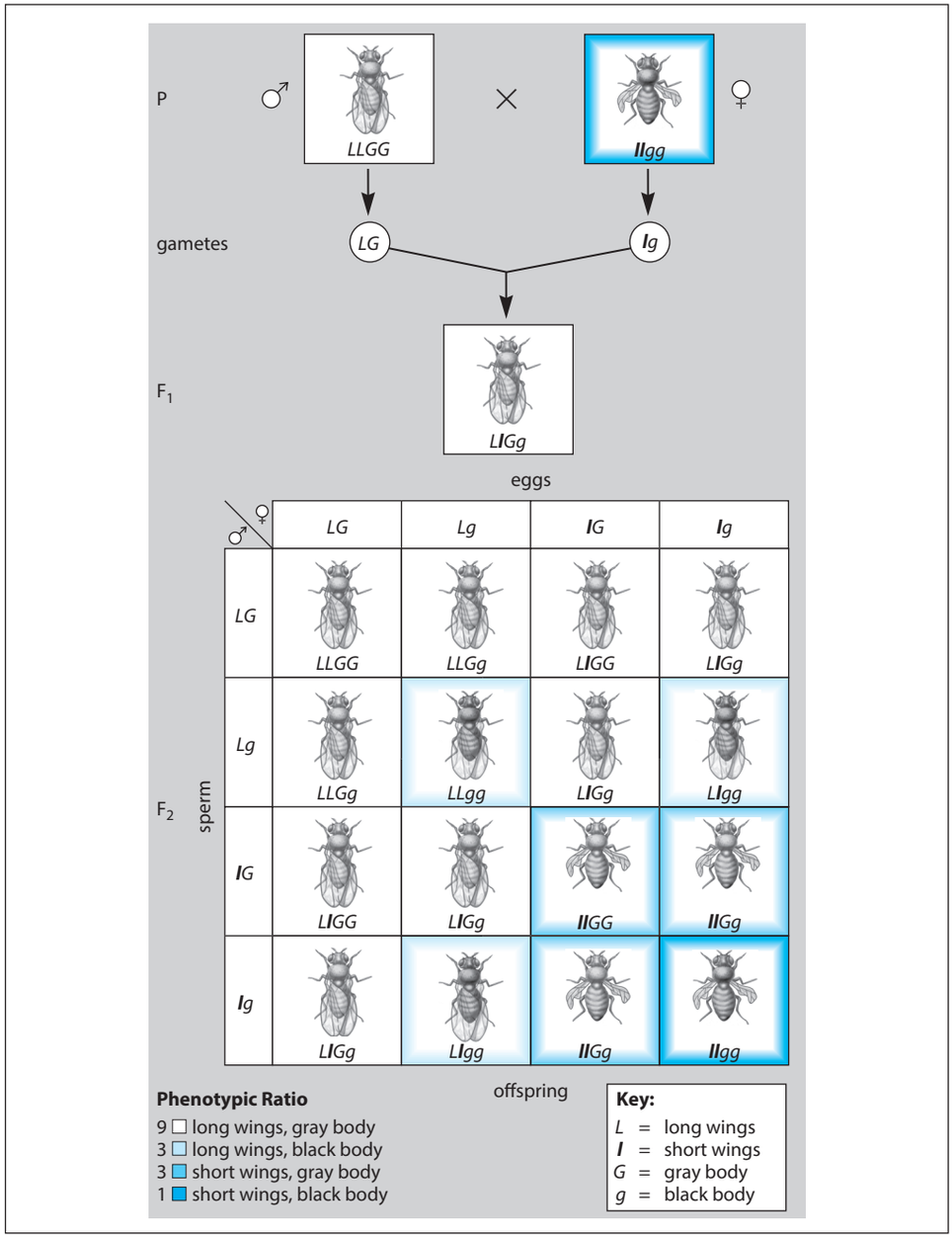

FIGURE 3-5 In a dihybrid cross, the inheritance of two unlinked traits are considered simultaneously. In this cross, Mendel's 9:3:3:1 phenotypic ratio is observed. *Source:* From Sylvia S. Mader, *Biology*, 8th ed., McGraw-Hill, 2004; reproduced with permission of The McGraw-Hill Companies.

they are always inherited together. However, if the loci of the alleles are far away from each other on the chromosome, then there is a possibility for crossing over and genetic recombination to occur. This process will be discussed in more detail with meiosis in a later section.

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TABLE 3-2 A Summary of Possible Genetic Situations

Genetic Situation	Key Characteristics	Examples
Simple dominant/recessive inheritance	One allele is dominant over the recessive allele. The only way to express the recessive phenotype is to be homozygous recessive. Individuals who are homozygous dominant or heterozygous express the dominant phenotype.	Mendel's traits observed in pea plants
Linked genes	These are separate genes located on the same chromosome. They do not assort independently and are generally inherited together. In the case that the linked genes are located far from each other on the same chromosome, there is a possibility for the genes to recombine during crossing over in meiosis.	Two genes located on the same chromosome
Multiple alleles	Some traits have more than two alleles to select from in the gene pool. Although an individual can receive only two alleles per trait (one from each parent), multiple alleles increase the diversity in the population.	Human blood type
Incomplete dominance	An individual who is heterozygous is expected to have a dominant phenotype. In incomplete dominance, both alleles are expressed somewhat so that the individual expresses a phenotype that is intermediate of the dominant and recessive phenotypes.	Snapdragon flower color
Codominance	An individual inherits two different alleles that are both dominant. Both alleles will be fully expressed, leading to an individual that expresses both dominant phenotypes.	Type AB blood in humans
Polygenic traits	More than one gene influences a single trait. This leads to multiple potential phenotypes.	Hair and skin color in humans
Epistasis	One gene can mask the presence of an expected phenotype of another gene.	Fur color in Labrador retriever dogs
Sex linkage	Recessive traits located on the single X chromosome in males are expressed, while they must be inherited on both X chromosomes to be expressed in women.	Color blindness

Multiple Alleles

For the traits Mendel observed with pea plants, there were always two alleles. One was dominant and one was recessive. Although an individual can inherit only two alleles (one from each parent) for any given trait, there is the possibility that there may be

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more than two alleles to select from in the gene pool, which consists of all genotypes in the population. These new alleles arise due to mutation and increase diversity in the population.

Human blood type is an example of multiple alleles. The ABO system has three alleles: I^A, I^B, and i. The alleles I^A and I^B are dominant, whereas the allele i is recessive. Each allele codes for either the presence or absence of particular antigens on the surface of red blood cells. With simple dominant/recessive traits, two phenotypes are expected—a dominant phenotype and a recessive phenotype. Any time multiple alleles are involved with a trait, more than two potential phenotypes will be expected. This is the case in blood type where four phenotypes can be observed: type A, type B, type AB, and type O.

Incomplete Dominance

According to Mendelian rules, a heterozygous individual always expresses the dominant phenotype. If alleles behave by incomplete dominance, this is not the case. Flower color in snapdragons is a classic example. If the allele *R* codes for red flowers and the allele *r* codes for white flowers, Mendelian rules would predict that the heterozygote (*Rr*) would have red flowers. However, because this trait behaves according to incomplete dominance, both alleles will be expressed to some degree, leading to a pink (intermediate) phenotype in the heterozygous offspring. In the case of incomplete dominance, only two alleles are involved, yet there are three potential phenotypes that can arise.

Codominance

Codominance is similar to incomplete dominance. For this to occur, the trait involved must first have multiple alleles and more than one of them must be dominant. If a heterozygous individual inherits two different dominant alleles, both alleles are expressed, leading to an individual who has both phenotypes (as opposed to a blended phenotype seen with incomplete dominance).

Human blood type is an example of codominance as well as multiple alleles. Should an individual inherit the genotype of I^AI^B, they will display the A phenotype as well as the B phenotype. In this case, the result is type AB blood. The following table contains more details on human blood type.

TABLE 3-3 The Genetic Basis of Human Blood Types

Blood Type	Potential Genotypes	Antigens Found on the Red Blood Cell Surface
Type A	I^AI^A or I^Ai	A
Type B	I^BI^B or I^Bi	B
Type AB	I^AI^B	A and B
Type O	Ii	none

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Polygenic Traits

Generally, a single gene influences one trait. **Polygenic traits** involve gene interaction. This means that more than one gene acts to influence a single trait. Skin color and hair color are both examples of polygenic traits in humans. Because more than one gene is involved, the number of potential phenotypes is increased, resulting in continuous variation.

Epistasis

Epistasis is a unique genetic situation where one gene interferes with the expression of another gene. In many cases, epistasis can lead to the masking of an expected trait. An example is coat color in Labrador retrievers. These dogs have black, chocolate, or yellow fur. In addition to the gene that controls fur color, the *B* gene, there is another allele that controls how pigment is distributed in the fur, the *E* gene. The *B* gene produces an enzyme that processes brown pigment to black pigment. Dogs that have the genotype *BB* or *Bb* produce black pigment, while those with the genotype *bb* produce brown pigment. The *E* gene allows the pigment to be deposited into the hair follicle. If the Labrador is *EE* or *Ee*, it is able to deposit the pigment. However, dogs with the genotype *ee* will not. Therefore, the gene *B* determines if a dog produces black or brown pigment, but these phenotypes can be expressed only if the dog is homozygous dominant or heterozygous for the *E* gene. Any dog that is homozygous recessive for the *E* gene, *ee*, will be yellow.

Pleiotropy

Pleiotropy occurs when a single gene influences two or more other traits. Most frequently, the effects of pleiotropy are seen in genetic diseases. In sickle cell disease, the mutation in the hemoglobin gene results in the production of hemoglobin protein with a reduced oxygen-carrying ability. This, in turn, affects multiple organ systems in the body, explaining the multiple symptoms of the disease.

Sex Linkage

When alleles are found on the X and Y sex chromosomes, the normal rules of genetics may not apply. Although the sex chromosomes do contain genes to influence gender, there are other traits found on these chromosomes that have nothing to do with gender. Women inherit an XX genotype, whereas men inherit an XY genotype. In men, traits that occur on the sex chromosomes are the exception to the normal rule of always having two alleles per trait. Because the sex chromosomes in men are not a true pair, they do not have two alleles per trait on their sex chromosomes. The Y chromosome contains relatively few genes as compared to the X chromosome.

When a recessive trait is located on the X chromosome, women must receive two copies of the recessive allele (one from each parent) to express the recessive trait.

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However, men who inherit a recessive allele on their only X chromosome will express the recessive phenotype. Color blindness and hemophilia are examples of traits that are sex linked. While women can express these traits, to do so they must receive the recessive alleles on both X chromosomes (meaning they must receive it from both of their parents). Therefore, these traits are more commonly observed in men, as they only need to receive the recessive trait on their single X chromosome.

Women who are heterozygous for a trait on the X chromosome do not express the trait; yet they are carriers for this and can pass the traits to their sons. Since women are genotypically XX, every egg cell they make contains the X chromosome. Men are XY and thus half their sperm contain the X chromosome and half contain the Y. In males, the Y chromosome must come from the father and the X comes from the mother.

PEDIGREE ANALYSIS

A **pedigree** is a diagram used to help determine a pattern of inheritance over multiple generations. In pedigrees, males are indicated by a square and females by a circle. Individuals in the pedigree who are affected by a certain trait are indicated by shading in the square or circle. Those that are not affected are indicated by no shading in the square or circle. In some cases, a square or circle may be half shaded, and this is to indicate a carrier or a heterozygote. Horizontal lines indicate matings, and vertical lines show offspring. An example of a pedigree indicating recessive inheritance is seen in Figure 3-6.

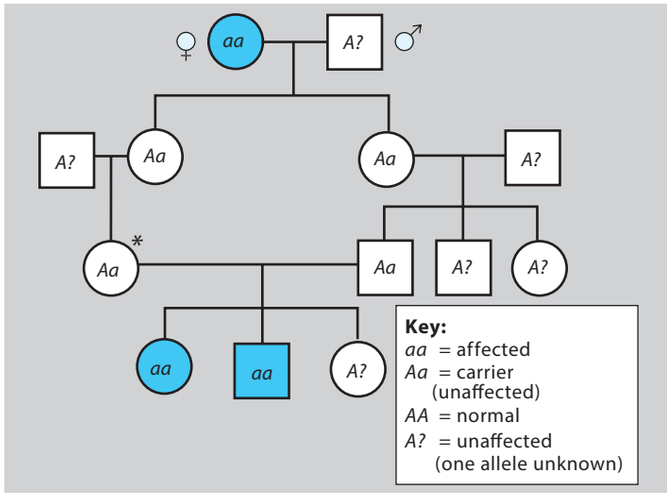

FIGURE 3-6 This pedigree shows a recessive pattern of inheritance. *Source:* From Sylvia S. Mader, *Biology*, 8th ed., McGraw-Hill, 2004; reproduced with permission of The McGraw-Hill Companies.

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If a pedigree shows many more males than females being affected, sex-linked inheritance should be suspected. If males and females both seem equally affected, look for skipping of generations. Dominant traits usually appear in each generation, whereas recessive traits often skip generations.

ENVIRONMENTAL INFLUENCES ON GENES AND EPIGENETICS

Although some genes behave according to very predictable rules, there are many cases where some external or internal environmental factor can interfere with the expression of a particular genotype. **Penetrance** of a genotype is a measure of the frequency at which a trait is actually expressed in the population. If a trait were described at 80% penetrance, it would mean that 80% of the people with the genotype for the particular trait would have the phenotype associated with the genotype. While some traits always show 100% penetrance, others do not. Within an individual, expressivity is a measure of the extent of **expression** of a phenotype. This means that, in some cases, expression of a phenotype is more extreme than others.

There are many examples of how the environment affects the expression of a particular phenotype. Hydrangea plants may have the genotype to produce blue flowers, but depending on the acidity of the soil that they are grown in (an environmental factor), they may express a different phenotype than expected (such as pink flowers). Women who have the BRCA 1 and 2 alleles are at a high but not guaranteed risk for developing breast cancer, meaning that something other than the allele determines the expression of the allele.

Many traits cannot be predicted by genotype alone (like intelligence, emotional behavior, and susceptibility to cancer). In many cases, the interaction of genes and the environment is a complicated relationship that is impossible to predict. Factors in humans such as age, gender, diet, and so forth are all factors known to influence the expression of certain genotypes. This is the concern of the field of epigenetics.

Epigenetics involves the study of heritable changes in gene expression that are not caused by DNA sequence changes. These changes can have many sources but frequently involve patterns of DNA methylation and histone modification that influence gene expression. These changes may involve down-regulation or up-regulation of gene expression. These patterns that influence gene expression are referred to as the **epigenome**, and they may be heritable for many generations. However, the epigenome is known to show change over time, particularly during the process of cellular differentiation during embryonic development.

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GENETIC VARIABILITY DURING CELL DIVISION

The cell is the basic unit of structure and function in an organism. For life to continue, cells must divide and reproduce. Cell division in eukaryotes happens through two processes: mitosis and meiosis. **Mitosis** is normal cell division used for growth and the replacement of cells. In mitosis, a parent cell is copied to produce two identical daughter cells.

There are times that producing genetically identical offspring cells is not appropriate, such as during sexual reproduction. During the process of sexual reproduction, genetically diverse gametes must be created. These gametes are produced by the process of meiosis. Both mitosis and meiosis have many features in common. The processes of mitosis and meiosis will be considered in detail in Chapter 7.

Cytoplasmic Inheritance

Recall that certain organelles such as mitochondria contain their own DNA. This DNA is circular and in single copy; therefore, there are no pairs of alleles—just a single copy of each allele. Any genes present on mitochondrial DNA will be inherited by the daughter cells during cytokinesis of mitosis or meiosis. Unlike chromosomal inheritance, any genes passed through mitochondrial DNA will not follow the normal Mendelian laws of genetics.

Mistakes in Meiosis

While the technicalities of meiosis will be discussed in Chapter 7, this section will discuss the consequences of meiotic mistakes in the context of factors that influence genetic variability. Mistakes that happen during meiosis can have drastic consequences. Because the gametes are used for reproduction, any chromosomal damage to the gametes is passed on to the next generation. There are several ways in which mistakes can occur, changing the number of chromosomes or damaging them.

If chromosomes fail to separate properly during meiosis, a **nondisjunction** has occurred. This leads to gametes that have the wrong number of chromosomes. If those gametes are fertilized, the resulting embryo will have the wrong diploid number. An example of this is **Down syndrome**, which often is the result of a nondisjunction in the female gamete. If a female egg contains 24 chromosomes instead of the expected 23 and is fertilized by a normal sperm, the resulting embryo has 47 chromosomes, which is one more than expected. This condition is referred to as a **trisomy**. In the case where a gamete is missing a chromosome as the result of a nondisjunction and is fertilized by a normal gamete, the result is an embryo with 45 chromosomes. This is termed a **monosomy**. With the exception of Down syndrome, which is a trisomy of human chromosome 21 (which is very small) and certain trisomies and monosomies of the

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sex chromosomes (X and Y), most embryos with trisomies and monosomies do not survive development.

Other forms of chromosomal damage can occur in meiosis. They typically have serious if not fatal consequences. They are as follows:

- **Deletion.** This occurs when a portion of a chromosome is broken off and lost during meiosis. Although the total chromosome number is normal, some alleles are lost.
- **Duplication.** This is when a chromosome contains all of the expected alleles and then receives a duplication of some alleles.
- **Inversion.** This occurs when a portion of a chromosome breaks off and reattaches to the same chromosome in the opposite direction.
- **Translocation.** This occurs when a portion of a chromosome breaks off and reattaches to another chromosome.

MUTATIONS

Mutations occur naturally through the process of DNA replication, but certain factors can greatly increase the spontaneous mutation rate. These factors are referred to as **mutagens**. The mutagens that are linked to development of cancer are called carcinogens. Should any of these mutations occur in gametes (germ cells), they would be considered heritable and are passed on to the next generation.

Mutations change the coding sequence of DNA. When the DNA changes, the mRNA codons change, and the amino acid sequence of the protein made may change. In some cases, this may produce a protein that functions better than the one intended by the DNA (thus providing an advantage), one that functions equivalently to the intended protein, or in the worst case, a protein that functions worse than the intended protein or does not function at all. Recall that mutations happen spontaneously and that the rate of mutation is increased by exposure to mutagens.

Mutations can occur in several ways—by the change in a single nucleotide, the mispairing of nucleotides, the addition or deletion of a nucleotide, or the movement of nucleotides. While these changes typically happen at the DNA level, there can also be transcription and translation errors that occur.

Point Mutations

When a single nucleotide is swapped for another, the resulting mutation is termed a **point (substitution) mutation**. This ultimately changes a single codon on the mRNA. In some cases, this mutation is silent, meaning that if the codon is changed and still codes for the intended amino acids, there will be no detectable consequence. However, sometimes even a single point mutation can have major consequences. If the new

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codon (a missense codon) codes for a different amino acid than what was intended, the new protein may not function properly. This can lead to a genetic disease such as sickle cell. It is also possible for a change in a single nucleotide (nonsense codon) to produce a stop codon in a new location, causing a nonsense mutation. In this case, the protein produced would be too short and most likely nonfunctional.

Frameshift Mutations

A **frameshift mutation** is the result of the addition or deletion of nucleotides. Unlike the point mutation where the overall number of nucleotides does not change, adding or deleting changes the number of nucleotides. Because mRNA is read in codons, an addition or deletion alters all of the codons from the point of the mutation onward. This disrupts the reading frame of the mRNA. Since many codons are changed, the frameshift mutation generally produces a damaged or nonfunctional protein.

EVOLUTION

Evolution simply means change. The changes referred to are genetic ones, thus putting the concept of mutation at the center of the process of evolution. Evolution is something that occurs over time, so a single individual does not evolve, but populations of individuals do evolve. **Microevolution** deals with genetic changes within a population, whereas **macroevolution** is concerned with changes that occur to a species on a larger scale over a longer period of time.

Mechanisms for Evolution

A variety of factors are responsible for the microevolution of a particular population. Natural selection, based on mutation, tends to be the major driving force for evolution, while genetic drift and gene flow can also influence the process.

MUTATION

New alleles are created by **mutation**. These new alleles code for proteins that may be beneficial, neutral, or detrimental as compared to the original protein intended by the allele. New alleles that code for beneficial proteins can provide advantages that are ultimately selected for by natural selection and are passed to the next generation, whereas detrimental alleles are selected against.

NATURAL SELECTION

A central concept to the process of how evolution occurs is that of Darwin's **natural selection**. Natural selection explains the increase in frequency of favorable alleles from

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one generation to the next. This results from differential reproductive success in which some individuals reproduce more often than others and thus are selected for. This increases the frequency of their alleles in the next generation. Those that reproduce less decrease the frequency of their alleles in the next generation.

Fitness Concept. The concept of **evolutionary fitness** is key to natural selection. In this context, **fit** refers to the reproductive success of an individual and their allelic contribution to the next generation. Those individuals who are more fit are more evolutionarily successful because their genetic traits are passed to the next generation, thus increasing the frequency of specific alleles in the gene pool.

Over generations, selective pressures that are exerted on a population can lead to adaptation. When selective pressures change, some organisms that may have been considered marginally fit before may now have increased fitness under the new conditions. Further, those individuals that may have been very fit previously may drastically decrease their fitness. Their genetic adaptations will be selected against. While individuals cannot change their genetics, over time, the population changes genetically, which is termed **adaptation**.

Differential Reproductive Success and Competition. Competitive interactions within a population are another critical factor for natural selection. The ability to outcompete other individuals for resources, including mates, is a key feature of fitness. In any given population, some individuals are better able to compete for resources and are considered more fit than others. This leads to **differential reproductive success**. This concept assumes that mating in the population is random. In some cases, such as with humans, mating is nonrandom, which leads to another form of selection to be discussed shortly.

Competition between species can also influence the evolutionary progression of all species involved. In some cases, **symbiotic relationships** exist where two species exist together for extended periods of time. In **mutualistic relationships**, both species benefit from the association. In **parasitic relationships**, one species benefits at the expense of the other species. In **commensalism**, one species benefits while the other species is relatively unharmed.

When two species are competing for the same ecological requirements or niche, the reproductive success and fitness, as well as the growth of one or both populations, may be inhibited based on the ability to compete for resources. This will change the microevolutionary course of the population.

GENE FLOW

When individuals leave a population, they take their alleles with them, resulting in **gene flow**. This can decrease genetic variation within the gene pool of the population, ultimately affecting the evolution of the population. Outbreeding occurs with the

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individuals that leave the population. They can add diversity to the gene pool of their new populations by adding alleles to it.

GENETIC DRIFT

Genetic drift involves changes to the allelic frequencies within a population due to chance. Although this is generally negligible in large populations, it can have major consequences in smaller populations. The **bottleneck effect** is a form of genetic drift where catastrophic events may wipe out a large percentage of a population. When the population is small, the few remaining alleles in the gene pool may not be characteristic of the larger population. The **founder effect** is a form of genetic drift that occurs when a small number of individuals leave a larger population and form their own small population where inbreeding is necessary. The new population only has the diversity brought to it by the founding members.

GENETIC BASIS FOR EVOLUTION AND THE HARDY–WEINBERG EQUATION

The **Hardy–Weinberg equation** can be used to calculate allelic frequencies within a population given the population is large and microevolution is not occurring—not necessarily a realistic situation. The Hardy–Weinberg equation is expressed as:

$$p^2 + 2pq + q^2 = 1$$

where p represents the frequency of a dominant allele and q represents the frequency of a recessive allele such that $p + q = 1$.

The equation can be used to show the frequency of homozygous dominant individuals (p^2), the frequency of heterozygotes ($2pq$), and the frequency of homozygous recessive individuals (q^2). Given information on the frequency of a single allele, all other pieces within the equation can be determined. Although these frequencies are temporarily accurate, any evolution occurring within the population would shift these predicted values.

For Hardy–Weinberg allelic frequencies to hold true, it is necessary that certain criteria be met. If any of these criteria are violated, the allelic frequencies will change over time. Any of the following will negate Hardy–Weinberg equilibrium:

- Nonrandom mating
- Gene flow
- Populations with a small number of individuals
- Mutations
- Bottleneck effect
- Founder effect

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TYPES OF NATURAL SELECTION

For any given trait, there can be several different phenotypes. If two phenotypes are present for a particular trait, **dimorphism** is the case. If three or more phenotypes are seen for a particular trait, **polymorphism** is at work. For example, flower color in snapdragons exhibits polymorphism with red, white, and pink phenotypes. Some phenotypes can be considered intermediates (like pink flowers) or can be extremes from either end of the intermediate phenotype (like red and white flowers). When natural selection occurs, it may select for intermediate phenotypes, either extreme phenotype, or both extreme phenotypes as seen in Figure 3-7.

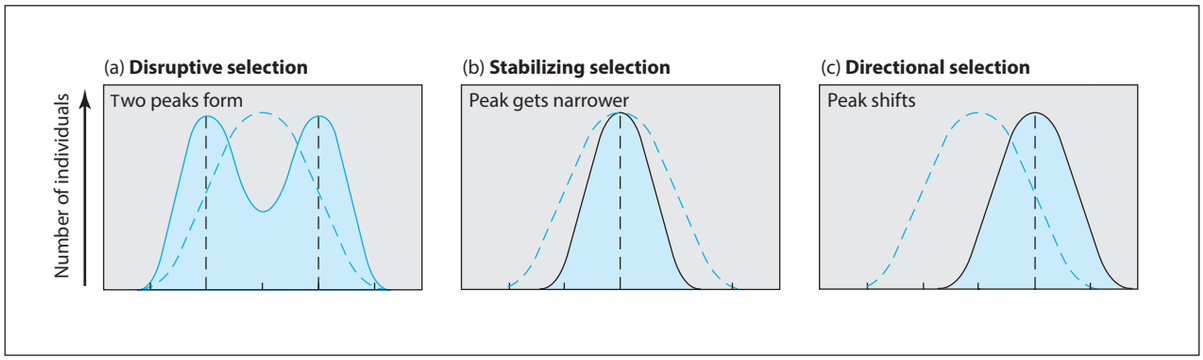

FIGURE 3-7 Types of natural selection. (a) In disruptive selection, the extreme phenotypes are selected for, whereas the intermediate phenotype is selected against. (b) In stabilizing selection, the intermediate phenotype is favored. (c) In directional selection, one extreme phenotype is favored. *Source:* From George B. Johnson, *The Living World*, 3rd ed., McGraw-Hill, 2003; reproduced with permission of The McGraw-Hill Companies.

Disruptive Selection

In some cases, the environment favors two extreme phenotypes simultaneously. In this case, **disruptive selection** occurs, where individuals with either extreme phenotype are favored, while those with the intermediate forms of the alleles are selected against. Over time, the continued selection of both extremes may eventually lead to the evolution of two distinct species.

Stabilizing Selection

Stabilizing selection leads to favoring the alleles that produce an intermediate phenotype. Human birth weight would be an example of stabilizing selection. Babies of an intermediate weight are favored over those who are too small to survive or too large to be easily delivered.

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Directional Selection

In **directional selection**, an allele that is considered advantageous is selected for. The allelic frequency continues to shift in the same direction generation after generation. In this case, one allele that produces an extreme phenotype is selected for. The selection of antibiotic resistance alleles in bacteria is an example of directional selection. Over time, selective pressures can result in an entire population possessing the same allele for a particular trait.

Artificial Selection

When particular alleles are purposely selected for based on nonrandom mating, **artificial selection** occurs. The breeding of domesticated dogs is an excellent example of the results of artificial selection. All breeds of dogs are members of the same species, all of which have been selectively bred from wolves for specific traits that are appealing to the breeder. Both toy poodles and Great Danes are examples of the extreme phenotypes that can be selected for when artificial selection is used. Traits that are artificially selected for are not necessarily the result of the most fit alleles. Many breeds of dog have medical conditions or predispositions as a result of artificial selection.

SPECIATION

Natural selection can ultimately result in the formation of new species. By definition, a **species** is a group of individuals who can breed with each other and not with members of other species. When a population becomes geographically isolated from each other, members of the same species may evolve differently in different locations. This is referred to as **allopatric speciation**. Even when geographic barriers do not exist to divide a population, there are factors that can ultimately prevent some members of the population from breeding with others. This is **sympatric speciation**. Over time, they may evolve into two different species that can no longer breed with each other.

There are a variety of mechanisms that occur to prevent interbreeding between species. When these mechanisms do not work, hybrid species may occur.

Prezygotic Isolation

Prezygotic isolation mechanisms occur to prevent fertilization between the gametes of members of two different species. They are as follows:

- **Temporal isolation.** Two different species may live in the same environment but have different breeding seasons or may have overlapping breeding seasons but breed during different times of the day.

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- **Ecological isolation.** The two species live in different habitats and thus rarely encounter each other to breed.
- **Behavioral isolation.** The mating behaviors of the two species are not compatible with each other.
- **Reproductive isolation.** Even if members of different species attempt to mate, their reproductive structures may not be compatible.
- **Gamete isolation.** The gametes of one species cannot fertilize the gametes of the other species, so reproduction is unsuccessful even if successful copulation occurs.

Postzygotic Isolation

If prezygotic isolation mechanisms fail, there are a variety of mechanisms that occur after fertilization to prevent successful reproduction between members of different species. The **postzygotic isolation** mechanisms are as follows:

- **Hybrid inviability.** If fertilization occurs between the gametes of two different species, the zygote will not be able to continue in development.
- **Hybrid sterility.** If fertilization and subsequent development successfully occurs, the hybrid offspring is sterile and unable to reproduce.
- **Hybrid breakdown.** Some hybrid offspring are fertile and can reproduce. However, the second-generation offspring are infertile.

TYPES OF EVOLUTION

The evolutionary process can proceed in a variety of directions or patterns such as convergent, divergent, and parallel evolution. These gradual and random changes in the genome are how evolutionary time is measured.

Convergent Evolution

When two populations exist in the same type of environment that provides the same selective pressures, the two populations will evolve in a similar manner via **convergent evolution**. While the populations may not be closely related, they may develop similar analogous structures to allow them to function in similar environments. Fish in Antarctica have evolved the ability to produce specialized glycoproteins that serve as a sort of antifreeze to prevent their tissues from freezing in the low-temperature water. Fish on the opposite side of the world, in the Arctic, have evolved the same kind of antifreeze protection mechanism. Genetic studies show that the two species of fish produce antifreeze proteins that are very different from each other, which strongly suggests that two independent events led to the evolution of these mechanisms.

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Divergent Evolution

In an individual population, it is possible that individuals within the population evolve differently. Over time, this may lead to the development of new species via **divergent evolution**. In many cases, changes to the population or geographic isolation may cause different adaptations within the population. This sort of evolution can lead to homologous structures. Vertebrate limbs are an excellent example of divergent evolution. The forearms of different vertebrate species have different structures and functions; however, they all diverged from a common origin.

Parallel Evolution

When two species share the same environment, the evolution of one species can affect the evolution of the other species. This is called **parallel evolution** or coevolution. Any changes to one species will require adaptations to the other species for them to continue to exist in the same environment. An example might be how the predation patterns of birds might influence the evolution of butterfly species sharing the same space. Some butterflies have evolved the ability to store poisonous chemicals that deter birds from eating them, while other butterflies simply mimic the poisonous ones to avoid being preyed upon.