

BIOL 30: GENETICS



Princess Gilbert
Monterey Peninsula College

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Licensing

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CHAPTER OVERVIEW

1: Introduction to Genetics

1.1: Genetics Overview

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1.1: Genetics Overview

Learning Objectives

- Define genes, genome, chromosomes and karyotype
- Explain how DNA carries and maintains genetic information
- Describe DNA sequencing and genomics
- Discuss how genomics can be useful for fields such as conservation biology, forensic science and epidemiology

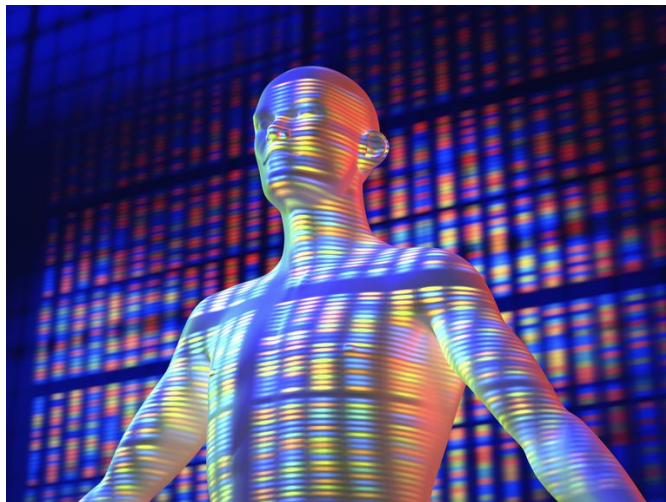


Figure 1.1.1: The Human Genome Project (HGP) made a map of the genes of a human being. (Copyright; author via source)

We are living in the “Post-genomic era” in which you can pay a company like “23 and Me” or “Ancestry” \$100-200, spit into a tube, and in about two months, find out everything you need to know about your ancestry, genetic traits, risks for getting certain genetic diseases, etc. These advances would not be possible without the efforts of scientists across the world studying human genetics and genomics. The US National Institute of Health (NIH) and the US Department of Energy (DOE) launched the Human Genome Project (HGP) in 1990, an international consortium with participating scientists from the UK, France, Australia, China, Germany, etc. aimed at sequencing the entire human genome. Scientists started out mapping the human genome (Figure 1), by determining the locations of many genes on the 23 haploid chromosomes. They then determined the nucleotide sequences of the genes (called coding sequences) and other non-coding sequences in the genome. The project took 13 years (completed in 2003), and provided the first blueprint of the human genome.

Genetics – is the scientific study of heredity and the variation of inherited characteristics. It includes the study of genes, themselves, how they function, interact, and produce the visible and measurable characteristics we see in individuals and populations of species as they change from one generation to the next, over time, and in different environments.

Heredity – Humans have always been aware that the characteristics of an individual plant or animal in a population could be passed down through the generations. Offspring look more like their parents. Humans also knew that some heritable characteristics (such as the size or color of fruit) varied between individuals, and that they could select or breed crops and animals for the most favorable traits. Knowledge of these hereditary properties has been of significant value in the history of human development. In the past, humans could only manipulate and select from naturally existing combinations of genes. More recently, with the discovery of the substance and nature of genetic material, DNA, we can now identify, clone, and create novel, better combinations of genes that will serve our goals. Understanding the mechanisms of genetics is fundamental to using it wisely and for the betterment of all.



Fig.1-1: Parent and offspring. Wolf's Monkey. (Flickr-eclectic echos-CC:AND)



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2: Biological Molecules

The large molecules necessary for life that are built from smaller organic molecules are called biological macromolecules. There are four major classes of biological macromolecules (carbohydrates, lipids, proteins, and nucleic acids), and each is an important component of the cell and performs a wide array of functions. Combined, these molecules make up the majority of a cell's mass. Biological macromolecules are organic, meaning that they contain carbon (with some exceptions, like carbon dioxide). In addition, they may contain hydrogen, oxygen, nitrogen, phosphorus, sulfur, and additional minor elements.

Carbon

It is often said that life is “carbon-based.” This means that carbon atoms, bonded to other carbon atoms or other elements, form the fundamental components of many, if not most, of the molecules found uniquely in living things. Other elements play important roles in biological molecules, but carbon certainly qualifies as the “foundation” element for molecules in living things. It is the bonding properties of carbon atoms that are responsible for its important role.

Carbon Bonding

Carbon contains four electrons in its outer shell. Therefore, it can form four covalent bonds with other atoms or molecules. The simplest organic carbon molecule is methane (CH_4), in which four hydrogen atoms bind to a carbon atom (Figure 2.1).

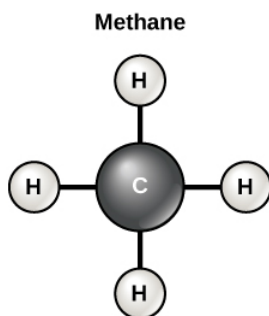
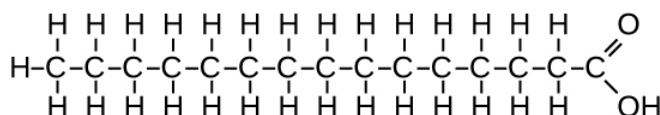
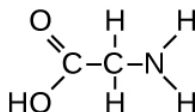


Figure 2.1: Carbon can form four covalent bonds to create an organic molecule. The simplest carbon molecule is methane (CH_4), depicted here.

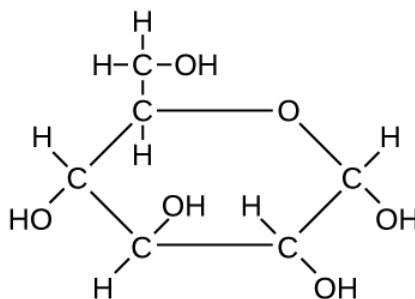
However, structures that are more complex are made using carbon. Any of the hydrogen atoms can be replaced with another carbon atom covalently bonded to the first carbon atom. In this way, long and branching chains of carbon compounds can be made (Figure 2.2a). The carbon atoms may bond with atoms of other elements, such as nitrogen, oxygen, and phosphorus (Figure 2.2b). The molecules may also form rings, which themselves can link with other rings (Figure 2.2c). This diversity of molecular forms accounts for the diversity of functions of the biological macromolecules and is based to a large degree on the ability of carbon to form multiple bonds with itself and other atoms.



(a)



(b)



(c)

Figure 2.2: These examples show three molecules (found in living organisms) that contain carbon atoms bonded in various ways to other carbon atoms and the atoms of other elements. (a) This molecule of stearic acid has a long chain of carbon atoms. (b) Glycine, a component of proteins, contains carbon, nitrogen, oxygen, and hydrogen atoms. (c) Glucose, a sugar, has a ring of carbon atoms and one oxygen atom.

Carbohydrates

Carbohydrates are macromolecules with which most consumers are somewhat familiar. To lose weight, some individuals adhere to “low-carb” diets. Athletes, in contrast, often “carb-load” before important competitions to ensure that they have sufficient energy to compete at a high level. Carbohydrates are, in fact, an essential part of our diet; grains, fruits, and vegetables are all natural sources of carbohydrates. Carbohydrates provide energy to the body, particularly through glucose, a simple sugar. Carbohydrates also have other important functions in humans, animals, and plants.

Carbohydrates can be represented by the formula $(CH_2O)_n$, where n is the number of carbon atoms in the molecule. In other words, the ratio of carbon to hydrogen to oxygen is 1:2:1 in carbohydrate molecules. Carbohydrates are classified into three subtypes: monosaccharides, disaccharides, and polysaccharides.

Monosaccharides (mono- = “one”; sacchar- = “sweet”) are simple sugars, the most common of which is glucose. In monosaccharides, the number of carbon atoms usually ranges from three to six. Most monosaccharide names end with the suffix -ose. Depending on the number of carbon atoms in the sugar, they may be known as trioses (three carbon atoms), pentoses (five carbon atoms), and hexoses (six carbon atoms).

Monosaccharides may exist as a linear chain or as ring-shaped molecules; in aqueous solutions, they are usually found in the ring form.

The chemical formula for glucose is $C_6H_{12}O_6$. In most living species, glucose is an important source of energy. During cellular respiration, energy is released from glucose, and that energy is used to help make adenosine triphosphate (ATP). Plants synthesize glucose using carbon dioxide and water by the process of photosynthesis, and the glucose, in turn, is used for the energy requirements of the plant. The excess synthesized glucose is often stored as starch that is broken down by other organisms that feed on plants.

Galactose (part of lactose, or milk sugar) and fructose (found in fruit) are other common monosaccharides. Although glucose, galactose, and fructose all have the same chemical formula ($C_6H_{12}O_6$), they differ structurally and chemically (and are known as isomers) because of differing arrangements of atoms in the carbon chain (Figure 2.3).

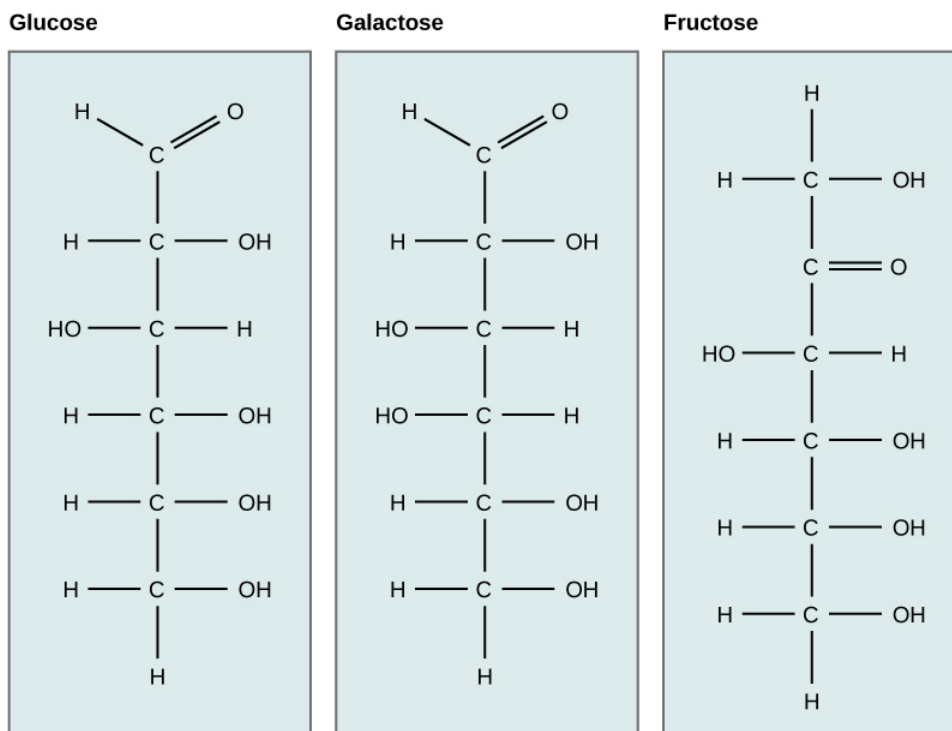


Figure 2.3: Glucose, galactose, and fructose are isomeric monosaccharides, meaning that they have the same chemical formula but slightly different structures.

Disaccharides (di- = “two”) form when two monosaccharides undergo a dehydration reaction (a reaction in which the removal of a water molecule occurs). During this process, the hydroxyl group ($-OH$) of one monosaccharide combines with a hydrogen atom of another monosaccharide, releasing a molecule of water (H_2O) and forming a covalent bond between atoms in the two sugar molecules.

Common disaccharides include lactose, maltose, and sucrose. Lactose is a disaccharide consisting of the monomers glucose and galactose. It is found naturally in milk. Maltose, or malt sugar, is a disaccharide formed from a dehydration reaction between two glucose molecules. The most common disaccharide is sucrose, or table sugar, which is composed of the monomers glucose and fructose.

A long chain of monosaccharides linked by covalent bonds is known as a polysaccharide (poly- = “many”). The chain may be branched or unbranched, and it may contain different types of monosaccharides. Polysaccharides may be very large molecules. Starch, glycogen, cellulose, and chitin are examples of polysaccharides.

Starch is the stored form of sugars in plants and is made up of amylose and amylopectin (both polymers of glucose). Plants are able to synthesize glucose, and the excess glucose is stored as starch in different plant parts, including roots and seeds. The starch that is consumed by animals is broken down into smaller molecules, such as glucose. The cells can then absorb the glucose.

Glycogen is the storage form of glucose in humans and other vertebrates, and is made up of monomers of glucose. Glycogen is the animal equivalent of starch and is a highly branched molecule usually stored in liver and muscle cells. Whenever glucose levels decrease, glycogen is broken down to release glucose.

Cellulose is one of the most abundant natural biopolymers. The cell walls of plants are mostly made of cellulose, which provides structural support to the cell. Wood and paper are mostly cellulosic in nature. Cellulose is made up of glucose monomers that are linked by bonds between particular carbon atoms in the glucose molecule.

Every other glucose monomer in cellulose is flipped over and packed tightly as extended long chains. This gives cellulose its rigidity and high tensile strength—which is so important to plant cells. Cellulose passing through our digestive system is called

dietary fiber. While the glucose-glucose bonds in cellulose cannot be broken down by human digestive enzymes, herbivores such as cows, buffalos, and horses are able to digest grass that is rich in cellulose and use it as a food source. In these animals, certain species of bacteria reside in the rumen (part of the digestive system of herbivores) and secrete the enzyme cellulase. The appendix also contains bacteria that break down cellulose, giving it an important role in the digestive systems of ruminants. Cellulases can break down cellulose into glucose monomers that can be used as an energy source by the animal.

Carbohydrates serve other functions in different animals. Arthropods, such as insects, spiders, and crabs, have an outer skeleton, called the exoskeleton, which protects their internal body parts. This exoskeleton is made of the biological macromolecule chitin, which is a nitrogenous carbohydrate. It is made of repeating units of a modified sugar containing nitrogen.

Thus, through differences in molecular structure, carbohydrates are able to serve the very different functions of energy storage (starch and glycogen) and structural support and protection (cellulose and chitin) (Figure 2.4).

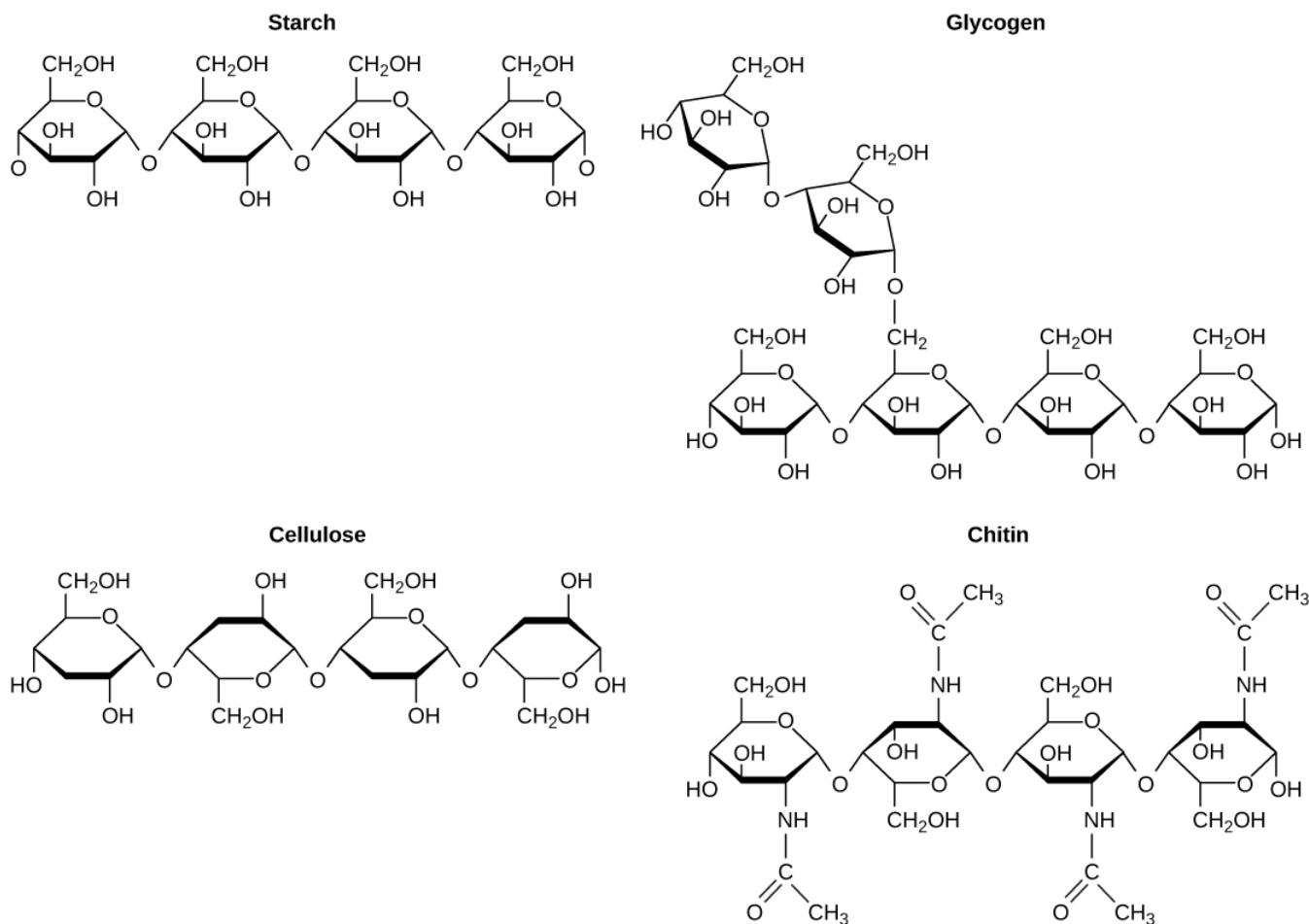


Figure 2.4: Although their structures and functions differ, all polysaccharide carbohydrates are made up of monosaccharides and have the chemical formula $(\text{CH}_2\text{O})_n$.

CAREERS IN ACTION: Registered Dietitian

Obesity is a worldwide health concern, and many diseases, such as diabetes and heart disease, are becoming more prevalent because of obesity. This is one of the reasons why registered dietitians are increasingly sought after for advice. Registered dietitians help plan food and nutrition programs for individuals in various settings. They often work with patients in health-care facilities, designing nutrition plans to prevent and treat diseases. For example, dietitians may teach a patient with diabetes how to manage blood-sugar levels by eating the correct types and amounts of carbohydrates. Dietitians may also work in nursing homes, schools, and private practices.

To become a registered dietitian, one needs to earn at least a bachelor's degree in dietetics, nutrition, food technology, or a related field. In addition, registered dietitians must complete a supervised internship program and pass a national exam. Those

who pursue careers in dietetics take courses in nutrition, chemistry, biochemistry, biology, microbiology, and human physiology. Dietitians must become experts in the chemistry and functions of food (proteins, carbohydrates, and fats).

Lipids

Lipids include a diverse group of compounds that are united by a common feature. Lipids are hydrophobic (“water-fearing”), or insoluble in water, because they are nonpolar molecules. This is because they are hydrocarbons that include only nonpolar carbon-carbon or carbon-hydrogen bonds. Lipids perform many different functions in a cell. Cells store energy for long-term use in the form of lipids called fats. Lipids also provide insulation from the environment for plants and animals (Figure 2.5). For example, they help keep aquatic birds and mammals dry because of their water-repelling nature. Lipids are also the building blocks of many hormones and are an important constituent of the plasma membrane. Lipids include fats, oils, waxes, phospholipids, and steroids.



Figure 2.5: Hydrophobic lipids in the fur of aquatic mammals, such as this river otter, protect them from the elements. (credit: Ken Bosma)

A fat molecule, such as a triglyceride, consists of two main components—glycerol and fatty acids. Glycerol is an organic compound with three carbon atoms, five hydrogen atoms, and three hydroxyl (-OH) groups. Fatty acids have a long chain of hydrocarbons to which an acidic carboxyl group is attached, hence the name “fatty acid.” The number of carbons in the fatty acid may range from 4 to 36; most common are those containing 12–18 carbons. In a fat molecule, a fatty acid is attached to each of the three oxygen atoms in the -OH groups of the glycerol molecule with a covalent bond (Figure 2.6).

Unsaturated fats or oils are usually of plant origin and contain unsaturated fatty acids. The double bond causes a bend or a “kink” that prevents the fatty acids from packing tightly, keeping them liquid at room temperature. Olive oil, corn oil, canola oil, and cod liver oil are examples of unsaturated fats. Unsaturated fats help to improve blood cholesterol levels, whereas saturated fats contribute to plaque formation in the arteries, which increases the risk of a heart attack.

In the food industry, oils are artificially hydrogenated to make them semi-solid, leading to less spoilage and increased shelf life. Simply speaking, hydrogen gas is bubbled through oils to solidify them. During this hydrogenation process, double bonds of the *cis*-conformation in the hydrocarbon chain may be converted to double bonds in the *trans*-conformation. This forms a *trans*-fat from a *cis*-fat. The orientation of the double bonds affects the chemical properties of the fat (Figure 2.7).

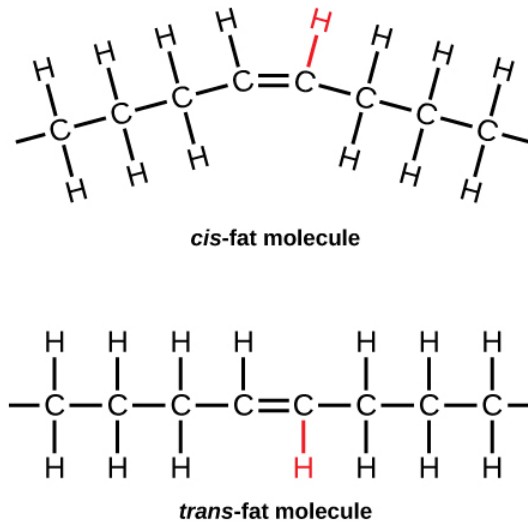


Figure 2.7: During the hydrogenation process, the orientation around the double bonds is changed, making a *trans*-fat from a *cis*-fat. This changes the chemical properties of the molecule.

Margarine, some types of peanut butter, and shortening are examples of artificially hydrogenated *trans*-fats. Recent studies have shown that an increase in *trans*-fats in the human diet may lead to an increase in levels of low-density lipoprotein (LDL), or “bad” cholesterol, which, in turn, may lead to plaque deposition in the arteries, resulting in heart disease. Many fast food restaurants have recently eliminated the use of *trans*-fats, and U.S. food labels are now required to list their *trans*-fat content.

Essential fatty acids are fatty acids that are required but not synthesized by the human body. Consequently, they must be supplemented through the diet. Omega-3 fatty acids fall into this category and are one of only two known essential fatty acids for humans (the other being omega-6 fatty acids). They are a type of polyunsaturated fat and are called omega-3 fatty acids because the third carbon from the end of the fatty acid participates in a double bond.

Salmon, trout, and tuna are good sources of omega-3 fatty acids. Omega-3 fatty acids are important in brain function and normal growth and development. They may also prevent heart disease and reduce the risk of cancer.

Like carbohydrates, fats have received a lot of bad publicity. It is true that eating an excess of fried foods and other “fatty” foods leads to weight gain. However, fats do have important functions. Fats serve as long-term energy storage. They also provide insulation for the body. Therefore, “healthy” unsaturated fats in moderate amounts should be consumed on a regular basis.

Phospholipids are the major constituent of the plasma membrane. Like fats, they are composed of fatty acid chains attached to a glycerol or similar backbone. Instead of three fatty acids attached, however, there are two fatty acids and the third carbon of the glycerol backbone is bound to a phosphate group. The phosphate group is modified by the addition of an alcohol.

A phospholipid has both hydrophobic and hydrophilic regions. The fatty acid chains are hydrophobic and exclude themselves from water, whereas the phosphate is hydrophilic and interacts with water.

Cells are surrounded by a membrane, which has a bilayer of phospholipids. The fatty acids of phospholipids face inside, away from water, whereas the phosphate group can face either the outside environment or the inside of the cell, which are both aqueous.

Steroids and Waxes

Unlike the phospholipids and fats discussed earlier, steroids have a ring structure. Although they do not resemble other lipids, they are grouped with them because they are also hydrophobic. All steroids have four, linked carbon rings and several of them, like cholesterol, have a short tail.

Cholesterol is a steroid. Cholesterol is mainly synthesized in the liver and is the precursor of many steroid hormones, such as testosterone and estradiol. It is also the precursor of vitamins E and K. Cholesterol is the precursor of bile salts, which help in the breakdown of fats and their subsequent absorption by cells. Although cholesterol is often spoken of in negative terms, it is necessary for the proper functioning of the body. It is a key component of the plasma membranes of animal cells.

Waxes are made up of a hydrocarbon chain with an alcohol ($-OH$) group and a fatty acid. Examples of animal waxes include beeswax and lanolin. Plants also have waxes, such as the coating on their leaves, that helps prevent them from drying out.

CONCEPT IN ACTION

For an additional perspective on lipids, explore “Biomolecules: The Lipids” through this interactive [animation](#).

Proteins

Proteins are one of the most abundant organic molecules in living systems and have the most diverse range of functions of all macromolecules. Proteins may be structural, regulatory, contractile, or protective; they may serve in transport, storage, or membranes; or they may be toxins or enzymes. Each cell in a living system may contain thousands of different proteins, each with a unique function. Their structures, like their functions, vary greatly. They are all, however, polymers of amino acids, arranged in a linear sequence.

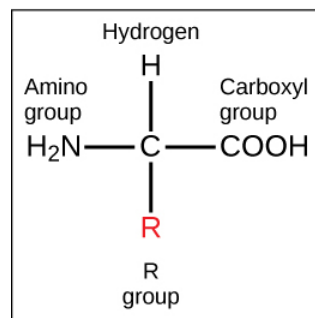
The functions of proteins are very diverse because there are 20 different chemically distinct amino acids that form long chains, and the amino acids can be in any order. For example, proteins can function as enzymes or hormones. Enzymes, which are produced by living cells, are catalysts in biochemical reactions (like digestion) and are usually proteins. Each enzyme is specific for the substrate (a reactant that binds to an enzyme) upon which it acts. Enzymes can function to break molecular bonds, to rearrange bonds, or to form new bonds. An example of an enzyme is salivary amylase, which breaks down amylose, a component of starch.

Hormones are chemical signaling molecules, usually proteins or steroids, secreted by an endocrine gland or group of endocrine cells that act to control or regulate specific physiological processes, including growth, development, metabolism, and reproduction. For example, insulin is a protein hormone that maintains blood glucose levels.

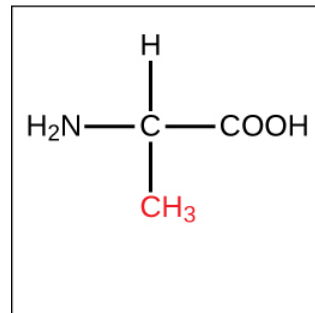
Proteins have different shapes and molecular weights; some proteins are globular in shape whereas others are fibrous in nature. For example, hemoglobin is a globular protein, but collagen, found in our skin, is a fibrous protein. Protein shape is critical to its function. Changes in temperature, pH, and exposure to chemicals may lead to permanent changes in the shape of the protein, leading to a loss of function or denaturation (to be discussed in more detail later). All proteins are made up of different arrangements of the same 20 kinds of amino acids.

Amino acids are the monomers that make up proteins. Each amino acid has the same fundamental structure, which consists of a central carbon atom bonded to an amino group ($-NH_2$), a carboxyl group ($-COOH$), and a hydrogen atom. Every amino acid also has another variable atom or group of atoms bonded to the central carbon atom known as the R group. The R group is the only difference in structure between the 20 amino acids; otherwise, the amino acids are identical (Figure 2.8).

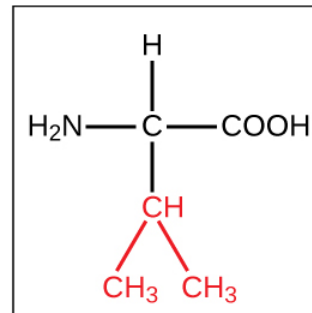
Fundamental structure



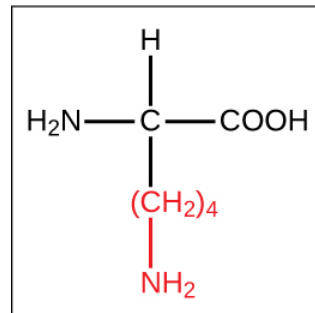
Alanine



Valine



Lysine



Aspartic acid

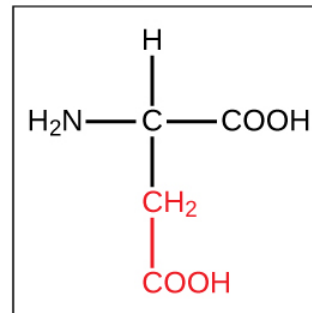


Figure 2.8: Amino acids are made up of a central carbon bonded to an amino group ($-\text{NH}_2$), a carboxyl group ($-\text{COOH}$), and a hydrogen atom. The central carbon's fourth bond varies among the different amino acids, as seen in these examples of alanine, valine, lysine, and aspartic acid.

The chemical nature of the R group determines the chemical nature of the amino acid within its protein (that is, whether it is acidic, basic, polar, or nonpolar).

The sequence and number of amino acids ultimately determine a protein's shape, size, and function. Each amino acid is attached to another amino acid by a covalent bond, known as a peptide bond, which is formed by a dehydration reaction. The carboxyl group of one amino acid and the amino group of a second amino acid combine, releasing a water molecule. The resulting bond is the peptide bond.

The products formed by such a linkage are called polypeptides. While the terms polypeptide and protein are sometimes used interchangeably, a polypeptide is technically a polymer of amino acids, whereas the term protein is used for a polypeptide or polypeptides that have combined together, have a distinct shape, and have a unique function.

EVOLUTION IN ACTION: The Evolutionary Significance of Cytochrome c

Cytochrome c is an important component of the molecular machinery that harvests energy from glucose. Because this protein's role in producing cellular energy is crucial, it has changed very little over millions of years. Protein sequencing has shown that there is a considerable amount of sequence similarity among cytochrome c molecules of different species; evolutionary relationships can be assessed by measuring the similarities or differences among various species' protein sequences.

For example, scientists have determined that human cytochrome c contains 104 amino acids. For each cytochrome c molecule that has been sequenced to date from different organisms, 37 of these amino acids appear in the same position in each cytochrome c. This indicates that all of these organisms are descended from a common ancestor. On comparing the human and chimpanzee protein sequences, no sequence difference was found. When human and rhesus monkey sequences were compared, a single difference was found in one amino acid. In contrast, human-to-yeast comparisons show a difference in 44 amino acids, suggesting that humans and chimpanzees have a more recent common ancestor than humans and the rhesus monkey, or humans and yeast.

Protein Structure

As discussed earlier, the shape of a protein is critical to its function. To understand how the protein gets its final shape or conformation, we need to understand the four levels of protein structure: primary, secondary, tertiary, and quaternary (Figure 2.9).

The unique sequence and number of amino acids in a polypeptide chain is its primary structure. The unique sequence for every protein is ultimately determined by the gene that encodes the protein. Any change in the gene sequence may lead to a different amino acid being added to the polypeptide chain, causing a change in protein structure and function. In sickle cell anemia, the hemoglobin β chain has a single amino acid substitution, causing a change in both the structure and function of the protein. What is most remarkable to consider is that a hemoglobin molecule is made up of two alpha chains and two beta chains that each consist of about 150 amino acids. The molecule, therefore, has about 600 amino acids. The structural difference between a normal hemoglobin molecule and a sickle cell molecule—that dramatically decreases life expectancy in the affected individuals—is a single amino acid of the 600.

Because of this change of one amino acid in the chain, the normally biconcave, or disc-shaped, red blood cells assume a crescent or “sickle” shape, which clogs arteries. This can lead to a myriad of serious health problems, such as breathlessness, dizziness, headaches, and abdominal pain for those who have this disease.

Folding patterns resulting from interactions between the non-R group portions of amino acids give rise to the secondary structure of the protein. The most common are the alpha (α)-helix and beta (β)-pleated sheet structures. Both structures are held in shape by hydrogen bonds. In the alpha helix, the bonds form between every fourth amino acid and cause a twist in the amino acid chain.

In the β -pleated sheet, the “pleats” are formed by hydrogen bonding between atoms on the backbone of the polypeptide chain. The R groups are attached to the carbons, and extend above and below the folds of the pleat. The pleated segments align parallel to each other, and hydrogen bonds form between the same pairs of atoms on each of the aligned amino acids. The α -helix and β -pleated sheet structures are found in many globular and fibrous proteins.

The unique three-dimensional structure of a polypeptide is known as its tertiary structure. This structure is caused by chemical interactions between various amino acids and regions of the polypeptide. Primarily, the interactions among R groups create the complex three-dimensional tertiary structure of a protein. There may be ionic bonds formed between R groups on different amino acids, or hydrogen bonding beyond that involved in the secondary structure. When protein folding takes place, the hydrophobic R groups of nonpolar amino acids lay in the interior of the protein, whereas the hydrophilic R groups lay on the outside. The former types of interactions are also known as hydrophobic interactions.

In nature, some proteins are formed from several polypeptides, also known as subunits, and the interaction of these subunits forms the quaternary structure. Weak interactions between the subunits help to stabilize the overall structure. For example, hemoglobin is a combination of four polypeptide subunits.

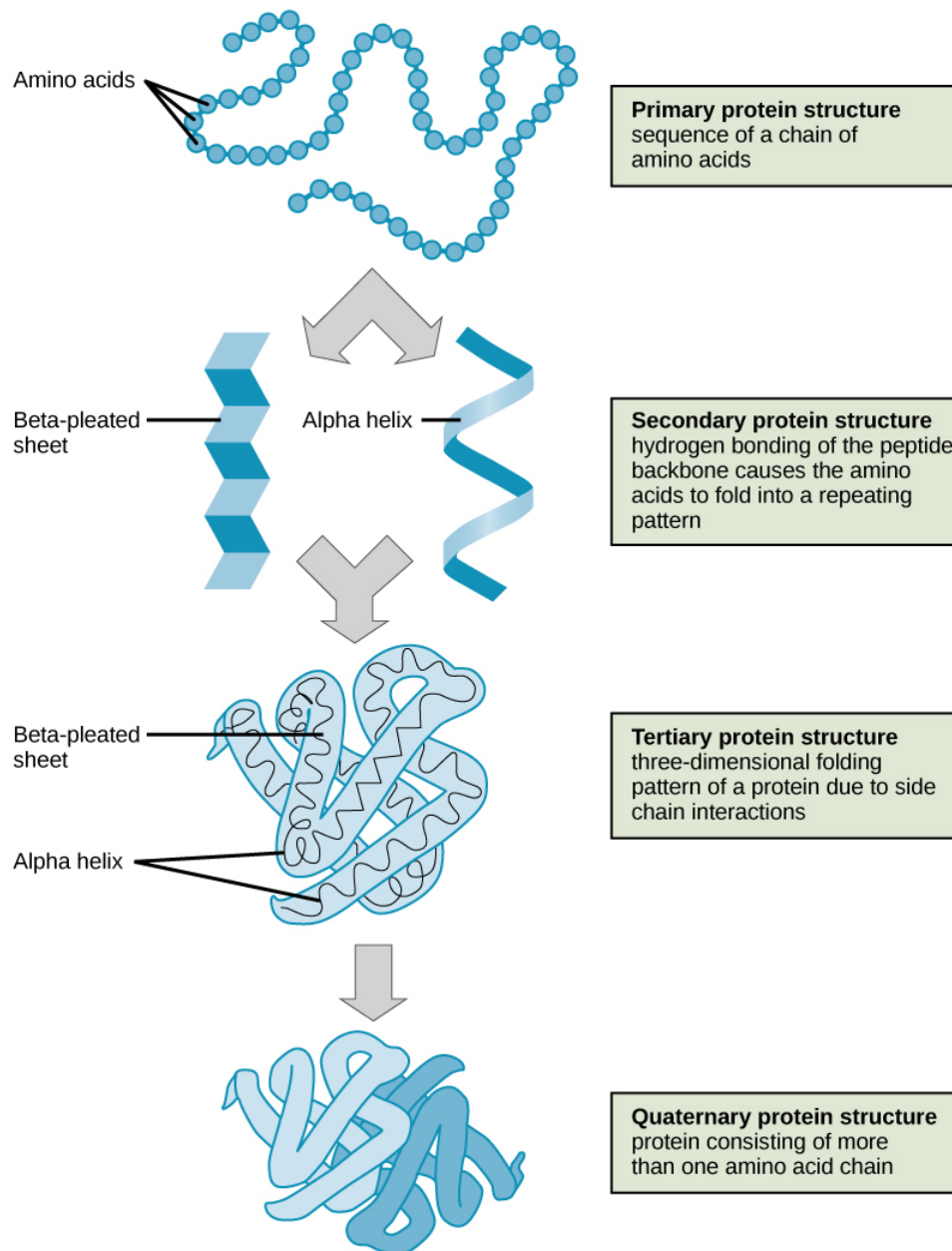


Figure 2.9: The four levels of protein structure can be observed in these illustrations. (credit: modification of work by National Human Genome Research Institute)

Each protein has its own unique sequence and shape held together by chemical interactions. If the protein is subject to changes in temperature, pH, or exposure to chemicals, the protein structure may change, losing its shape in what is known as denaturation as discussed earlier. Denaturation is often reversible because the primary structure is preserved if the denaturing agent is removed, allowing the protein to resume its function. Sometimes denaturation is irreversible, leading to a loss of function. One example of protein denaturation can be seen when an egg is fried or boiled. The albumin protein in the liquid egg white is denatured when placed in a hot pan, changing from a clear substance to an opaque white substance. Not all proteins are denatured at high temperatures; for instance, bacteria that survive in hot springs have proteins that are adapted to function at those temperatures.

CONCEPT IN ACTION

For an additional perspective on proteins, explore “Biomolecules: The Proteins” through this interactive [animation](https://bio.libretexts.org/@go/page/155979).

Nucleic Acids

Nucleic acids are key macromolecules in the continuity of life. They carry the genetic blueprint of a cell and carry instructions for the functioning of the cell.

The two main types of nucleic acids are deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). DNA is the genetic material found in all living organisms, ranging from single-celled bacteria to multicellular mammals.

The other type of nucleic acid, RNA, is mostly involved in protein synthesis. The DNA molecules never leave the nucleus, but instead use an RNA intermediary to communicate with the rest of the cell. Other types of RNA are also involved in protein synthesis and its regulation.

DNA and RNA are made up of monomers known as nucleotides. The nucleotides combine with each other to form a polynucleotide, DNA or RNA. Each nucleotide is made up of three components: a nitrogenous base, a pentose (five-carbon) sugar, and a phosphate group (Figure 2.10). Each nitrogenous base in a nucleotide is attached to a sugar molecule, which is attached to a phosphate group.

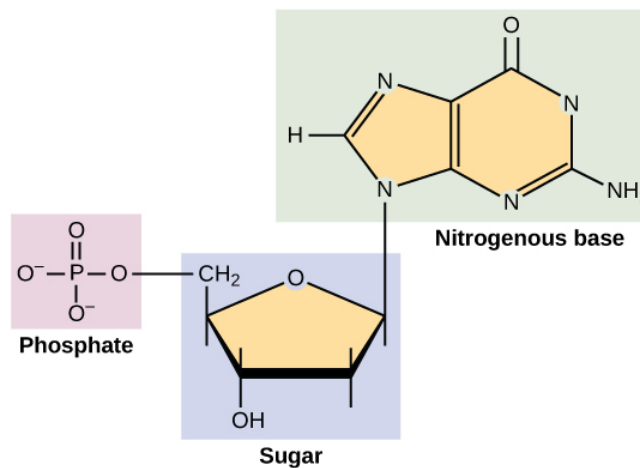


Figure 2.10: A nucleotide is made up of three components: a nitrogenous base, a pentose sugar, and a phosphate group.

DNA Double-Helical Structure

DNA has a double-helical structure (Figure 2.11). It is composed of two strands, or polymers, of nucleotides. The strands are formed with bonds between phosphate and sugar groups of adjacent nucleotides. The strands are bonded to each other at their bases with hydrogen bonds, and the strands coil about each other along their length, hence the “double helix” description, which means a double spiral.

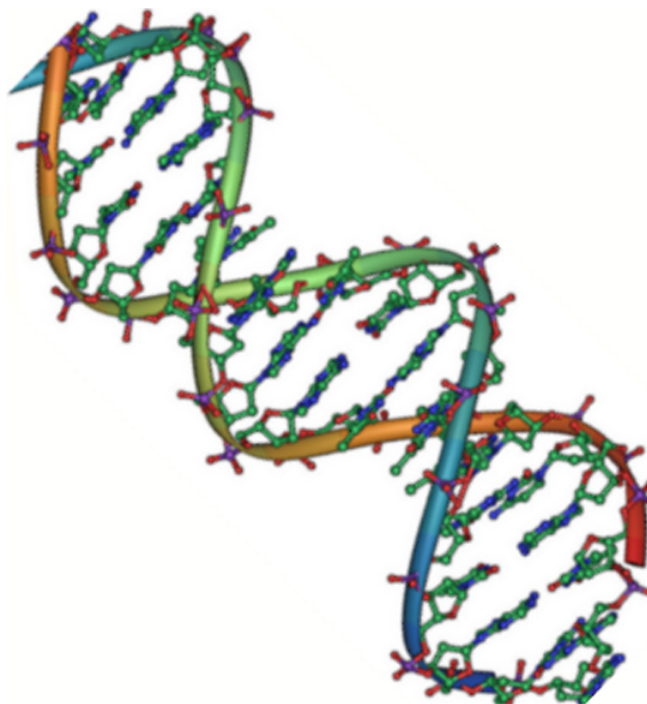


Figure 2.11: The double-helix model shows DNA as two parallel strands of intertwining molecules. (credit: Jerome Walker, Dennis Myts)

The alternating sugar and phosphate groups lie on the outside of each strand, forming the backbone of the DNA. The nitrogenous bases are stacked in the interior, like the steps of a staircase, and these bases pair; the pairs are bound to each other by hydrogen bonds. The bases pair in such a way that the distance between the backbones of the two strands is the same all along the molecule.

Summary

Living things are carbon-based because carbon plays such a prominent role in the chemistry of living things. The four covalent bonding positions of the carbon atom can give rise to a wide diversity of compounds with many functions, accounting for the importance of carbon in living things. Carbohydrates are a group of macromolecules that are a vital energy source for the cell, provide structural support to many organisms, and can be found on the surface of the cell as receptors or for cell recognition. Carbohydrates are classified as monosaccharides, disaccharides, and polysaccharides, depending on the number of monomers in the molecule.

Lipids are a class of macromolecules that are nonpolar and hydrophobic in nature. Major types include fats and oils, waxes, phospholipids, and steroids. Fats and oils are a stored form of energy and can include triglycerides. Fats and oils are usually made up of fatty acids and glycerol.

Proteins are a class of macromolecules that can perform a diverse range of functions for the cell. They help in metabolism by providing structural support and by acting as enzymes, carriers or as hormones. The building blocks of proteins are amino acids. Proteins are organized at four levels: primary, secondary, tertiary, and quaternary. Protein shape and function are intricately linked; any change in shape caused by changes in temperature, pH, or chemical exposure may lead to protein denaturation and a loss of function.

Nucleic acids are molecules made up of repeating units of nucleotides that direct cellular activities such as cell division and protein synthesis. Each nucleotide is made up of a pentose sugar, a nitrogenous base, and a phosphate group. There are two types of nucleic acids: DNA and RNA.

Glossary

amino acid

a monomer of a protein

carbohydrate

a biological macromolecule in which the ratio of carbon to hydrogen to oxygen is 1:2:1; carbohydrates serve as energy sources and structural support in cells

cellulose

a polysaccharide that makes up the cell walls of plants and provides structural support to the cell

chitin

a type of carbohydrate that forms the outer skeleton of arthropods, such as insects and crustaceans, and the cell walls of fungi

denaturation

the loss of shape in a protein as a result of changes in temperature, pH, or exposure to chemicals

deoxyribonucleic acid (DNA)

a double-stranded polymer of nucleotides that carries the hereditary information of the cell

disaccharide

two sugar monomers that are linked together by a peptide bond

enzyme

a catalyst in a biochemical reaction that is usually a complex or conjugated protein

fat

a lipid molecule composed of three fatty acids and a glycerol (triglyceride) that typically exists in a solid form at room temperature

glycogen

a storage carbohydrate in animals

hormone

a chemical signaling molecule, usually a protein or steroid, secreted by an endocrine gland or group of endocrine cells; acts to control or regulate specific physiological processes

lipids

a class of macromolecules that are nonpolar and insoluble in water

macromolecule

a large molecule, often formed by polymerization of smaller monomers

monosaccharide

a single unit or monomer of carbohydrates

nucleic acid

a biological macromolecule that carries the genetic information of a cell and carries instructions for the functioning of the cell

nucleotide

a monomer of nucleic acids; contains a pentose sugar, a phosphate group, and a nitrogenous base

oil

an unsaturated fat that is a liquid at room temperature

phospholipid

a major constituent of the membranes of cells; composed of two fatty acids and a phosphate group attached to the glycerol backbone

polypeptide

a long chain of amino acids linked by peptide bonds

polysaccharide

a long chain of monosaccharides; may be branched or unbranched

protein

a biological macromolecule composed of one or more chains of amino acids

ribonucleic acid (RNA)

a single-stranded polymer of nucleotides that is involved in protein synthesis

saturated fatty acid

a long-chain hydrocarbon with single covalent bonds in the carbon chain; the number of hydrogen atoms attached to the carbon skeleton is maximized

starch

a storage carbohydrate in plants

steroid

a type of lipid composed of four fused hydrocarbon rings

***trans*-fat**

a form of unsaturated fat with the hydrogen atoms neighboring the double bond across from each other rather than on the same side of the double bond

triglyceride

a fat molecule; consists of three fatty acids linked to a glycerol molecule

unsaturated fatty acid

a long-chain hydrocarbon that has one or more than one double bonds in the hydrocarbon chain

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3: The Structure of DNA

In the 1950s, Francis Crick and James Watson worked together at the University of Cambridge, England, to determine the structure of DNA. Other scientists, such as Linus Pauling and Maurice Wilkins, were also actively exploring this field. Pauling had discovered the secondary structure of proteins using X-ray crystallography. X-ray crystallography is a method for investigating molecular structure by observing the patterns formed by X-rays shot through a crystal of the substance. The patterns give important information about the structure of the molecule of interest. In Wilkins' lab, researcher Rosalind Franklin was using X-ray crystallography to understand the structure of DNA. Watson and Crick were able to piece together the puzzle of the DNA molecule using Franklin's data (Figure 3.1). Watson and Crick also had key pieces of information available from other researchers such as Chargaff's rules. Chargaff had shown that of the four kinds of monomers (nucleotides) present in a DNA molecule, two types were always present in equal amounts and the remaining two types were also always present in equal amounts. This meant they were always paired in some way. In 1962, James Watson, Francis Crick, and Maurice Wilkins were awarded the Nobel Prize in Medicine for their work in determining the structure of DNA.

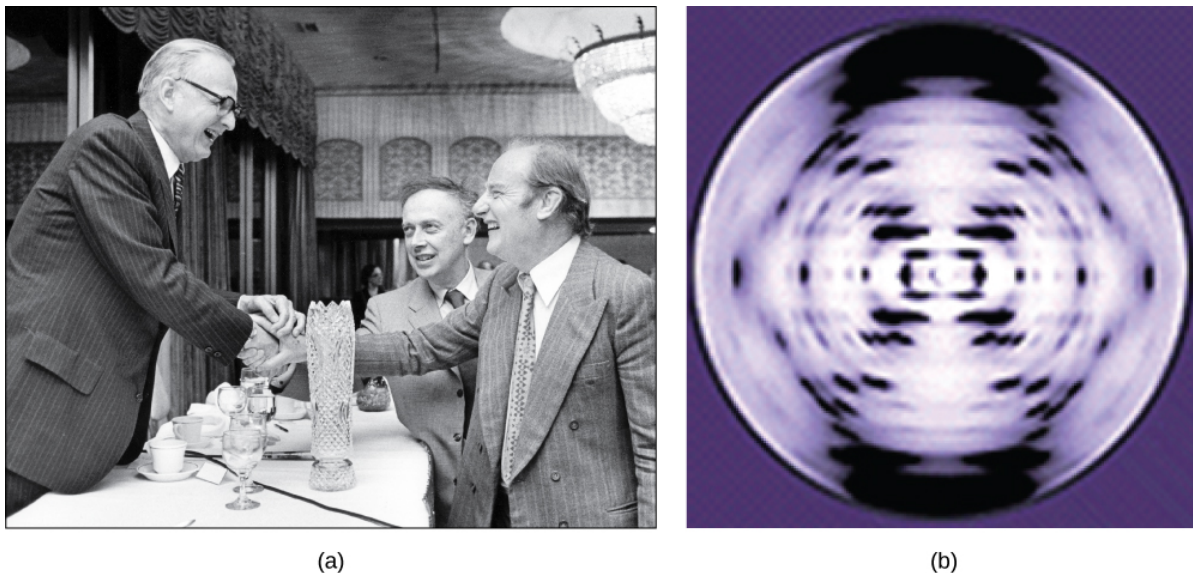


Figure 3.1: Pioneering scientists (a) James Watson and Francis Crick are pictured here with American geneticist Maclyn McCarty. Scientist Rosalind Franklin discovered (b) the X-ray diffraction pattern of DNA, which helped to elucidate its double helix structure. (credit a: modification of work by Marjorie McCarty; b: modification of work by NIH)

Now let's consider the structure of the two types of nucleic acids, deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). The building blocks of DNA are nucleotides, which are made up of three parts: a deoxyribose (5-carbon sugar), a phosphate group, and a nitrogenous base (Figure 3.2). There are four types of nitrogenous bases in DNA. Adenine (A) and guanine (G) are double-ringed purines, and cytosine (C) and thymine (T) are smaller, single-ringed pyrimidines. The nucleotide is named according to the nitrogenous base it contains.

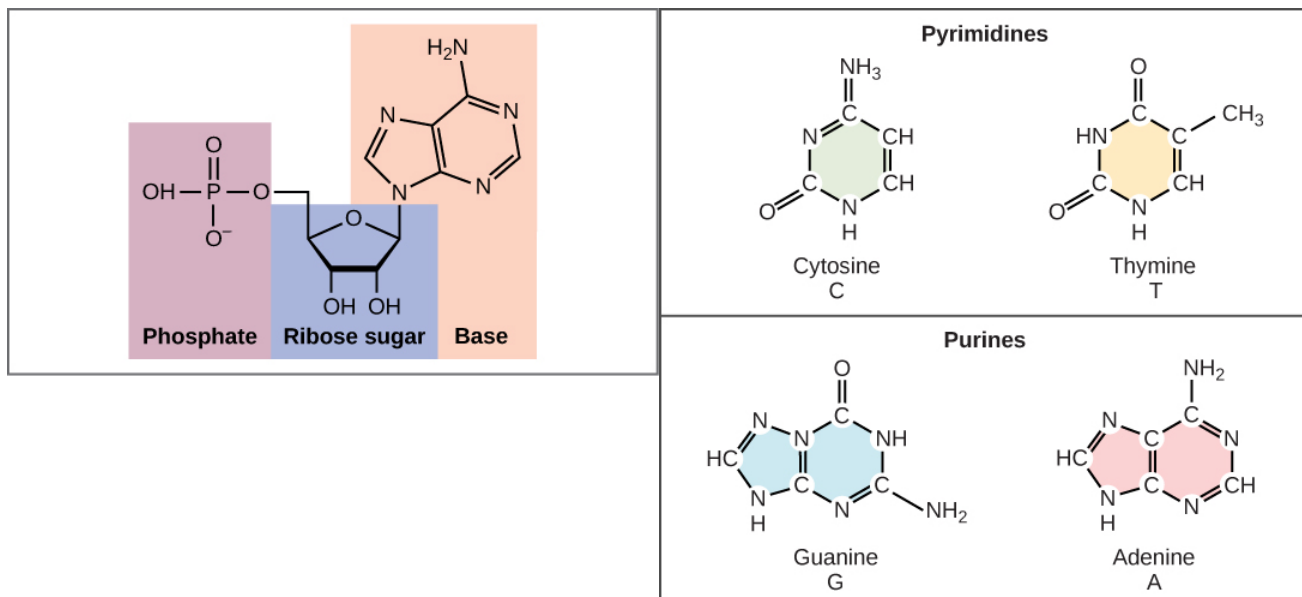


Figure 3.2: (a) Each DNA nucleotide is made up of a sugar, a phosphate group, and a base. (b) Cytosine and thymine are pyrimidines. Guanine and adenine are purines.

The phosphate group of one nucleotide bonds covalently with the sugar molecule of the next nucleotide, and so on, forming a long polymer of nucleotide monomers. The sugar-phosphate groups line up in a “backbone” for each single strand of DNA, and the nucleotide bases stick out from this backbone. The carbon atoms of the five-carbon sugar are numbered clockwise from the oxygen as 1', 2', 3', 4', and 5' (1' is read as “one prime”). The phosphate group is attached to the 5' carbon of one nucleotide and the 3' carbon of the next nucleotide. In its natural state, each DNA molecule is actually composed of two single strands held together along their length with hydrogen bonds between the bases.

Watson and Crick proposed that the DNA is made up of two strands that are twisted around each other to form a right-handed helix, called a double helix. Base-pairing takes place between a purine and pyrimidine: namely, A pairs with T, and G pairs with C. In other words, adenine and thymine are complementary base pairs, and cytosine and guanine are also complementary base pairs. This is the basis for Chargaff’s rule; because of their complementarity, there is as much adenine as thymine in a DNA molecule and as much guanine as cytosine. Adenine and thymine are connected by two hydrogen bonds, and cytosine and guanine are connected by three hydrogen bonds. The two strands are anti-parallel in nature; that is, one strand will have the 3' carbon of the sugar in the “upward” position, whereas the other strand will have the 5' carbon in the upward position. The diameter of the DNA double helix is uniform throughout because a purine (two rings) always pairs with a pyrimidine (one ring) and their combined lengths are always equal (Figure 3.3).

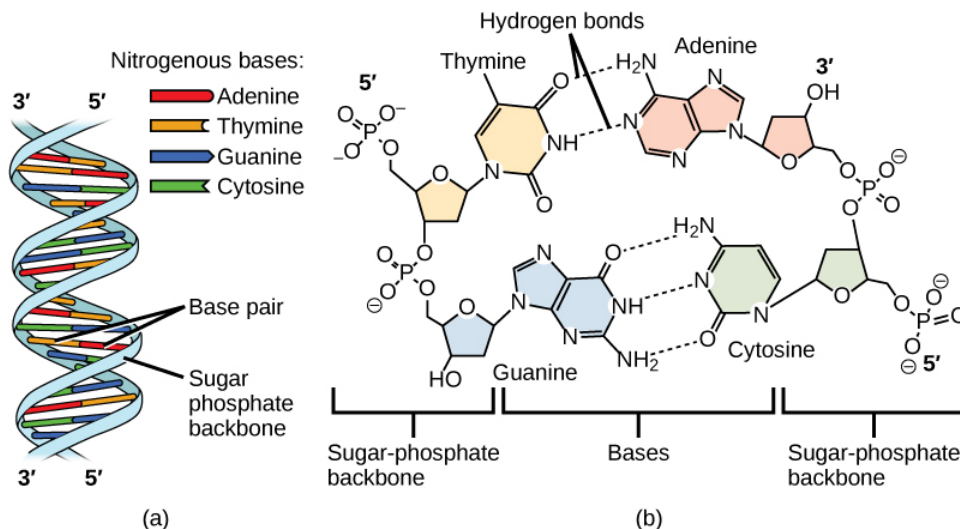


Figure 3.3: DNA (a) forms a double stranded helix, and (b) adenine pairs with thymine and cytosine pairs with guanine. (credit a: modification of work by Jerome Walker, Dennis Myts)

The Structure of RNA

There is a second nucleic acid in all cells called ribonucleic acid, or RNA. Like DNA, RNA is a polymer of nucleotides. Each of the nucleotides in RNA is made up of a nitrogenous base, a five-carbon sugar, and a phosphate group. In the case of RNA, the five-carbon sugar is ribose, not deoxyribose. Ribose has a hydroxyl group at the 2' carbon, unlike deoxyribose, which has only a hydrogen atom (Figure 3.4).

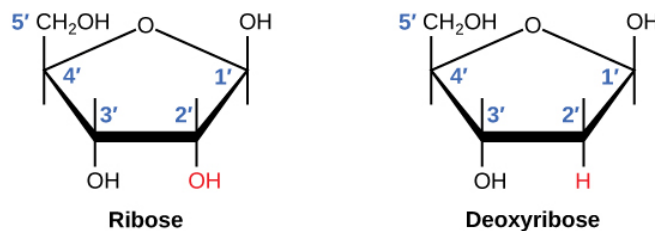


Figure 3.4: The difference between the ribose found in RNA and the deoxyribose found in DNA is that ribose has a hydroxyl group at the 2' carbon.

RNA nucleotides contain the nitrogenous bases adenine, cytosine, and guanine. However, they do not contain thymine, which is instead replaced by uracil, symbolized by a “U.” RNA exists as a single-stranded molecule rather than a double-stranded helix. Molecular biologists have named several kinds of RNA on the basis of their function. These include messenger RNA (mRNA), transfer RNA (tRNA), and ribosomal RNA (rRNA)—molecules that are involved in the production of proteins from the DNA code.

How DNA Is Arranged in the Cell

DNA is a working molecule; it must be replicated when a cell is ready to divide, and it must be “read” to produce the molecules, such as proteins, to carry out the functions of the cell. For this reason, the DNA is protected and packaged in very specific ways. In addition, DNA molecules can be very long. Stretched end-to-end, the DNA molecules in a single human cell would come to a length of about 2 meters. Thus, the DNA for a cell must be packaged in a very ordered way to fit and function within a structure (the cell) that is not visible to the naked eye. The chromosomes of prokaryotes are much simpler than those of eukaryotes in many of their features (Figure 3.5). Most prokaryotes contain a single, circular chromosome that is found in an area in the cytoplasm called the nucleoid.

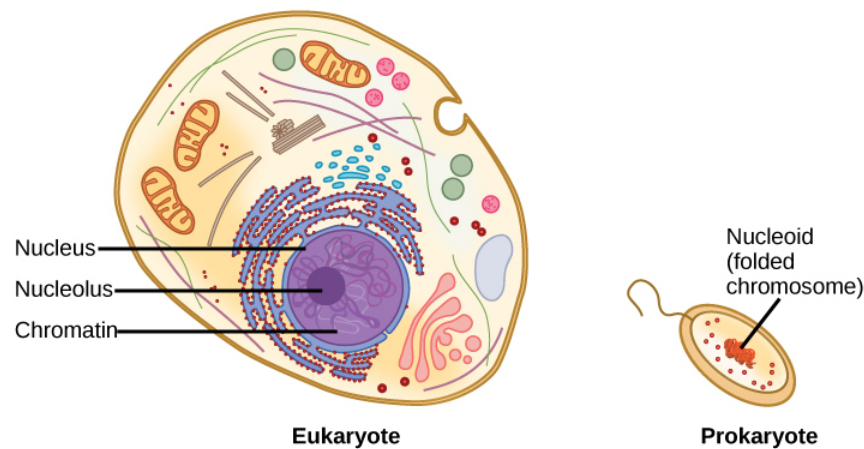


Figure 3.5: A eukaryote contains a well-defined nucleus, whereas in prokaryotes, the chromosome lies in the cytoplasm in an area called the nucleoid.

The size of the genome in one of the most well-studied prokaryotes, *Escherichia coli*, is 4.6 million base pairs, which would extend a distance of about 1.6 mm if stretched out. So how does this fit inside a small bacterial cell? The DNA is twisted beyond the double helix in what is known as supercoiling. Some proteins are known to be involved in the supercoiling; other proteins and enzymes help in maintaining the supercoiled structure.

Eukaryotes, whose chromosomes each consist of a linear DNA molecule, employ a different type of packing strategy to fit their DNA inside the nucleus (Figure 3.6). At the most basic level, DNA is wrapped around proteins known as histones to form structures called nucleosomes. The DNA is wrapped tightly around the histone core. This nucleosome is linked to the next one by a short strand of DNA that is free of histones. This is also known as the “beads on a string” structure; the nucleosomes are the “beads” and the short lengths of DNA between them are the “string.” The nucleosomes, with their DNA coiled around them, stack compactly onto each other to form a 30-nm-wide fiber. This fiber is further coiled into a thicker and more compact structure. At the metaphase stage of mitosis, when the chromosomes are lined up in the center of the cell, the chromosomes are at their most compacted. They are approximately 700 nm in width, and are found in association with scaffold proteins.

In interphase, the phase of the cell cycle between mitoses at which the chromosomes are decondensed, eukaryotic chromosomes have two distinct regions that can be distinguished by staining. There is a tightly packaged region that stains darkly, and a less dense region. The darkly staining regions usually contain genes that are not active, and are found in the regions of the centromere and telomeres. The lightly staining regions usually contain genes that are active, with DNA packaged around nucleosomes but not further compacted.

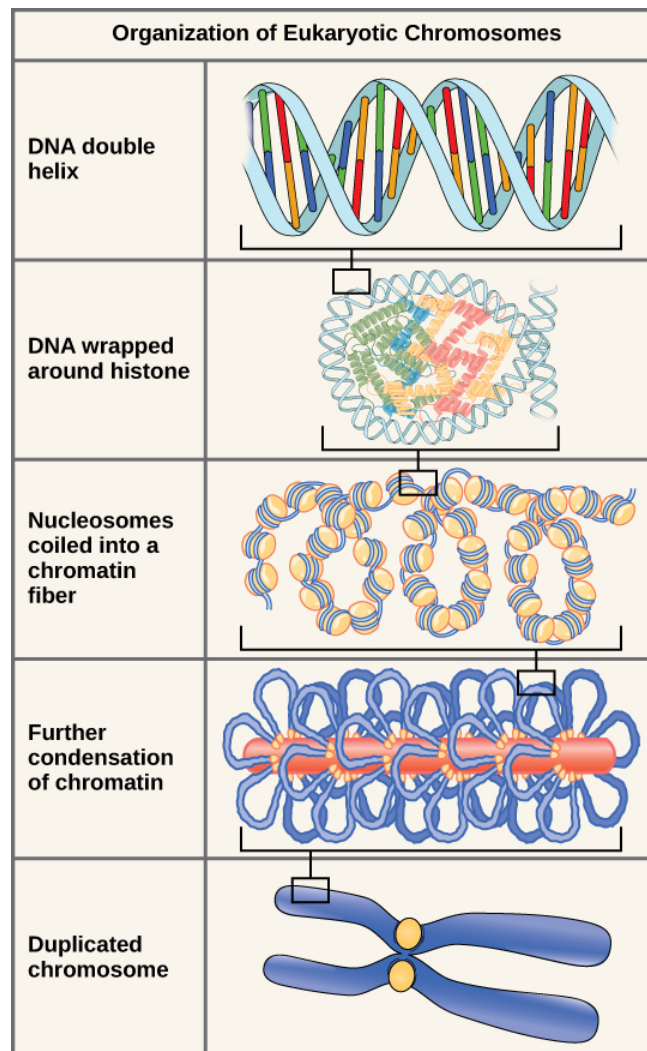


Figure 3.6: These figures illustrate the compaction of the eukaryotic chromosome.

CONCEPT IN ACTION



How DNA is Packaged (Advanced)

A Learning Center



www.dnalc.org Watch on  **YouTube**

Watch this [animation](#) of DNA packaging.

Summary

The model of the double-helix structure of DNA was proposed by Watson and Crick. The DNA molecule is a polymer of nucleotides. Each nucleotide is composed of a nitrogenous base, a five-carbon sugar (deoxyribose), and a phosphate group. There

are four nitrogenous bases in DNA, two purines (adenine and guanine) and two pyrimidines (cytosine and thymine). A DNA molecule is composed of two strands. Each strand is composed of nucleotides bonded together covalently between the phosphate group of one and the deoxyribose sugar of the next. From this backbone extend the bases. The bases of one strand bond to the bases of the second strand with hydrogen bonds. Adenine always bonds with thymine, and cytosine always bonds with guanine. The bonding causes the two strands to spiral around each other in a shape called a double helix. Ribonucleic acid (RNA) is a second nucleic acid found in cells. RNA is a single-stranded polymer of nucleotides. It also differs from DNA in that it contains the sugar ribose, rather than deoxyribose, and the nucleotide uracil rather than thymine. Various RNA molecules function in the process of forming proteins from the genetic code in DNA.

Prokaryotes contain a single, double-stranded circular chromosome. Eukaryotes contain double-stranded linear DNA molecules packaged into chromosomes. The DNA helix is wrapped around proteins to form nucleosomes. The protein coils are further coiled, and during mitosis and meiosis, the chromosomes become even more greatly coiled to facilitate their movement. Chromosomes have two distinct regions which can be distinguished by staining, reflecting different degrees of packaging and determined by whether the DNA in a region is being expressed (euchromatin) or not (heterochromatin).

Glossary

deoxyribose

a five-carbon sugar molecule with a hydrogen atom rather than a hydroxyl group in the 2' position; the sugar component of DNA nucleotides

double helix

the molecular shape of DNA in which two strands of nucleotides wind around each other in a spiral shape

nitrogenous base

a nitrogen-containing molecule that acts as a base; often referring to one of the purine or pyrimidine components of nucleic acids

phosphate group

a molecular group consisting of a central phosphorus atom bound to four oxygen atoms

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4: The Genome

The continuity of life from one cell to another has its foundation in the reproduction of cells by way of the cell cycle. The cell cycle is an orderly sequence of events in the life of a cell from the division of a single parent cell to produce two new daughter cells, to the subsequent division of those daughter cells. The mechanisms involved in the cell cycle are highly conserved across eukaryotes. Organisms as diverse as protists, plants, and animals employ similar steps.

Genomic DNA

Before discussing the steps a cell undertakes to replicate, a deeper understanding of the structure and function of a cell's genetic information is necessary. A cell's complete complement of DNA is called its genome. In prokaryotes, the genome is composed of a single, double-stranded DNA molecule in the form of a loop or circle. The region in the cell containing this genetic material is called a nucleoid. Some prokaryotes also have smaller loops of DNA called plasmids that are not essential for normal growth.

In eukaryotes, the genome comprises several double-stranded, linear DNA molecules (Figure 4.1) bound with proteins to form complexes called chromosomes. Each species of eukaryote has a characteristic number of chromosomes in the nuclei of its cells. Human body cells (somatic cells) have 46 chromosomes. A somatic cell contains two matched sets of chromosomes, a configuration known as diploid. The letter n is used to represent a single set of chromosomes; therefore a diploid organism is designated $2n$. Human cells that contain one set of 23 chromosomes are called gametes, or sex cells; these eggs and sperm are designated n , or haploid.

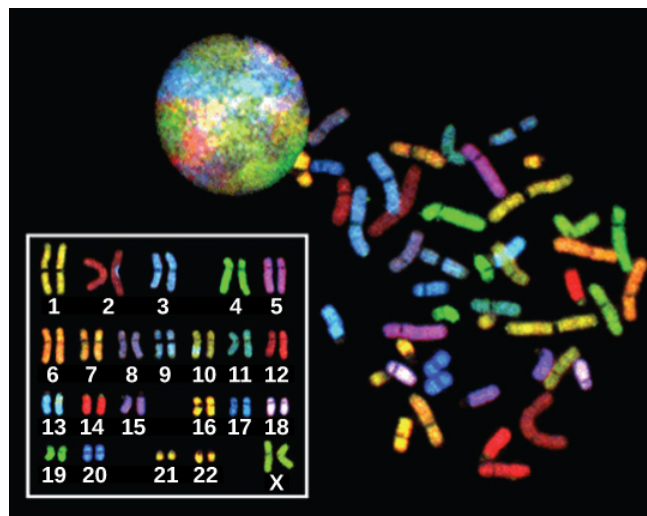


Figure 4.1: There are 23 pairs of homologous chromosomes in a female human somatic cell. These chromosomes are viewed within the nucleus (top), removed from a cell in mitosis (right), and arranged according to length (left) in an arrangement called a karyotype. In this image, the chromosomes were exposed to fluorescent stains to distinguish them. (credit: "718 Bot"/Wikimedia Commons, National Human Genome Research)

The matched pairs of chromosomes in a diploid organism are called homologous chromosomes. Homologous chromosomes are the same length and have specific nucleotide segments called genes in exactly the same location, or locus. Genes, the functional units of chromosomes, determine specific characteristics by coding for specific proteins. Traits are the different forms of a characteristic. For example, the shape of earlobes is a characteristic with traits of free or attached.

Each copy of the homologous pair of chromosomes originates from a different parent; therefore, the copies of each of the genes themselves may not be identical. The variation of individuals within a species is caused by the specific combination of the genes inherited from both parents. For example, there are three possible gene sequences on the human chromosome that codes for blood type: sequence A, sequence B, and sequence O. Because all diploid human cells have two copies of the chromosome that determines blood type, the blood type (the trait) is determined by which two versions of the marker gene are inherited. It is possible to have two copies of the same gene sequence, one on each homologous chromosome (for example, AA, BB, or OO), or two different sequences, such as AB.

Minor variations in traits such as those for blood type, eye color, and height contribute to the natural variation found within a species. The sex chromosomes, X and Y, are the single exception to the rule of homologous chromosomes; other than a small amount of homology that is necessary to reliably produce gametes, the genes found on the X and Y chromosomes are not the same.

Summary

Prokaryotes have a single loop chromosome, whereas eukaryotes have multiple, linear chromosomes surrounded by a nuclear membrane. Human somatic cells have 46 chromosomes consisting of two sets of 22 homologous chromosomes and a pair of nonhomologous sex chromosomes. This is the $2n$, or diploid, state. Human gametes have 23 chromosomes or one complete set of chromosomes. This is the n , or haploid, state. Genes are segments of DNA that code for a specific protein or RNA molecule. An organism's traits are determined in large part by the genes inherited from each parent, but also by the environment that they experience. Genes are expressed as characteristics of the organism and each characteristic may have different variants called traits that are caused by differences in the DNA sequence for a gene.

Glossary

diploid

describes a cell, nucleus, or organism containing two sets of chromosomes ($2n$)

gamete

a haploid reproductive cell or sex cell (sperm or egg)

gene

the physical and functional unit of heredity; a sequence of DNA that codes for a specific peptide or RNA molecule

genome

the entire genetic complement (DNA) of an organism

haploid

describes a cell, nucleus, or organism containing one set of chromosomes (n)

homologous chromosomes

chromosomes of the same length with genes in the same location; diploid organisms have pairs of homologous chromosomes, and the members of each pair come from different parents

locus

the position of a gene on a chromosome

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CHAPTER OVERVIEW

5: Mendelian Genetics

5.1: A History of Genetics Research

5.1.1: Mendel's Experiments

5.2: Laws of Inheritance

5.3: Mendelian Genetics

5.3.1: Monohybrid Crosses and Segregation

5.3.2: Dihybrid Crosses and Independent Assortment

5.4: Model Organisms Facilitate Genetic Advances

Thumbnail: A single white butterfly pea flower was blooming alone on its branches. (Unsplash License; [Worrakarn Boonyasngawongs](#) via [Unsplash](#))

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5.1: A History of Genetics Research

Learning Objectives

- Discuss Gregor Mendel's discovery of the Principles of Heredity and comment on why he was successful.
- Establish and analyze monohybrid crosses.
- Describe how monohybrid crosses reveal the Principle of Segregation and the Concept of Dominance.
- Apply a test cross to identify genotypes in individuals expressing one dominant trait.
- Apply probability to predict genotypic and phenotypic ratios in monohybrid crosses.

Genes

Mendel first made his discoveries of inheritance in the 1850s. In his 1866 publication, *Experiments on Plant Hybridization*, he didn't use the word "gene" as the fundamental unit of heredity because it wasn't coined until 1909 by Danish botanist [Wilhelm Johannsen](#). [Thomas Hunt Morgan](#) proposed that genes resided on chromosomes in 1910, and occupied distinct regions on those chromosomes. DNA as a substance was discovered in the 1860s, but it took until the 1940s to realize that DNA was the molecule that contained the genetic information. Then in the 1950s Watson and Crick discovered the structure of DNA. Take a look at the video, "A History of Research on Genetics" by Sigma Documentaries on YouTube, which summarizes some of these landmark studies.

Watch this video, *History of Research on Genetics*, by Sigma Documentaries (2017) on YouTube.



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Key Milestones in Genetics and Molecular Biology

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Note: If you are not using the online version of this guide, you can find the timeline in Appendix A:Key Milestones in Genetics and Molecular Biology Timeline

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Long Descriptions

- **Figure 1.1.1** Headshot of Gregor Mendel, the Augustinian monk, pictured with a receding hairline, wearing a black robe, spectacles and a cross around his neck. [Back to [Figure 1.1.1](#)]
- **Figure 1.1.2** Garden peas appear with many green leaves, tall green stems, green pea pods, and a few white flowers. [Back to [Figure 1.1.2](#)]
- **Figure 1.1.3** A medium bristled paint brush with yellow pollen dust on the tip of the brush is used for hand-pollination. [Back to [Figure 1.1.3](#)]
- **Figure 1.1.4** The seven traits in garden peas Mendel experimented with are: 1) round and wrinkled seeds; 2) yellow and green seeds; 3) white and purple flowers; 4) full and constricted pods; 5) yellow and green pods; 6) axial and terminal stems; and 7) long and short stems. [Back to [Figure 1.1.4](#)]
- **Figure 1.1.5** A Punnett square illustrates the segregation of alleles in male and female parents in a monohybrid cross, to produce four offspring in a 3:1 phenotypic ratio. A purple flowered male is crossed with a purple flowered female. Both parents are heterozygous (Bb). Offspring are 1 homozygous dominant (BB), 2 heterozygous (Bb) and 1 homozygous recessive (bb). [Back to [Figure 1.1.5](#)]

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5.1.1: Mendel's Experiments

Johann Gregor Mendel (1822–1884) (Figure 5.1.1.1) was a lifelong learner, teacher, scientist, and man of faith. As a young adult, he joined the Augustinian Abbey of St. Thomas in Brno in what is now the Czech Republic. Supported by the monastery, he taught physics, botany, and natural science courses at the secondary and university levels. In 1856, he began a decade-long research pursuit involving inheritance patterns in honeybees and plants, ultimately settling on pea plants as his primary model system (a system with convenient characteristics that is used to study a specific biological phenomenon to gain understanding to be applied to other systems). In 1865, Mendel presented the results of his experiments with nearly 30,000 pea plants to the local natural history society. He demonstrated that traits are transmitted faithfully from parents to offspring in specific patterns. In 1866, he published his work, *Experiments in Plant Hybridization*,¹ in the proceedings of the Natural History Society of Brunn.



Figure 5.1.1.1: Johann Gregor Mendel set the framework for the study of genetics.

Mendel's work went virtually unnoticed by the scientific community, which incorrectly believed that the process of inheritance involved a blending of parental traits that produced an intermediate physical appearance in offspring. This hypothetical process appeared to be correct because of what we know now as continuous variation. Continuous variation is the range of small differences we see among individuals in a characteristic like human height. It does appear that offspring are a “blend” of their parents' traits when we look at characteristics that exhibit continuous variation. Mendel worked instead with traits that show discontinuous variation. Discontinuous variation is the variation seen among individuals when each individual shows one of two—or a very few—easily distinguishable traits, such as violet or white flowers. Mendel's choice of these kinds of traits allowed him to see experimentally that the traits were not blended in the offspring as would have been expected at the time, but that they were inherited as distinct traits. In 1868, Mendel became abbot of the monastery and exchanged his scientific pursuits for his pastoral duties. He was not recognized for his extraordinary scientific contributions during his lifetime; in fact, it was not until 1900 that his work was rediscovered, reproduced, and revitalized by scientists on the brink of discovering the chromosomal basis of heredity.

Mendel's Crosses

Mendel's seminal work was accomplished using the garden pea, *Pisum sativum*, to study inheritance. This species naturally self-fertilizes, meaning that pollen encounters ova within the same flower. The flower petals remain sealed tightly until pollination is completed to prevent the pollination of other plants. The result is highly inbred, or “true-breeding,” pea plants. These are plants that always produce offspring that look like the parent. By experimenting with true-breeding pea plants, Mendel avoided the appearance of unexpected traits in offspring that might occur if the plants were not true breeding. The garden pea also grows to maturity within one season, meaning that several generations could be evaluated over a relatively short time. Finally, large quantities of garden peas could be cultivated simultaneously, allowing Mendel to conclude that his results did not come about simply by chance.

Mendel performed hybridizations, which involve mating two true-breeding individuals that have different traits. In the pea, which is naturally self-pollinating, this is done by manually transferring pollen from the anther of a mature pea plant of one variety to the stigma of a separate mature pea plant of the second variety.

Plants used in first-generation crosses were called P, or parental generation, plants (Figure 5.1.1.2). Mendel collected the seeds produced by the P plants that resulted from each cross and grew them the following season. These offspring were called the F₁, or the first filial (filial = daughter or son), generation. Once Mendel examined the characteristics in the F₁ generation of plants, he allowed them to self-fertilize naturally. He then collected and grew the seeds from the F₁ plants to produce the F₂, or second filial, generation. Mendel's experiments extended beyond the F₂ generation to the F₃ generation, F₄ generation, and so on, but it was the ratio of characteristics in the P, F₁, and F₂ generations that were the most intriguing and became the basis of Mendel's postulates.

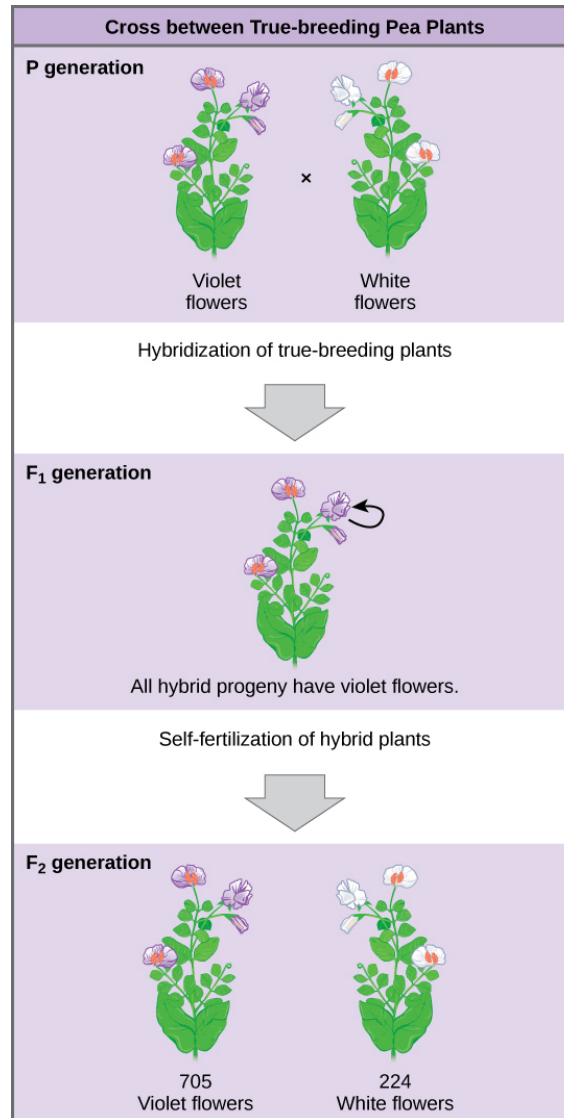


Figure 5.1.1.2: Mendel's process for performing crosses included examining flower color.

Garden Pea Characteristics Revealed the Basics of Heredity

In his 1865 publication, Mendel reported the results of his crosses involving seven different characteristics, each with two contrasting traits. A trait is defined as a variation in the physical appearance of a heritable characteristic. The characteristics included plant height, seed texture, seed color, flower color, pea-pod size, pea-pod color, and flower position. For the characteristic of flower color, for example, the two contrasting traits were white versus violet. To fully examine each characteristic, Mendel generated large numbers of F₁ and F₂ plants and reported results from thousands of F₂ plants.

What results did Mendel find in his crosses for flower color? First, Mendel confirmed that he was using plants that bred true for white or violet flower color. Irrespective of the number of generations that Mendel examined, all self-crossed offspring of parents with white flowers had white flowers, and all self-crossed offspring of parents with violet flowers had violet flowers. In addition,

Mendel confirmed that, other than flower color, the pea plants were physically identical. This was an important check to make sure that the two varieties of pea plants only differed with respect to one trait, flower color.

Once these validations were complete, Mendel applied the pollen from a plant with violet flowers to the stigma of a plant with white flowers. After gathering and sowing the seeds that resulted from this cross, Mendel found that 100 percent of the F_1 hybrid generation had violet flowers. Conventional wisdom at that time would have predicted the hybrid flowers to be pale violet or for hybrid plants to have equal numbers of white and violet flowers. In other words, the contrasting parental traits were expected to blend in the offspring. Instead, Mendel's results demonstrated that the white flower trait had completely disappeared in the F_1 generation.

Importantly, Mendel did not stop his experimentation there. He allowed the F_1 plants to self-fertilize and found that 705 plants in the F_2 generation had violet flowers and 224 had white flowers. This was a ratio of 3.15 violet flowers to one white flower, or approximately 3:1. When Mendel transferred pollen from a plant with violet flowers to the stigma of a plant with white flowers and vice versa, he obtained approximately the same ratio irrespective of which parent—male or female—contributed which trait. This is called a reciprocal cross—a paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross. For the other six characteristics that Mendel examined, the F_1 and F_2 generations behaved in the same way that they behaved for flower color. One of the two traits would disappear completely from the F_1 generation, only to reappear in the F_2 generation at a ratio of roughly 3:1 (Figure 5.1.1.3).

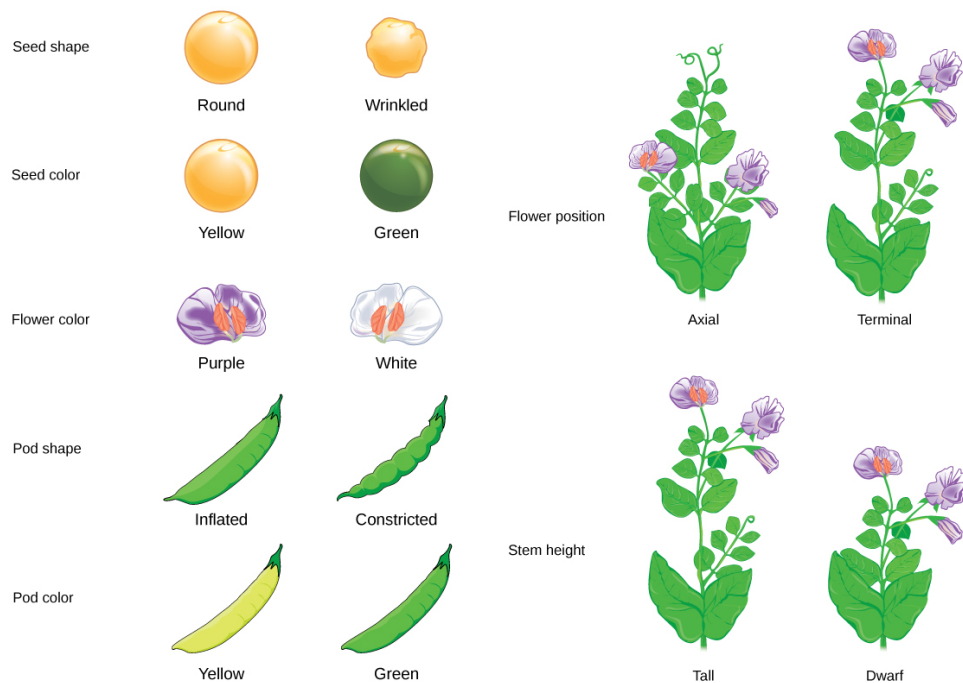


Figure 5.1.1.3: Mendel identified seven pea plant characteristics.

Upon compiling his results for many thousands of plants, Mendel concluded that the characteristics could be divided into expressed and latent traits. He called these dominant and recessive traits, respectively. Dominant traits are those that are inherited unchanged in a hybridization. Recessive traits become latent, or disappear in the offspring of a hybridization. The recessive trait does, however, reappear in the progeny of the hybrid offspring. An example of a dominant trait is the violet-colored flower trait. For this same characteristic (flower color), white-colored flowers are a recessive trait. The fact that the recessive trait reappeared in the F_2 generation meant that the traits remained separate (and were not blended) in the plants of the F_1 generation. Mendel proposed that this was because the plants possessed two copies of the trait for the flower-color characteristic, and that each parent transmitted one of their two copies to their offspring, where they came together. Moreover, the physical observation of a dominant trait could mean that the genetic composition of the organism included two dominant versions of the characteristic, or that it included one dominant and one recessive version. Conversely, the observation of a recessive trait meant that the organism lacked any dominant versions of this characteristic.

Section Summary

Working with garden pea plants, Mendel found that crosses between parents that differed for one trait produced F_1 offspring that all expressed one parent's traits. The traits that were visible in the F_1 generation are referred to as dominant, and traits that disappear in the F_1 generation are described as recessive. When the F_1 plants in Mendel's experiment were self-crossed, the F_2 offspring exhibited the dominant trait or the recessive trait in a 3:1 ratio, confirming that the recessive trait had been transmitted faithfully from the original P parent. Reciprocal crosses generated identical F_1 and F_2 offspring ratios. By examining sample sizes, Mendel showed that traits were inherited as independent events.

Footnotes

1. Johann Gregor Mendel, "Versuche über Pflanzenhybriden." *Verhandlungen des naturforschenden Vereines in Brünn*, Bd. IV für das Jahr, 1865 Abhandlungen (1866):3–47. [for English translation, see www.mendelweb.org/Mendel.plain.html]

Glossary

continuous variation

a variation in a characteristic in which individuals show a range of traits with small differences between them

discontinuous variation

a variation in a characteristic in which individuals show two, or a few, traits with large differences between them

dominant

describes a trait that masks the expression of another trait when both versions of the gene are present in an individual

F_1

the first filial generation in a cross; the offspring of the parental generation

F_2

the second filial generation produced when F_1 individuals are self-crossed or fertilized with each other

hybridization

the process of mating two individuals that differ, with the goal of achieving a certain characteristic in their offspring

model system

a species or biological system used to study a specific biological phenomenon to gain understanding that will be applied to other species

P

the parental generation in a cross

recessive

describes a trait whose expression is masked by another trait when the alleles for both traits are present in an individual

reciprocal cross

a paired cross in which the respective traits of the male and female in one cross become the respective traits of the female and male in the other cross

trait

a variation in an inherited characteristic

Contributors and Attributions

- Samantha Fowler (Clayton State University), Rebecca Roush (Sandhills Community College), James Wise (Hampton University). Original content by OpenStax (CC BY 4.0; Access for free at <https://cnx.org/contents/b3c1e1d2-83...4-e119a8aafbdd>).

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5.2: Laws of Inheritance

The seven characteristics that Mendel evaluated in his pea plants were each expressed as one of two versions, or traits. Mendel deduced from his results that each individual had two discrete copies of the characteristic that are passed individually to offspring. We now call those two copies genes, which are carried on chromosomes. The reason we have two copies of each gene is that we inherit one from each parent. In fact, it is the chromosomes we inherit and the two copies of each gene are located on paired chromosomes. Recall that in meiosis these chromosomes are separated out into haploid gametes. This separation, or segregation, of the homologous chromosomes means also that only one of the copies of the gene gets moved into a gamete. The offspring are formed when that gamete unites with one from another parent and the two copies of each gene (and chromosome) are restored.

For cases in which a single gene controls a single characteristic, a diploid organism has two genetic copies that may or may not encode the same version of that characteristic. For example, one individual may carry a gene that determines white flower color and a gene that determines violet flower color. Gene variants that arise by mutation and exist at the same relative locations on homologous chromosomes are called alleles. Mendel examined the inheritance of genes with just two allele forms, but it is common to encounter more than two alleles for any given gene in a natural population.

Phenotypes and Genotypes

Two alleles for a given gene in a diploid organism are expressed and interact to produce physical characteristics. The observable traits expressed by an organism are referred to as its phenotype. An organism's underlying genetic makeup, consisting of both the physically visible and the non-expressed alleles, is called its genotype. Mendel's hybridization experiments demonstrate the difference between phenotype and genotype. For example, the phenotypes that Mendel observed in his crosses between pea plants with differing traits are connected to the diploid genotypes of the plants in the P, F₁, and F₂ generations. We will use a second trait that Mendel investigated, seed color, as an example. Seed color is governed by a single gene with two alleles. The yellow-seed allele is dominant and the green-seed allele is recessive. When true-breeding plants were cross-fertilized, in which one parent had yellow seeds and one had green seeds, all of the F₁ hybrid offspring had yellow seeds. That is, the hybrid offspring were phenotypically identical to the true-breeding parent with yellow seeds. However, we know that the allele donated by the parent with green seeds was not simply lost because it reappeared in some of the F₂ offspring (Figure 5.2.1). Therefore, the F₁ plants must have been genotypically different from the parent with yellow seeds.

The P plants that Mendel used in his experiments were each homozygous for the trait he was studying. Diploid organisms that are homozygous for a gene have two identical alleles, one on each of their homologous chromosomes. The genotype is often written as YY or yy, for which each letter represents one of the two alleles in the genotype. The dominant allele is capitalized and the recessive allele is lower case. The letter used for the gene (seed color in this case) is usually related to the dominant trait (yellow allele, in this case, or "Y"). Mendel's parental pea plants always bred true because both produced gametes carried the same allele. When P plants with contrasting traits were cross-fertilized, all of the offspring were heterozygous for the contrasting trait, meaning their genotype had different alleles for the gene being examined. For example, the F₁ yellow plants that received a Y allele from their yellow parent and a y allele from their green parent had the genotype Yy.

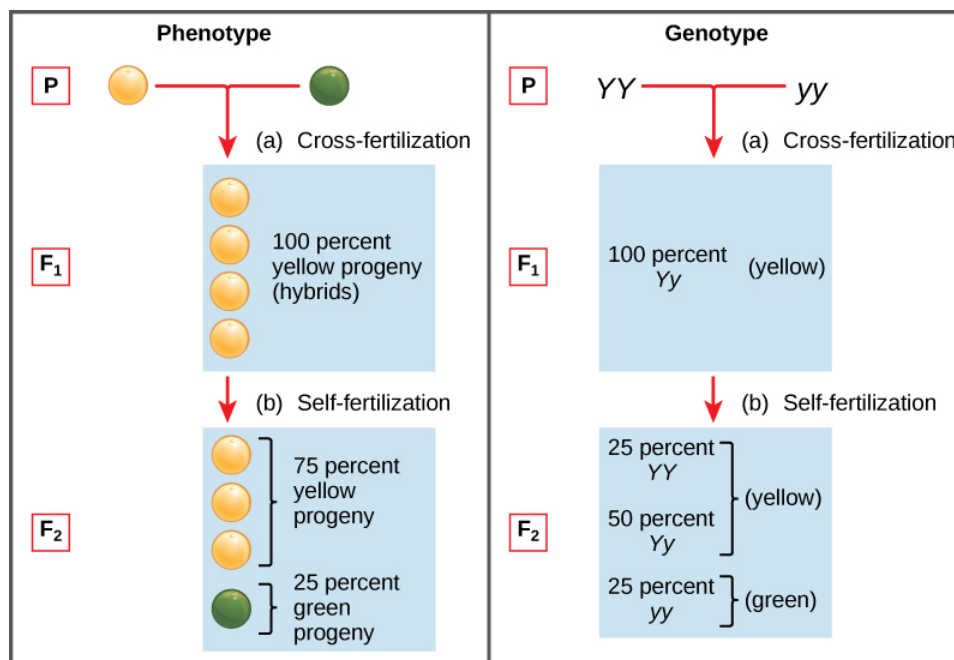


Figure 5.2.1: Phenotypes are physical expressions of traits that are transmitted by alleles. Capital letters represent dominant alleles and lowercase letters represent recessive alleles. The phenotypic ratios are the ratios of visible characteristics. The genotypic ratios are the ratios of gene combinations in the offspring, and these are not always distinguishable in the phenotypes.

Law of Dominance

Our discussion of homozygous and heterozygous organisms brings us to why the F_1 heterozygous offspring were identical to one of the parents, rather than expressing both alleles. In all seven pea-plant characteristics, one of the two contrasting alleles was dominant, and the other was recessive. Mendel called the dominant allele the expressed unit factor; the recessive allele was referred to as the latent unit factor. We now know that these so-called unit factors are actually genes on homologous chromosomes. For a gene that is expressed in a dominant and recessive pattern, homozygous dominant and heterozygous organisms will look identical (that is, they will have different genotypes but the same phenotype), and the recessive allele will only be observed in homozygous recessive individuals (Table 5.2.1).

Table 5.2.1: Correspondence between Genotype and Phenotype for a Dominant-Recessive Characteristic.

	Homozygous	Heterozygous	Homozygous
Genotype	YY	Yy	yy
Phenotype	yellow	yellow	green

Mendel's law of dominance states that in a heterozygote, one trait will conceal the presence of another trait for the same characteristic. For example, when crossing true-breeding violet-flowered plants with true-breeding white-flowered plants, all of the offspring were violet-flowered, even though they all had one allele for violet and one allele for white. Rather than both alleles contributing to a phenotype, the dominant allele will be expressed exclusively. The recessive allele will remain latent, but will be transmitted to offspring in the same manner as that by which the dominant allele is transmitted. The recessive trait will only be expressed by offspring that have two copies of this allele (Figure 5.2.2), and these offspring will breed true when self-crossed.



Figure 5.2.2: The allele for albinism, expressed here in humans, is recessive. Both of this child's parents carried the recessive allele.

Monohybrid Cross and the Punnett Square

When fertilization occurs between two true-breeding parents that differ by only the characteristic being studied, the process is called a monohybrid cross, and the resulting offspring are called monohybrids. Mendel performed seven types of monohybrid crosses, each involving contrasting traits for different characteristics. Out of these crosses, all of the F_1 offspring had the phenotype of one parent, and the F_2 offspring had a 3:1 phenotypic ratio. On the basis of these results, Mendel postulated that each parent in the monohybrid cross contributed one of two paired unit factors to each offspring, and every possible combination of unit factors was equally likely.

The results of Mendel's research can be explained in terms of probabilities, which are mathematical measures of likelihood. The probability of an event is calculated by the number of times the event occurs divided by the total number of opportunities for the event to occur. A probability of one (100 percent) for some event indicates that it is guaranteed to occur, whereas a probability of zero (0 percent) indicates that it is guaranteed to not occur, and a probability of 0.5 (50 percent) means it has an equal chance of occurring or not occurring.

To demonstrate this with a monohybrid cross, consider the case of true-breeding pea plants with yellow versus green seeds. The dominant seed color is yellow; therefore, the parental genotypes were YY for the plants with yellow seeds and yy for the plants with green seeds. A Punnett square, devised by the British geneticist Reginald Punnett, is useful for determining probabilities because it is drawn to predict all possible outcomes of all possible random fertilization events and their expected frequencies. Figure 5.2.5 shows a Punnett square for a cross between a plant with yellow peas and one with green peas. To prepare a Punnett square, all possible combinations of the parental alleles (the genotypes of the gametes) are listed along the top (for one parent) and side (for the other parent) of a grid. The combinations of egg and sperm gametes are then made in the boxes in the table on the basis of which alleles are combining. Each box then represents the diploid genotype of a zygote, or fertilized egg. Because each possibility is equally likely, genotypic ratios can be determined from a Punnett square. If the pattern of inheritance (dominant and recessive) is known, the phenotypic ratios can be inferred as well. For a monohybrid cross of two true-breeding parents, each parent contributes one type of allele. In this case, only one genotype is possible in the F_1 offspring. All offspring are Yy and have yellow seeds.

When the F_1 offspring are crossed with each other, each has an equal probability of contributing either a Y or a y to the F_2 offspring. The result is a 1 in 4 (25 percent) probability of both parents contributing a Y , resulting in an offspring with a yellow phenotype; a 25 percent probability of parent A contributing a Y and parent B a y , resulting in offspring with a yellow phenotype; a 25 percent probability of parent A contributing a y and parent B a Y , also resulting in a yellow phenotype; and a (25 percent) probability of both parents contributing a y , resulting in a green phenotype. When counting all four possible outcomes, there is a 3 in 4 probability of offspring having the yellow phenotype and a 1 in 4 probability of offspring having the green phenotype. This explains why the results of Mendel's F_2 generation occurred in a 3:1 phenotypic ratio. Using large numbers of crosses, Mendel was able to calculate probabilities, found that they fit the model of inheritance, and use these to predict the outcomes of other crosses.

Law of Segregation

Observing that true-breeding pea plants with contrasting traits gave rise to F_1 generations that all expressed the dominant trait and F_2 generations that expressed the dominant and recessive traits in a 3:1 ratio, Mendel proposed the law of segregation. This law states that paired unit factors (genes) must segregate equally into gametes such that offspring have an equal likelihood of inheriting either factor. For the F_2 generation of a monohybrid cross, the following three possible combinations of genotypes result: homozygous dominant, heterozygous, or homozygous recessive. Because heterozygotes could arise from two different pathways (receiving one dominant and one recessive allele from either parent), and because heterozygotes and homozygous dominant individuals are phenotypically identical, the law supports Mendel's observed 3:1 phenotypic ratio. The equal segregation of alleles is the reason we can apply the Punnett square to accurately predict the offspring of parents with known genotypes. The physical basis of Mendel's law of segregation is the first division of meiosis in which the homologous chromosomes with their different versions of each gene are segregated into daughter nuclei. This process was not understood by the scientific community during Mendel's lifetime (Figure 5.2.3).

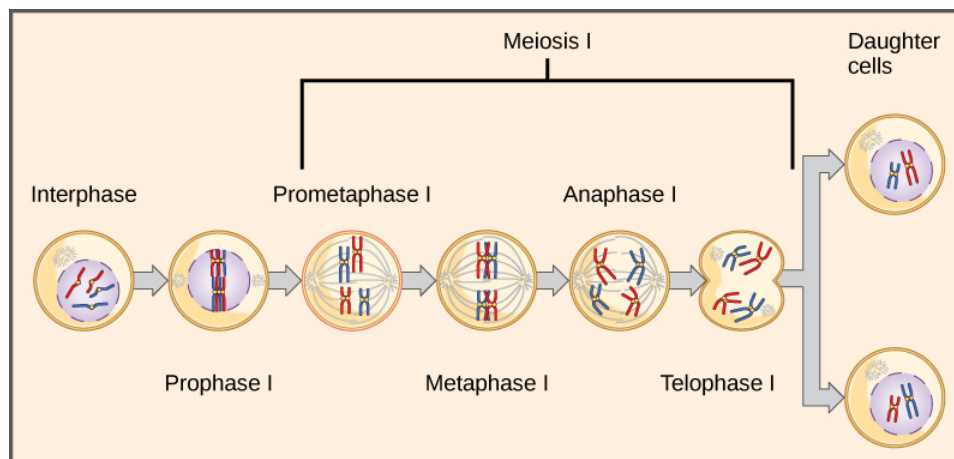


Figure 5.2.3: The first division in meiosis is shown.

Test Cross

Beyond predicting the offspring of a cross between known homozygous or heterozygous parents, Mendel also developed a way to determine whether an organism that expressed a dominant trait was a heterozygote or a homozygote. Called the test cross, this technique is still used by plant and animal breeders. In a test cross, the dominant-expressing organism is crossed with an organism that is homozygous recessive for the same characteristic. If the dominant-expressing organism is a homozygote, then all F_1 offspring will be heterozygotes expressing the dominant trait (Figure 5.2.4). Alternatively, if the dominant-expressing organism is a heterozygote, the F_1 offspring will exhibit a 1:1 ratio of heterozygotes and recessive homozygotes (Figure 5.2.4). The test cross further validates Mendel's postulate that pairs of unit factors segregate equally.

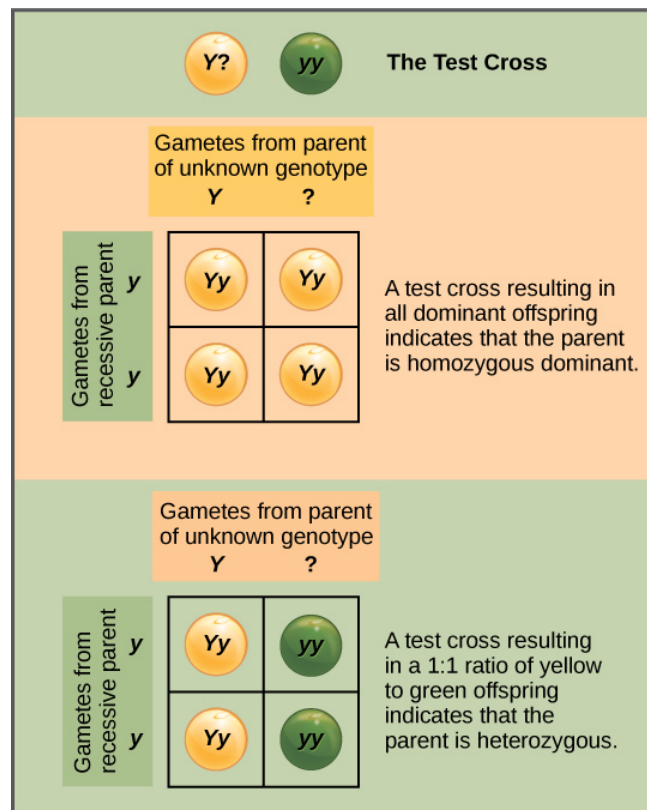


Figure 5.2.4: A test cross can be performed to determine whether an organism expressing a dominant trait is a homozygote or a heterozygote.

ART CONNECTION

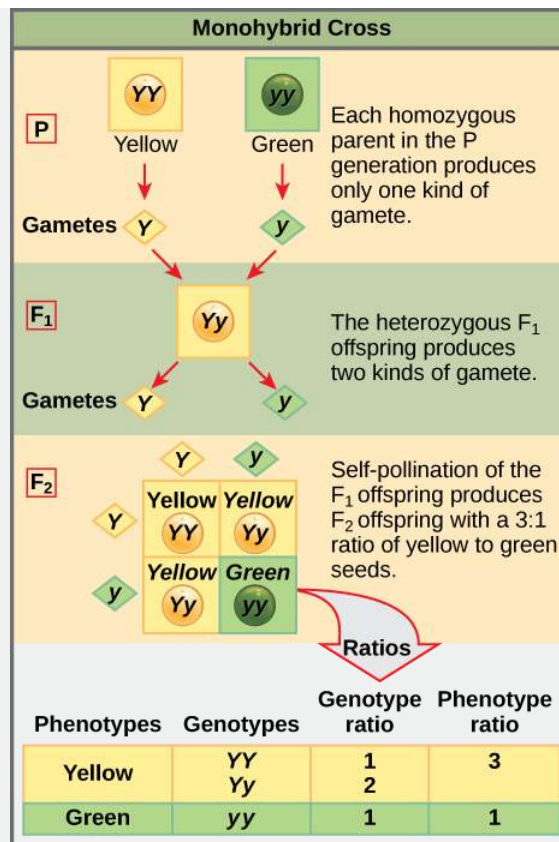


Figure 5.2.5: This Punnett square shows the cross between plants with yellow seeds and green seeds. The cross between the true-breeding P plants produces F₁ heterozygotes that can be self-fertilized. The self-cross of the F₁ generation can be analyzed with a Punnett square to predict the genotypes of the F₂ generation. Given an inheritance pattern of dominant–recessive, the genotypic and phenotypic ratios can then be determined.

In pea plants, round peas (*R*) are dominant to wrinkled peas (*r*). You do a test cross between a pea plant with wrinkled peas (genotype *rr*) and a plant of unknown genotype that has round peas. You end up with three plants, all which have round peas. From this data, can you tell if the parent plant is homozygous dominant or heterozygous?

Law of Independent Assortment

Mendel's law of independent assortment states that genes do not influence each other with regard to the sorting of alleles into gametes, and every possible combination of alleles for every gene is equally likely to occur. Independent assortment of genes can be illustrated by the dihybrid cross, a cross between two true-breeding parents that express different traits for two characteristics. Consider the characteristics of seed color and seed texture for two pea plants, one that has wrinkled, green seeds (*rryy*) and another that has round, yellow seeds (*RRYY*). Because each parent is homozygous, the law of segregation indicates that the gametes for the wrinkled–green plant all are *ry*, and the gametes for the round–yellow plant are all *RY*. Therefore, the F₁ generation of offspring all are *RrYy* (Figure 5.2.6).

ART CONNECTION

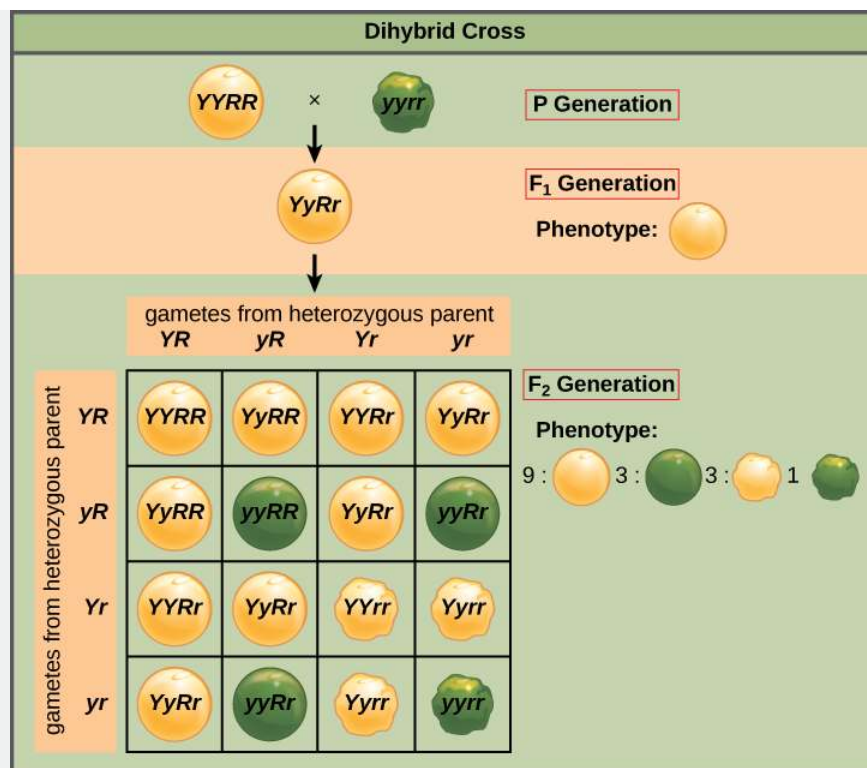


Figure 5.2.6: A dihybrid cross in pea plants involves the genes for seed color and texture. The P cross produces F₁ offspring that are all heterozygous for both characteristics. The resulting 9:3:3:1 F₂ phenotypic ratio is obtained using a Punnett square.

In pea plants, purple flowers (P) are dominant to white (p), and yellow peas (Y) are dominant to green (y). What are the possible genotypes and phenotypes for a cross between $PpYY$ and $ppYy$ pea plants? How many squares would you need to complete a Punnett square analysis of this cross?

The gametes produced by the F₁ individuals must have one allele from each of the two genes. For example, a gamete could get an R allele for the seed shape gene and either a Y or a y allele for the seed color gene. It cannot get both an R and an r allele; each gamete can have only one allele per gene. The law of independent assortment states that a gamete into which an r allele is sorted would be equally likely to contain either a Y or a y allele. Thus, there are four equally likely gametes that can be formed when the $RrYy$ heterozygote is self-crossed, as follows: RY , rY , Ry , and ry . Arranging these gametes along the top and left of a 4×4 Punnett square (Figure 5.2.6) gives us 16 equally likely genotypic combinations. From these genotypes, we find a phenotypic ratio of 9 round–yellow:3 round–green:3 wrinkled–yellow:1 wrinkled–green (Figure 5.2.6). These are the offspring ratios we would expect, assuming we performed the crosses with a large enough sample size.

The physical basis for the law of independent assortment also lies in meiosis I, in which the different homologous pairs line up in random orientations. Each gamete can contain any combination of paternal and maternal chromosomes (and therefore the genes on them) because the orientation of tetrads on the metaphase plane is random (Figure 5.2.7).

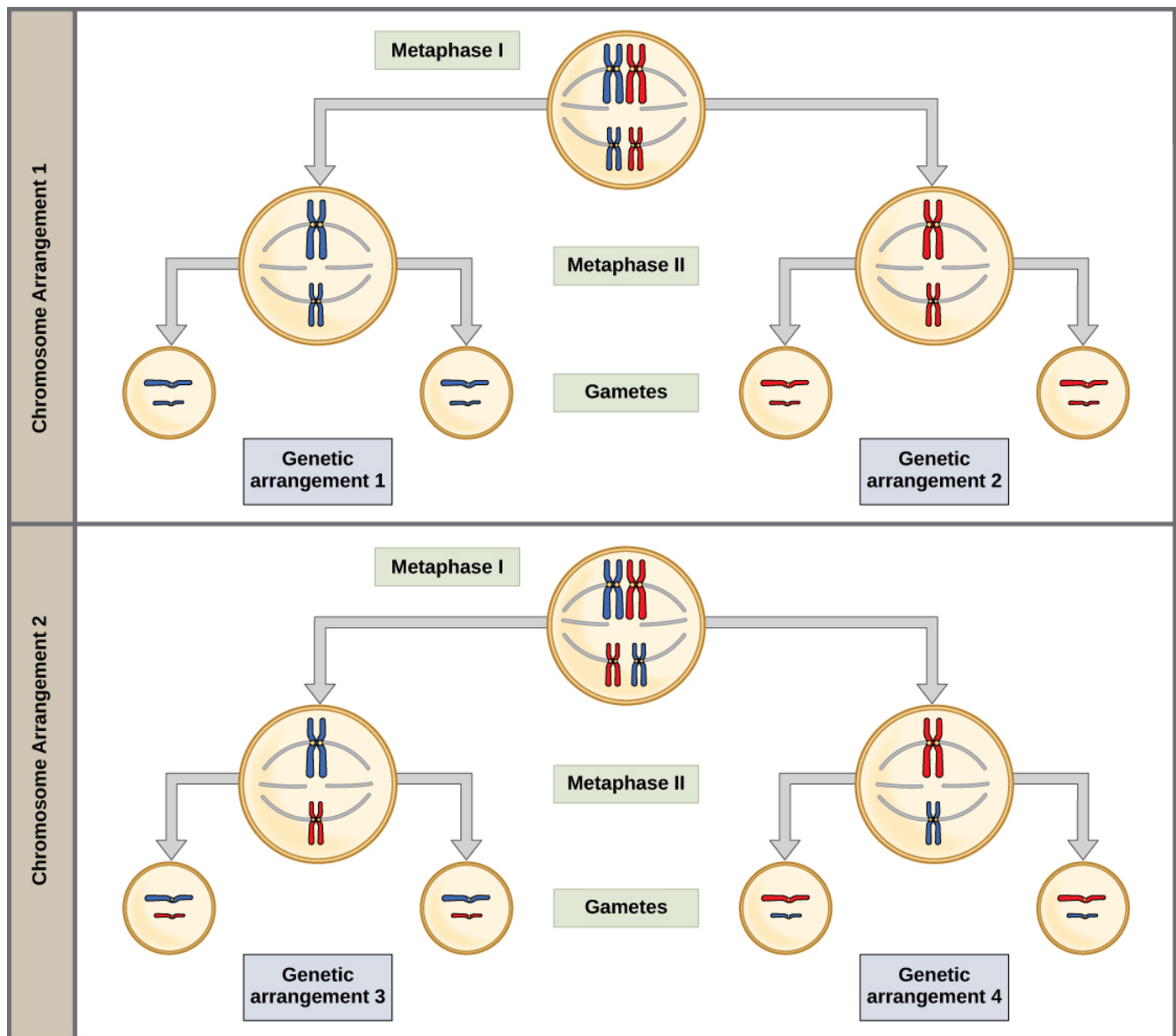


Figure 5.2.7: The random segregation into daughter nuclei that happens during the first division in meiosis can lead to a variety of possible genetic arrangements.

Section Summary

When true-breeding, or homozygous, individuals that differ for a certain trait are crossed, all of the offspring will be heterozygous for that trait. If the traits are inherited as dominant and recessive, the F_1 offspring will all exhibit the same phenotype as the parent homozygous for the dominant trait. If these heterozygous offspring are self-crossed, the resulting F_2 offspring will be equally likely to inherit gametes carrying the dominant or recessive trait, giving rise to offspring of which one quarter are homozygous dominant, half are heterozygous, and one quarter are homozygous recessive. Because homozygous dominant and heterozygous individuals are phenotypically identical, the observed traits in the F_2 offspring will exhibit a ratio of three dominant to one recessive.

Mendel postulated that genes (characteristics) are inherited as pairs of alleles (traits) that behave in a dominant and recessive pattern. Alleles segregate into gametes such that each gamete is equally likely to receive either one of the two alleles present in a diploid individual. In addition, genes are assorted into gametes independently of one another. That is, in general, alleles are not more likely to segregate into a gamete with a particular allele of another gene.

Art Connections

Figure 5.2.5: In pea plants, round peas (R) are dominant to wrinkled peas (r). You do a test cross between a pea plant with wrinkled peas (genotype rr) and a plant of unknown genotype that has round peas. You end up with three plants, all which have round peas. From this data, can you tell if the parent plant is homozygous dominant or heterozygous?

Answer

You cannot be sure if the plant is homozygous or heterozygous as the data set is too small: by random chance, all three plants might have acquired only the dominant gene even if the recessive one is present.

Figure 5.2.6: In pea plants, purple flowers (P) are dominant to white (p), and yellow peas (Y) are dominant to green (y). What are the possible genotypes and phenotypes for a cross between $PpYY$ and $ppYy$ pea plants? How many squares would you need to complete a Punnett square analysis of this cross?

Answer

The possible genotypes are $PpYY$, $PpYy$, $ppYY$, and $ppYy$. The former two genotypes would result in plants with purple flowers and yellow peas, while the latter two genotypes would result in plants with white flowers with yellow peas, for a 1:1 ratio of each phenotype. You only need a 2×2 Punnett square (four squares total) to do this analysis because two of the alleles are homozygous.

Glossary

allele

one of two or more variants of a gene that determines a particular trait for a characteristic

dihybrid

the result of a cross between two true-breeding parents that express different traits for two characteristics

genotype

the underlying genetic makeup, consisting of both physically visible and non-expressed alleles, of an organism

heterozygous

having two different alleles for a given gene on the homologous chromosomes

homozygous

having two identical alleles for a given gene on the homologous chromosomes

law of dominance

in a heterozygote, one trait will conceal the presence of another trait for the same characteristic

law of independent assortment

genes do not influence each other with regard to sorting of alleles into gametes; every possible combination of alleles is equally likely to occur

law of segregation

paired unit factors (i.e., genes) segregate equally into gametes such that offspring have an equal likelihood of inheriting any combination of factors

monohybrid

the result of a cross between two true-breeding parents that express different traits for only one characteristic

phenotype

the observable traits expressed by an organism

Punnett square

a visual representation of a cross between two individuals in which the gametes of each individual are denoted along the top and side of a grid, respectively, and the possible zygotic genotypes are recombined at each box in the grid

test cross

a cross between a dominant expressing individual with an unknown genotype and a homozygous recessive individual; the offspring phenotypes indicate whether the unknown parent is heterozygous or homozygous for the dominant trait

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5.3: Mendelian Genetics

Learning Objectives

- Define and identify examples of: homozygous, heterozygous, allele, gene, locus, dominant, recessive, genotype, phenotype.
- Construct genetic crosses for P₀, F₁, and F₂ generations and predict the genotypes and phenotypes of offspring.
- Explain Mendel's laws of segregation and independent assortment, and how they predict the 3:1 dominant-to-recessive phenotypic ratio among the F₂ of a monohybrid cross, or the 9:3:3:1 phenotypic ratio in a dihybrid cross, respectively. Relate the key events of meiosis that explain Mendel's first and second laws.
- Be able to draw chromosomes during meiosis with alleles labeled.
- Interpret phenotypic ratios of progeny in experimental organisms to infer how particular traits are inherited.
- Predict genotypic and phenotypic ratios or probabilities of outcomes among progeny of single factor and multifactor crosses using simple rules of probability (sum rule and product rule).
- Cite the most common molecular explanations for dominant and recessive alleles.

Introduction to Gregor Mendel and his Work



Video 5.3.1: Shirley Tilghman describes Gregor Mendel's Famous Genetics Experiment. (CC BY NC XBio via Wonder Collaborative on youtube.com <https://youtu.be/XgXLVaBjQqw>)

Mendel Studied Visible Character Traits in Pea Plants

Through careful study of patterns of inheritance, Mendel recognized that a single trait could exist in different versions, or **alleles**, even within an individual plant or animal. Recalling that genes contain information needed to make proteins, we now understand that alleles are differences in gene sequence. If these differences alter the production, structure, or function of the protein, an observable or measurable change in the organism may occur. For example, Mendel identified two forms of a gene for seed color: one allele gave green seeds and the other gave yellow seeds.

Seed		Flower	Pod		Stem	
Form	Color	Color	Form	Color	Place	Size
						
Round	Yellow	White	Full	Yellow	Axial pods, Flowers along	Long (6-7ft)
						
Wrinkled	Green	Purple	Constricted	Green	Terminal pods, Flowers top	Short (~1ft)
1	2	3	4	5	6	7

Figure 5.3.1: Seven traits Mendel studied in peas. (Wikipedia-Mariana Ruiz-PD)

Representing genes and alleles

Alleles are forms of genes, if genes are DNA sequences, then alleles are variations in the sequence of a gene.

In genetics, you may encounter different ways of representing alleles. Traditionally, genes are represented as letters when studying Mendelian genetics. This representation is usually easy to follow in problems involving crosses; however, using letters does not reflect our modern understanding of the genetic differences between alleles, which often involves knowing whether or not a product is functional or how the allele was identified.

- A plus can be used to indicate that the gene product of an allele is functional.
- A minus can be used to indicate that the gene product is not functional.
- If sequence information is known, the nucleotide or amino acid change can be identified.
- In model organisms, alleles are often given numbers when they are identified as mutants and these numbers can be used to identify different alleles of the gene.

The table shows some examples of how we might represent genes and alleles.

Examples of ways genes can be represented	Examples of representing wild-type alleles	Examples of representing the mutant alleles
<i>C</i> representing a hypothetical gene	<i>C</i> or <i>C</i> ⁺	<i>c</i> or <i>C</i> ⁻ or <i>C</i> ¹
<i>white</i> or <i>w</i> gene in <i>Drosophila</i>	<i>white</i> or <i>white</i> ⁺ or <i>w</i> ⁺	<i>w</i> ⁻ or <i>w</i> ¹¹¹⁸
<i>ced-1</i> gene in <i>C. elegans</i>	<i>ced-1</i> or <i>ced-1</i> (+) or <i>ced-1</i> ⁺	<i>ced-1</i> (-) or <i>ced-1</i> (<i>e1754</i>)
<i>CFTR</i> gene in humans <i>Cfr</i> gene in mouse	<i>CFTR</i> or <i>CFTR</i> ⁺	<i>CFTR</i> (<i>delF508</i>) or <i>CFTR</i> (<i>482G-A</i>)

Heterozygous and homozygous

Mendel's work and discoveries are especially remarkable because he made his observations and conclusions (in 1865) without knowing about the relationships between genes, chromosomes, and DNA. We now know the reason why more than one allele of a gene can be present in an individual: most eukaryotic organisms have at least two sets of homologous chromosomes. For organisms that are predominantly diploid, such as humans or Mendel's peas, chromosomes exist as pairs, with one homolog inherited from each parent. Diploid cells therefore contain two different alleles of each gene, with one allele on each member of a pair of homologous chromosomes. If both alleles of a particular gene are identical, the individual is said to be **homozygous** for that gene. On the other hand, if the alleles are different from each other, the genotype is **heterozygous**.

Although a typical diploid individual can have at most two different alleles of a particular gene, many more than two different alleles can exist in a population of individuals. In a natural population the most common allelic form is usually called the **wild-type** allele. However, in many populations there can be multiple **variants** at the DNA sequence level that are visibly indistinguishable because all produce a normal, wild-type appearance. There can also be various **mutant** alleles (in wild populations and in lab strains) that vary from wild type in their appearance, each with a different change at the DNA sequence level. Such collections of mutations are known as an **allelic series**.

wild type vs wild-type

The noun **wild type** (without a hyphen) means the the most common form of a gene, phenotype, or organism under standard conditions.

- Example: The mutant worms produce fewer offspring than **wild type**.

The adjective **wild-type** (with a hyphen) can be used to describe a gene, allele, organism, phenotype, or trait as what is most commonly present among a population or under standard conditions.

- Example: A **wild-type** worm produces about 300 offspring.

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5.3.1: Monohybrid Crosses and Segregation

True Breeding Lines

Mendel used **true-breeding** lines of pea plants, which are in-bred populations of plants or animals in which all parents and their offspring (over many generations) have the same phenotypes with respect to a particular trait. True breeding lines are useful, because they are typically assumed to be homozygous for the alleles that affect the trait of interest. When two individuals that are homozygous for the same alleles are crossed, all of their offspring will all also be homozygous. The continuation of such crosses constitutes a true breeding line or strain. A large variety of different strains, each with a different, true breeding character, can be collected and maintained for genetic research.

Monohybrid Crosses

A **monohybrid cross** is one in which both parents are heterozygous (or a hybrid) for a single (mono) trait. The trait might be petal color in pea plants. When conducting crosses, the first generation is called P (or P_0), the second generation is F_1 (F is for filial), and the next generation is F_2 .

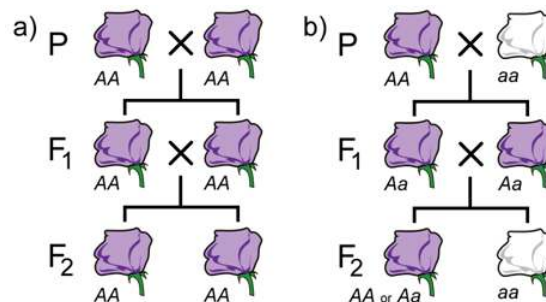


Figure 5.3.1.1: (a) A true-breeding line (b) A monohybrid cross produced by mating two different pure-breeding lines. (Original-Deholos-CC:AN)

Using monohybrid crosses, Mendel observed that although different alleles could influence a single trait, they remained indivisible and could be inherited separately. Additionally, the allele could be present but invisible in one generation, only to reappear in the next generation.

Definition: Mendel's First Law

The Law of Segregation states that during gamete formation, the two alleles at a gene locus segregate from each other; each gamete has an equal probability of containing either allele.

Punnett Squares

Given the genotypes of any two parents, we can predict the genotypes of gametes that will be produced during meiosis. Using that information, we can predict all of the possible genotypes of the offspring. Furthermore, if we also know the dominance relationships for all of the alleles, we can predict the phenotypes of the offspring. A convenient method for calculating the expected genotype and phenotype ratios from a cross was invented by Reginald Punnett. A **Punnett square** is a matrix in which all of the possible gametes produced by one parent are listed along one axis, and the gametes from the other parent are listed along the other axis. Each possible combination of gametes is listed at the intersection of each row and column. Punnett squares can also be used to calculate the frequency of types offspring that are expected.

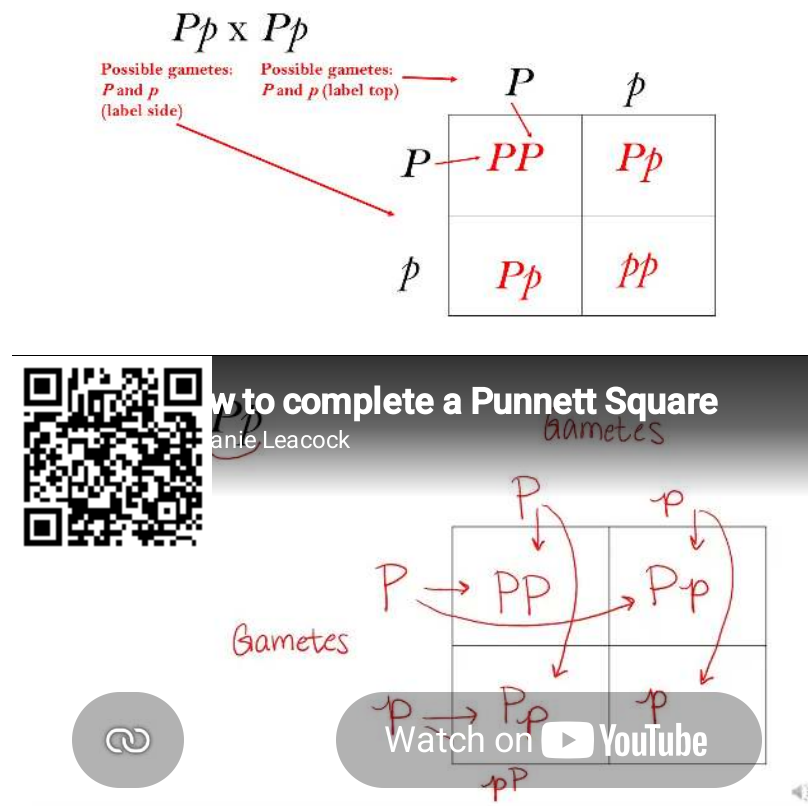
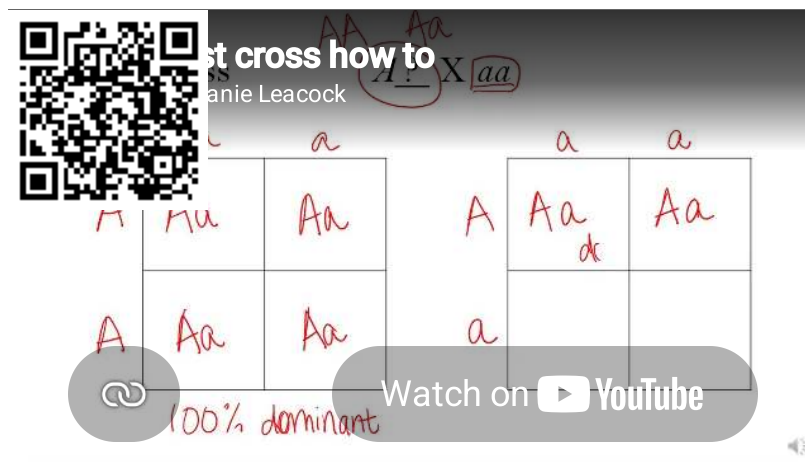


Figure 5.3.1.1: Top: A Punnett square shows the possible offspring of a monohybrid cross between Pp and Pp parents. Bottom Video: How to set up and complete the square (Copyright CC BY Leacock)

Test Crosses

Knowing the genotypes of an individual is usually an important part of a genetic experiment. However, genotypes cannot be observed directly; they must be inferred based on phenotypes. Because of dominance, it is often not possible to distinguish between a heterozygote and a homozygote based on phenotype alone. To determine the genotype of a specific individual, a **test cross** can be performed, in which the individual with an uncertain genotype is crossed with an individual that is homozygous recessive for all of the loci being tested.

For example, if you were given a pea plant with purple flowers it might be a homozygote (AA) or a heterozygote (Aa). You could cross this purple-flowered plant to a white-flowered plant, because you know the genotype of the plant is homozygous recessive aa . Depending on the genotype of the purple-flowered parent, you will observe different phenotypic ratios in the F_1 generation. If the purple-flowered parent was a homozygote, all of the F_1 progeny will be purple. If the purple-flowered parent was a heterozygote, the F_1 progeny should segregate purple-flowered and white-flowered plants in a 1:1 ratio.



Video 5.3.1.2: How to use a test cross (Copyright CC BY Leacock)

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Molecular basis of dominant and recessive alleles

What determines whether alleles are dominant or recessive?

The alleles have different DNA sequences. Because the sequence of DNA contains information to make products, different sequences can lead to different products. One advantage of diploid species is that there are two copies of every sequence. If one sequence makes a "faulty" or non-functional product, it would be called a **loss-of-function** allele. However, there is likely another sequence that produces a "correct" or functional product. For most genes a single wild-type (usually the normal / functional) allele is capable of producing enough product for the cell, resulting in a **dominant** phenotype. However, if both copies of the gene are loss-of-function alleles, there will not be any functional protein and the **recessive** phenotype will be observed.

An example from pea plants

Mendel studied pea plants in which the peas could be round or wrinkled, with wrinkled being the recessive characteristic. The gene that determines this trait (originally represented as *R*) has since been identified and named *SBE1* (for starch-branching enzyme). The wild-type sequence encodes an enzyme that catalyzes a chemical reaction of carbohydrates to form branched chains of starch in plants. When *SBE1* protein is present and functional, the peas produce branched starch and exhibit a round shape. When *SBE1* protein is absent, the amount of branched starch is reduced, but the levels of disaccharides are higher, resulting in increased water absorption. Later, this water will be lost and the pea will become wrinkled.

Molecular studies of DNA in round (*RR*) and wrinkled (*rr*) plants revealed an insertion of about 800 base pairs in an exon of the *SBE1* gene in the *rr* plants (Bhattacharyya et al, 1990). The insertion is derived from a transposon and disrupts the *SBE1* protein-coding sequence. Therefore, the *RR* plants have two copies of a functional *SBE1* allele (*SBE1*+/+) to make functional *SBE1* enzyme, while *rr* plants have two copies of the *SBE1* gene in which the sequence is disrupted (*SBE1*-/-) and cannot produce any functional *SBE1* enzyme.

So what happens in a heterozygous *Rr* (*SBE1*+/-) plant? These plants have one chromosome with the *SBE1*+ allele and one chromosome with the *SBE1*- allele. The *SBE1*- allele is still transcribed, but does not code for the functional protein during translation. The *SBE1*+ allele is transcribed and translated into a functional enzyme, starch-branching occurs, and the peas have the round phenotype. Although the total amount of *SBE1* enzyme is less than homozygous dominant peas, it is sufficient for the round phenotype and therefore dominant. In this case, molecular discoveries 125 years after Mendel's work reveal the reason for the dominance of the *R* allele over the *r* allele.

References

Bhattacharyya MK, Smith AM, Ellis TH, Hedley C, Martin C. The wrinkled-seed character of pea described by Mendel is caused by a transposon-like insertion in a gene encoding starch-branching enzyme. *Cell*. 1990 Jan 12;60(1):115-22. doi: 10.1016/0092-8674(90)90721-p. PMID: 2153053.

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5.3.2: Dihybrid Crosses and Independent Assortment

Mendel's Second Law

Before Mendel, it had not yet been established that heritable traits were controlled by discrete factors. Therefore an important question was whether distinct traits were controlled by separate factors that were inherited independently of one another. To answer this, Mendel took two apparently unrelated traits, such as seed shape and seed color, and studied their inheritance together in one individual. He studied two variants of each trait: seed color was either green or yellow, and seed shape was either round or wrinkled. When either of these traits was studied individually, the phenotypes segregated in the classical 3:1 ratio among the progeny of a monohybrid cross, with $\frac{3}{4}$ of the seeds green and $\frac{1}{4}$ yellow in one cross, and $\frac{3}{4}$ round and $\frac{1}{4}$ wrinkled in the other cross. Would this be true when both were in the same individual?

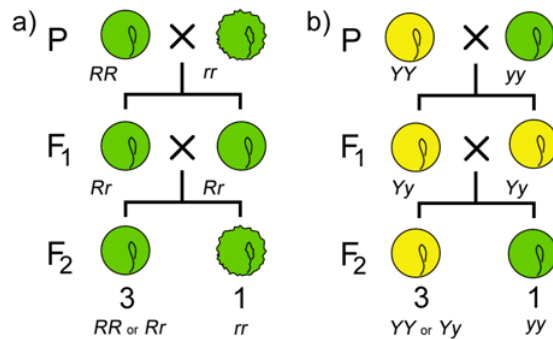


Figure 5.3.2.1: Monohybrid crosses involving two distinct traits, shape which can be round or wrinkled and color which can be yellow or green. a) A monohybrid cross between RR (round peas) and rr (wrinkled peas) plants and b) A monohybrid cross between YY (yellow peas) and yy (green peas) plants. (Original-Deyholos-CC:AN)

To analyze the segregation of both traits at the same time in the same individual, Mendel crossed a pure breeding line of green, wrinkled peas with a pure breeding line of yellow, round peas to produce F_1 progeny that were all yellow and round, and which were also **dihybrids**; they carried two alleles at each of two loci. If the alleles for the two genes for pea shape and pea color cannot be separated from each other, then in the F_2 generation, the offspring should be only green, round pea plants or yellow, wrinkled plants, like the P generation plants.

If the genes controlling shape and color can be inherited independently, then what is the probability of phenotypes in the F_2 generation? Using the product rule, we can multiply the individual probabilities of obtaining a round phenotype ($\frac{3}{4}$) with the probability of obtaining a yellow phenotype ($\frac{3}{4}$), then $\frac{3}{4} \times \frac{3}{4} = \frac{9}{16}$ of the progeny would be both round and green. Likewise, $\frac{3}{4} \times \frac{1}{4} = \frac{3}{16}$ of the progeny would be both round and yellow, and so on. By applying the product rule to all combinations of phenotypes, we can predict a **9:3:3:1** phenotypic ratio among the progeny of a dihybrid cross, if certain conditions are met, including the independent segregation of the alleles at each locus.

Definition: Product Rule

The probability of two or more independent events occurring is the product obtained by multiplying the probabilities of the individual events. If you can use the word "and" to describe the occurrence of the events, then you can usually use the **product rule**. For example, the probability of an organism with phenotype 1 and phenotype 2 is the product of the probability of phenotype 1 times the probability of phenotype 2.

The dihybrid crosses that Mendel performed consistently revealed the 9:3:3:1 ratio in dihybrid crosses, leading him to conclude that the factors controlling the traits are inherited independent of one another, a rule commonly known as the Law of Independent Assortment. Importantly, the F_2 generation includes two phenotype classes (in this example, round, green peas and yellow, wrinkled peas) that are new combinations of phenotypes, distinct from the parental generation plants. Thus, independent assortment contribute to genetic diversity.

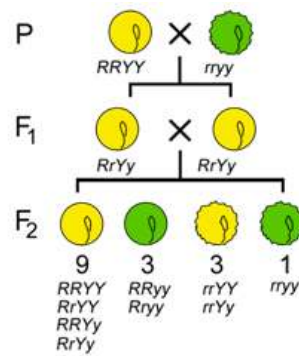


Figure 5.3.2.2: Pure-breeding lines *RRYY* and *rryy* are crossed to produce dihybrids in the F_1 generation. The cross of the F_1 *RrYy* dihybrids produces four phenotypic classes in a ratio of 9:3:3:1. (Original-Deyholos-CC:AN)

Definition: Mendel's Second Law

The **Law of Independent Assortment** states that two loci assort independently of each other during gamete formation.

Table 5.3.2.1: Phenotypic classes expected in monohybrid and dihybrid crosses for two seed traits in pea.

						Frequency of phenotypic crosses within separate monohybrid crosses: seed shape: $\frac{3}{4}$ round $\frac{1}{4}$ wrinkled seed color: $\frac{3}{4}$ yellow $\frac{1}{4}$ green Frequency of phenotypic crosses within a dihybrid cross: $\frac{3}{4}$ round $\times$ $\frac{3}{4}$ yellow = $\frac{9}{16}$ round & yellow $\frac{3}{4}$ round $\times$ $\frac{1}{4}$ green = $\frac{3}{16}$ round & green $\frac{1}{4}$ wrinkled $\times$ $\frac{3}{4}$ yellow = $\frac{3}{16}$ wrinkled & yellow $\frac{1}{4}$ wrinkled $\times$ $\frac{1}{4}$ green = $\frac{1}{16}$ wrinkled & green
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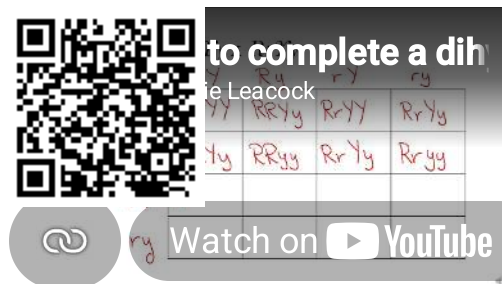
The 9:3:3:1 phenotypic ratio calculated using the product rule can also be obtained using Punnett Square. First, we list the genotypes of the possible gametes along each axis of the Punnett Square. In a diploid with two heterozygous genes of interest, there are up to four combinations of alleles in the gametes of each parent. The gametes from the respective rows and column are then combined in the each cell of the array. When working with two loci, genotypes are written with the symbols for both alleles of one

locus, followed by both alleles of the next locus (e.g. $AaBb$, not $ABab$). Note that the order in which the loci are written does not imply anything about the actual position of the loci on the chromosomes.

	RY	Ry	rY	ry
RY	$RRYY$	$RRYy$	$RrYY$	$RrYy$
Ry	$RRYy$	$RRyy$	$RrYy$	$Rryy$
rY	$RrYY$	$RrYy$	$rrYY$	$rrYy$
ry	$RrYy$	$Rryy$	$rrYy$	$rryy$

Figure 5.3.2.3: A Punnett Square showing the results of a dihybrid cross. Each of the four phenotypic classes is represented by a different color of shading. (Original-Deyholos-CC:AN)

To calculate the expected phenotypic ratios, we assign a phenotype to each of the 16 genotypes in the Punnett Square, based on our knowledge of the alleles and their dominance relationships. In the case of Mendel's seeds, any genotype with at least one R allele and one Y allele will be round and yellow; these genotypes are shown in the nine, green-shaded cells. We can represent all of four of the different genotypes shown in these cells with the notation ($R_Y_$), where the blank line ($_$), means "any allele". The three offspring that have at least one R allele and are homozygous recessive for y (i.e. R_yy) will have a round, green phenotype. Conversely the three progeny that are homozygous recessive r , but have at least one Y allele ($rrY_$) will have wrinkled, yellow seeds. Finally, the rarest phenotypic class of wrinkled, yellow seeds is produced by the doubly homozygous recessive genotype, $rryy$, which is expected to occur in only one of the sixteen possible offspring represented in the square.



Video 5.3.2.1: Watch the video to see how to set up and complete a dihybrid Punnett Square.

Assumptions of the 9:3:3:1 ratio

Both the product rule and the Punnett Square approaches showed that a 9:3:3:1 phenotypic ratio is expected among the progeny of a dihybrid cross such as Mendel's $RrYy \times RrYy$. In making these calculations, we assumed that:

1. both loci assort independently;
2. one allele at each locus is completely dominant; and
3. each of four possible phenotypes can be distinguished unambiguously, with no interactions between the two genes that would alter the phenotypes.

Deviations from the 9:3:3:1 phenotypic ratio may indicate that one or more of the above conditions has not been met. Modified ratios in the progeny of a dihybrid cross can therefore reveal useful information about the genes involved.

Applying the product rule

Mendel's conclusions about the segregation of alleles and independent assortment of genes continue to hold true for inheritance in diploid organisms, in which meiosis produces gametes that have one copy of thousands of genes - not just one or two. However, making a Punnett Square for more than two genes becomes tricky and cumbersome. The Product Rule can be used to predict outcomes when considering more than two genes at a time. Find the probability of each genotype or phenotype and multiply each probability.

Exercise 5.3.2.1

If two organisms with genotype $GgHhJi$ are crossed, what percent of offspring are expected to be homozygous dominant for all three genes?

Hint: GG and HH and JJ

Answer

(Probability GG : $1/4$) $\times$ (Probability HH : $1/4$) $\times$ (Probability JJ : $1/4$) = $1/64$

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5.4: Model Organisms Facilitate Genetic Advances

Model organisms

Many of the great advances in genetics were made using species that are not especially important from a medical, economic, or even ecological perspective. Geneticists, from Mendel onwards, have sought the best organisms for their experiments. Today, a small number of species are widely used as **model organisms** in genetics (Fig 1.17). All of these species have specific characteristics that make large number of them easy to grow and analyze in laboratories: (1) they are small, (2) fast growing with a short generation time, (3) produce lots of progeny from matings that can be easily controlled, (4) have small genomes (small C-value), and (5) are diploid (i.e. chromosomes are present in pairs).

The most commonly used model organism are:

- The prokaryote bacterium, *Escherichia coli*, is the simplest genetic model organism and is often used to clone DNA sequences from other model species.
- Yeast (*Saccharomyces cerevisiae*) is a good general model for the basic functions of eukaryotic cells.
- The roundworm, *Caenorhabditis elegans* is a useful model for the development of multicellular organisms, in part because it is transparent throughout its life cycle, and its cells undergo a well-characterized series of divisions to produce the adult body.
- The fruit fly (*Drosophila melanogaster*) has been studied longer, and probably in more detail, than any of the other genetic model organisms still in use, and is a useful model for studying development as well as physiology and even behavior.
- The mouse (*Mus musculus*) is the model organism most closely related to humans, however there are some practical difficulties working with mice, such as cost, slow reproductive time, and ethical considerations.
- The zebrafish (*Danio rerio*) has more recently been developed by researchers as a genetic model for vertebrates. Unlike mice, zebrafish embryos develop quickly and externally to their mothers, and are transparent, making it easier to study the development of internal structures and organs.
- Finally, a small weed, *Arabidopsis thaliana*, is the most widely studied plant genetic model organism. This provides knowledge that can be applied to other plant species, such as wheat, rice, and corn.

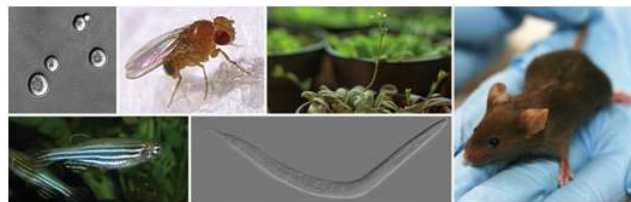


Figure 5.4.17: Some of the most important genetic model organisms in use today. Clockwise from top left: yeast, fruit fly, arabidopsis, mouse, roundworm, zebrafish. (Original/Flickr/Wikipedia – Deyholos/M.Westby/D.Joly/Z.F.Altun/Masur/Azul – CC:AS/ANS/AN &GFDL)

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macromolecule

- 2: Biological Molecules

Mendel's First Law

- 5.3: Mendelian Genetics

Mendel's Second Law

- 5.3.2: Dihybrid Crosses and Independent Assortment

model system

- 5.1.1: Mendel's Experiments

monohybrid

- 5.2: Laws of Inheritance

monohybrid crosses

- 5.3.1: Monohybrid Crosses and Segregation
- 5.3.2: Dihybrid Crosses and Independent Assortment

Monosaccharide

- 2: Biological Molecules

N

nitrogenous base

- 3: The Structure of DNA

nucleic acid

- 2: Biological Molecules

nucleotide

- 2: Biological Molecules

O

OGG

- 5.3: Mendelian Genetics
- 5.3.1: Monohybrid Crosses and Segregation
- 5.4: Model Organisms Facilitate Genetic Advances

oil

- 2: Biological Molecules

OOG

- 5.3: Mendelian Genetics
- 5.3.1: Monohybrid Crosses and Segregation
- 5.4: Model Organisms Facilitate Genetic Advances

P

p

- 5.1.1: Mendel's Experiments

phenotype

- 5.2: Laws of Inheritance

phosphate group

- 3: The Structure of DNA

phospholipid

- 2: Biological Molecules

polypeptide

- 2: Biological Molecules

polysaccharide

- 2: Biological Molecules

protein

- 2: Biological Molecules

Punnett square

- 5.2: Laws of Inheritance
- 5.3.1: Monohybrid Crosses and Segregation

R

recessive

- 5.1.1: Mendel's Experiments

reciprocal cross

- 5.1.1: Mendel's Experiments

Ribonucleic Acid (RNA)

- 2: Biological Molecules

S

Saturated fatty acid

- 2: Biological Molecules

starch

- 2: Biological Molecules

steroid

- 2: Biological Molecules

T

test cross

- 5.2: Laws of Inheritance

trait

- 5.1.1: Mendel's Experiments

triglyceride

- 2: Biological Molecules

U

unsaturated fatty acid

2: Biological Molecules

Glossary

Sample Word 1 | Sample Definition 1

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