

# THE INHERITANCE OF DNA, CHROMOSOMES, AND GENES

## DNA, CHROMOSOMES, AND GENES

Several billion people have lived on the earth since the time of Adam and Eve, and people living in different parts of the world today have very distinct physical characteristics. There are people with very light skin and people with very dark skin. People have various shades of eye color that range from blue to green to brown. There are people of all shapes and sizes, with different interests and even different body chemistry. Is it possible from what we know about human inheritance that everyone could have descended from just two people?

The physical appearance of each person is determined primarily by a substance called *deoxyribonucleic acid* (DNA), which is found in every human cell. DNA is a large molecule consisting of sugars (deoxyribose), phosphates, and four different bases: adenine, cytosine, guanine, and thymine. The bases have specific base partners. Adenine binds with thymine, and cytosine binds with guanine.

A sugar, phosphate, and base are bonded together to form what is called a *nucleotide*. Nucleotides bind to each other to form a long strand of DNA by connecting the phosphate of one nucleotide to the sugar of another nucleotide. DNA is double-stranded, with the two strands of nucleotides connected in the middle by weak bonds that are formed between the bases. The actual structure of DNA looks like a spiral staircase, with the sugar and phosphates on the sides and the steps of the staircase made from the base pairs (Figure 1). This structure can be understood by comparing the double-stranded molecule of DNA to a zipper. The teeth connecting the two sides of a zipper are analogous to the bases that connect the two strands of DNA, and the fabric that holds the zipper teeth are analogous to the sugar-phosphate molecules that hold the DNA bases.

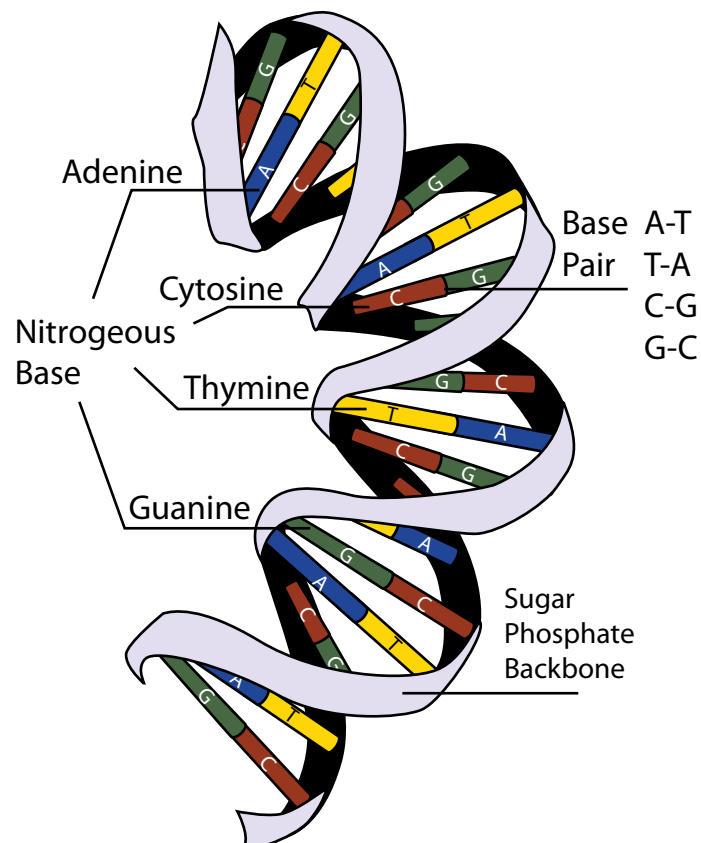


Figure 1



There are three billion pairs of these bases in all of your cells. They provide the information necessary to form your body and make it function. The part of DNA that provides the information for what you or any other living thing looks like is found in the arrangement of the four DNA bases. These bases communicate with the cell by “spelling out” instructions, much the same way the letters on this page spell out words to communicate information to the reader. Sections of DNA, or genes, are “unzipped” by enzymes when it is necessary to instruct the cell to make certain proteins. These proteins in turn determine the characteristics of each kind of living thing.

The large amount of DNA in human cells is organized into 46 packages called chromosomes. Chromosomes are made of DNA and proteins. The protein portion of the chromosome helps to protect and regulate the activities of the DNA. The 46 chromosomes include two copies of 23 different chromosomes ( $2 \times 23 = 46$ ). A person normally receives one of each chromosome from the mother and one from the father. You inherited 23 different chromosomes from your mother, and 23 additional chromosomes from your father, providing you with two sets. Your two sets of chromosomes include two gender-determining chromosomes and 22 pairs of *autosomes*, which are simply the chromosomes that are not sex-determining. Sex-determining chromosomes are represented with the letters X and Y. Girls have two X chromosomes and boys have one X and one Y chromosome. Girls received an X chromosome from each parent, while boys received an X chromosome from their mother and a Y chromosome from their father.

*Genes* are sections of DNA located at specific loci (sites) on a chromosome. These sections can have anywhere from just over 1,000 DNA bases to several thousand bases. A gene includes DNA bases that code for a specific protein and the additional DNA sequences required for the production of the encoded protein. Typically, human genes use approximately 50,000 base pairs of DNA to encode an average-size protein. Most of the gene (95 percent of the sequence) does not encode a protein but is instead involved in the overall production and regulation. Since every cell has a pair of the 23 different chromosomes, it also has a pair of every gene at a specific locus on the chromosome.

Genes have information for a specific trait in the sequence of their DNA. More precisely, genes have information to produce proteins that in turn determine the structure of a living thing and how it functions. As few as one or two differences in the DNA base sequence of genes, or the regulatory sequences that control them, can make humans physically different from each other. These differences affect the amount and activity of the proteins that come from the information in genes. Humans and animals have many of the same genes. Large differences in the sequence of genes, the location of a particular gene on a chromosome, and the regulation of these genes account for many of the observable differences among animals, and between animals and humans.

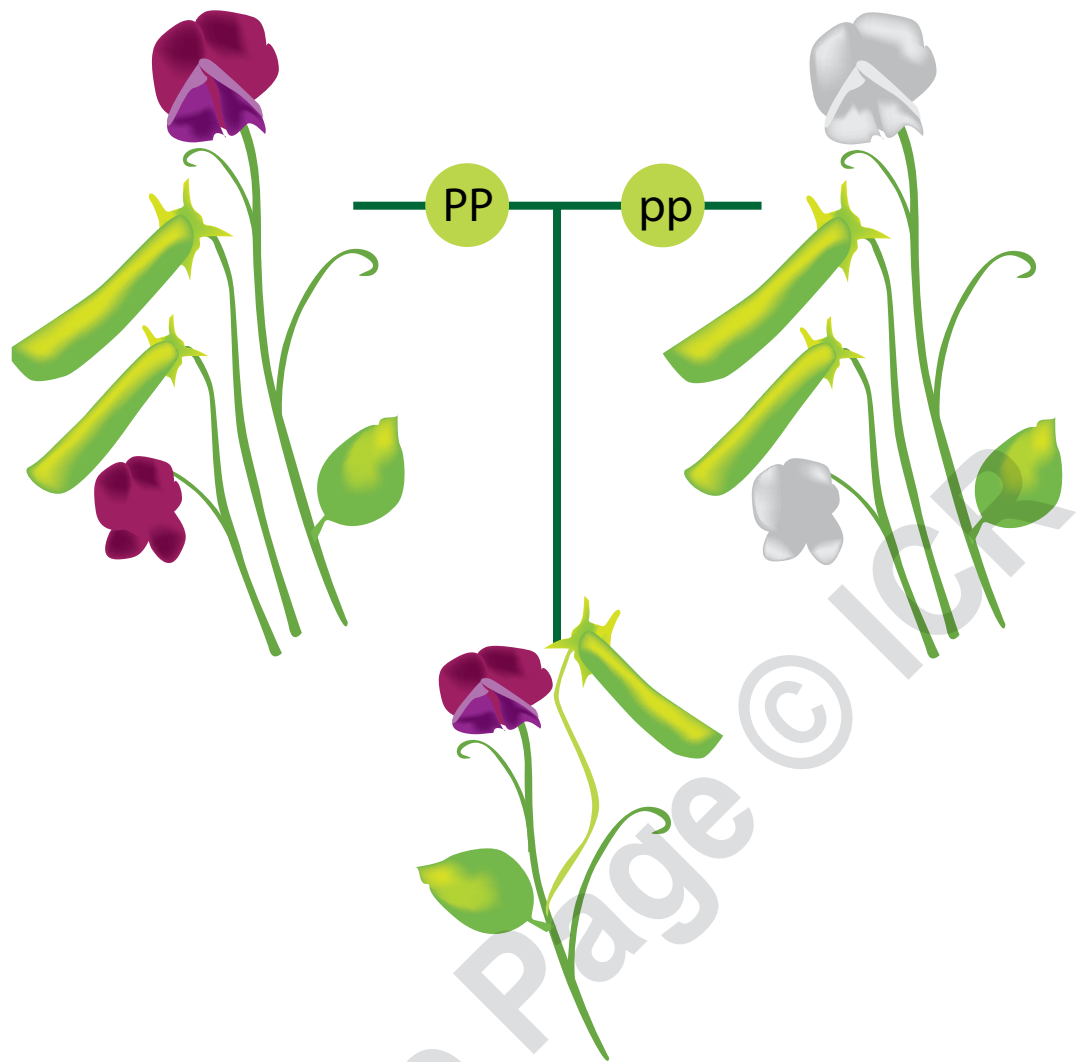
## BASIC MENDELIAN GENETICS

Our earliest understanding of how genetic inheritance works was made possible by an Austrian monk named Gregor Mendel. Mendel served in the St. Thomas Monastery in Brunn, Austria (now Brno, the Czech Republic), during the middle of the 19th century. Mendel raised pea plants (*Pisum sativum*) in the botanical garden at the monastery to verify what he termed were the “factors” that determined the inheritance pattern of individual plants. Mendel published his results in 1866 and died 18 years later, never knowing the impact his experiments would have on the science of genetics, the study of heredity or inheritance. Mendel’s work was cited 15 times after publication, but the importance of his work was not realized until 1900, when several prominent scientists of the time, including Hugo de Vries and Carl Correns, independently obtained the same results as Mendel in separate experiments. Because of his pioneering efforts, Gregor Mendel is frequently referred to as the “Father of Genetics.”



A brief look at Mendel’s pea plant experiments will provide a basic understanding of how individual traits are inherited and illustrate a few basic principles for human genetics. Mendel published the results of his pea plant experiments using several clearly distinguishable traits observed in these plants: seed (pea) color, pea shape, pod shape, pod color, flower position, and stem length. Our discussion of Mendel’s experiments will be limited to one trait—pea color.

Many important scientific discoveries have been made through the observation of simple events or patterns in nature. Mendel’s discovery of pea color inheritance is one example. He observed that pea plants in his garden produced two different colors of peas, yellow and green, which aroused his curiosity about how these characteristics were inherited from the parental plants. Mendel cross-pollinated a yellow pea plant with a green pea plant to find out what color would be produced in the offspring. To cross-pollinate the plants, Mendel took pollen containing the sperm from one pea plant variety (yellow peas) and fertilized the ova (eggs) of a second plant variety (green peas). All of the offspring from Mendel’s cross-pollination resulted in plants that produced only yellow peas, although they also inherited a gene for green peas (Figure 2). The offspring are called the F1 generation, an abbreviation for the first filial generation. Mendel theorized that the “factor,” which we now call a gene, for yellow peas was dominant over the “factor” for green peas. To test his theory, he crossed the F1 offspring—the plants that had a gene for yellow peas and one for green peas—with each other to observe what their offspring would look like. Mendel’s second cross,



the F2 (second filial) generation, produced three plants with yellow peas for every plant that had green peas (Figure 3).

The F2 generation of pea plants confirmed what Mendel had theorized from the first plant cross, leading to several genetic laws that also hold true for human genetics. Mendel showed that genes can have two variations (and we now know that some genes have even more). The pea plants have one gene for pea coloration, but two variations of that gene. Today, the variation of a particular gene is called an *allele*. Pea plants can have one allele for yellow peas and one allele for green peas. Mendel's pea plants also demonstrated that one trait can be dominant over another trait. In other words, the effect of the *dominant* allele is visible in the plant (yellow peas), while the *recessive* allele (green peas), although present with the dominant allele, is not seen in the appearance of the plant. If the yellow pea allele is inherited with a green pea allele, all the peas produced will be yellow.

The F1 generation appeared to show that only one allele is normally inherited from each parent. The F1 offspring all had yellow peas. However, this generation was able to produce offspring that had green peas, indicating that they not only received a yellow pea allele from one

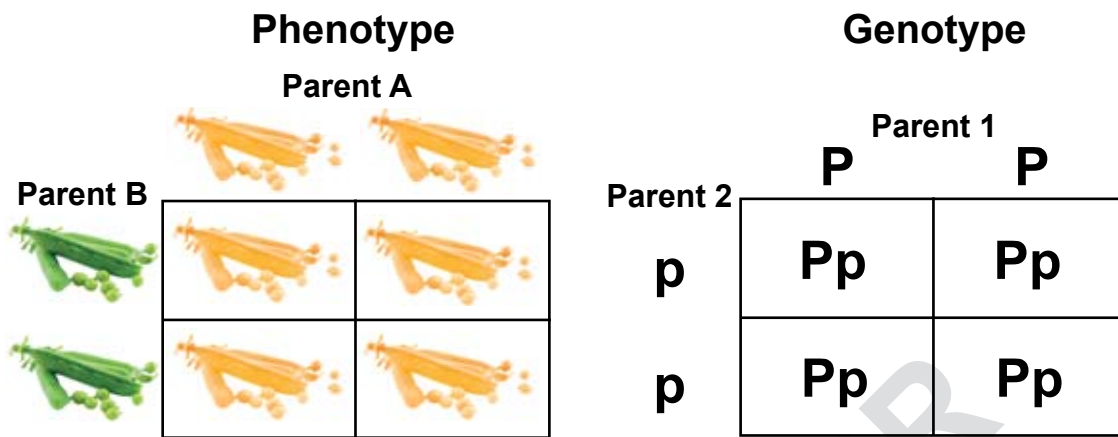


Figure 2: F1 generation

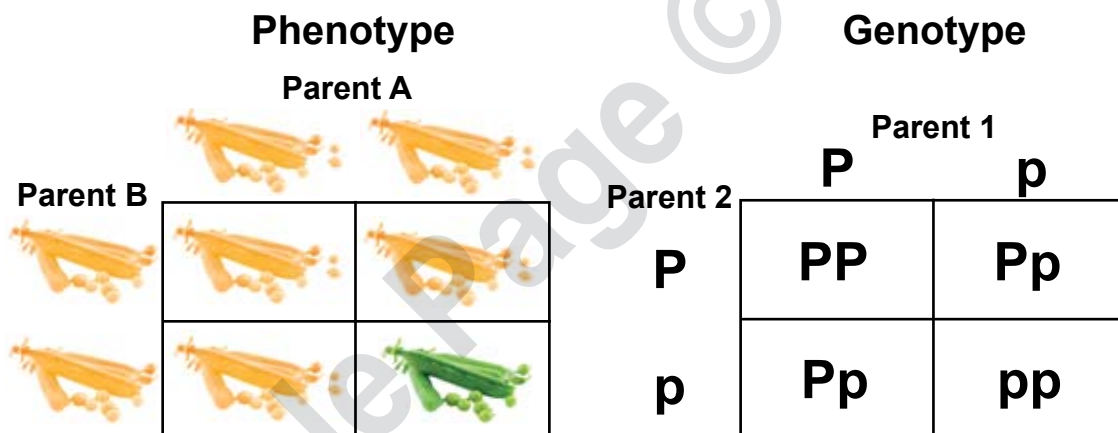


Figure 3: F2 generation

parent, but also received a green pea allele from the other. Remember that offspring inherit only one set of chromosomes from each parent. The inheritance of only one set of chromosomes, and consequently one set of genes, from each parent is due to a process called *segregation*. When sex cells (sperm and egg) are formed, chromosome pairs segregate so that each sex cell receives just one of each kind of chromosome. This makes it possible for the offspring to receive one set of chromosomes from each parent and become a combination of the genetic material of those parents. Because one set of chromosomes is inherited from each parent, only one of every gene, or more accurately allele, is inherited from each parent. Mendel's F1 pea plants received an allele for yellow peas and one for green peas from the two parental plants, indicating that the chromosomes in the parental strains successfully segregated into sex cells that recombined to become the F1 generation. The production of some plants with green peas in the F2 generation verified that the F1 plants were carriers of alleles for green peas.

Homozygous Dominant				Homozygous Recessive			
Parent 1				Parent 1			
Parent 2		E	E	Parent 2		e	e
E	EE	EE	EE	e	ee	ee	ee
E	EE	EE	EE	e	ee	ee	ee

Heterozygous			
Parent 1			
Parent 2		E	E
e	Ee	Ee	Ee
e	Ee	Ee	Ee

Figure 4: Typical Punnett squares showing monohybrid crosses

The results of Mendel's work can be visualized by making Punnett squares of a monohybrid cross for pea coloration (Figure 4). Punnett squares predict the probability of what offspring will look like and what their allelic combinations might be from a specific parental cross. A monohybrid cross is the genetic cross for just one trait. The results of a Punnett square are given as ratios, fractions, or a percentage of the total offspring. In the previous example, alleles for yellow peas were inherited from the male and alleles for green peas were inherited from the female. Each parent had two identical alleles for pea coloration, yellow in one and green in the other. When an individual has two identical alleles, they are *homozygous* for that trait. The parental plant with yellow peas had two alleles for yellow peas and the parental plant with green peas had two alleles for green peas. The reproductive cells, or gametes, from each parent receive only one allele to give to the offspring because of chromosome segregation. In this case, the sperm cells have one allele for yellow peas and the egg cells have one allele for green peas. When the sperm fertilizes the egg, the resulting offspring inherit one allele for yellow peas and one allele for green peas in the F1 generation. The F1 pea plants are all *heterozygous*, having two different alleles for one trait (pea color).

The F1 generation produces offspring that can have either yellow or green peas. The F1 male sperm cells can have one allele for yellow



peas or one for green. The same is true for the F1 female gamete. The egg cells can have one allele for yellow peas or one for green. When fertilization occurs, the F2 offspring can have a variety of alleles, depending on which sperm fertilized which egg. If a sperm with a yellow allele fertilizes an egg with a yellow allele, the offspring will have two alleles for yellow coloration and be homozygous. If a sperm with one yellow allele fertilizes an egg with a green allele, the offspring will have one allele of each color and be heterozygous.

Constructing a Punnett square shows several possible combinations of alleles for the offspring. To construct a Punnett square for a monohybrid cross, draw a box and divide it into four squares. Designate the male gametes (sperm) at the top of the box with “Y” for the yellow allele (dominant) over one column of squares and a lower case “y” for the green allele (recessive) over the other column of squares. The male contribution to the offspring will be written in the squares below each allele. The female gametes, one “Y” and one “y,” are written on the left side of the box with the female contribution to the offspring written in the squares to the right of each allele. The Punnett square gives three possible genotypes for the offspring in these ratios: a quarter of them YY, half Yy, and a quarter yy.

	<b>Y</b>	<b>y</b>
<b>Y</b>	<b>YY</b>	<b>Yy</b>
<b>y</b>	<b>Yy</b>	<b>yy</b>

A *genotype* is the “type of genes” or alleles that an organism has. The YY and Yy genotypes result in plants that have yellow peas, and the yy genotype results in plants that have green peas. The expression of the genes that determines what an organism looks like is its *phenotype*. The phenotype produced from the alleles YY and Yy is a plant with yellow peas, and plants with yy will have a green pea phenotype. Remember, the Punnett square only predicts what the ratios of the offspring should be. In this case, the predicted ratio of plants with yellow peas to plants with green peas is 3:1. Three-fourths of the plants should have yellow peas and one-fourth should have green peas. This is what Mendel observed in his results. His F2 pea plants produced three times as many plants with yellow peas as plants with green peas. Mendel confirmed his theories by observing the same results for other traits. Each time, his F2 crosses produced offspring that had three times as many dominant phenotypes as recessive phenotypes.

The discussion about yellow and green peas might suggest that two distinctly different alleles for pea coloration exist. This is not completely

Parents	Offspring Phenotype	Offspring Genotype	Offspring Ratios
2 homozygous dominant	100% show dominant trait	100% homozygous dominant	All are YY
2 homozygous recessive	100% show recessive trait	100% homozygous recessive	All are yy
1 homozygous recessive 1 homozygous dominant	100% show dominant trait	100% heterozygous	All 4 are Yy
1 homozygous dominant 1 heterozygous	100% show dominant trait	50% homozygous dominant 50% heterozygous	1/2 YY 1/2 Yy
2 heterozygous	75% show dominant trait 25% show recessive trait	25% homozygous dominant 25% homozygous recessive 50% heterozygous	1/4 YY 1/4 yy 1/2 Yy

**Table 1:** Possible combination of alleles

accurate. The location for the pea coloration gene is on chromosome 1 in pea plants. DNA sequence analysis in plants with yellow and green peas has identified one normal gene for yellow coloration, and a mutated form of this same gene responsible for green coloration. The name of this gene is the “stay green” (SGR) gene. A mutation in the green peas abolishes the activity of the SGR protein that plays a critical role in the breakdown of chlorophyll in peas.

Chlorophyll is a green pigment required for photosynthesis, a process that converts light energy into chemical energy. In normal yellow-colored peas, chlorophyll—which is not needed in peas that are covered in a pod and not exposed to strong light—is degraded, leaving the yellow pea coloration. The green peas retain their green coloration because the mutated gene is unable to produce a functional enzyme. In yellow peas, the enzyme leads to the breakdown of chlorophyll. If a pea plant inherits one of each allele for pea coloration, the peas will be yellow because the functional *SGR* allele will produce the enzymes necessary to break down chlorophyll. Consequently, the terms “dominant” and “recessive,” in this case, simply refer to a gene that produces a functional enzyme versus a gene that produces a non-functional enzyme.

The example of pea coloration appears to be typical for all the pea plant traits that Mendel investigated. Mendel also investigated the inheritance pattern for pea shape (wrinkled or smooth) and stem length (tall or short). He discovered that smooth peas are dominant over wrinkled peas in the same ratio as yellow and green peas. We now know that smooth peas are formed from the activity of an enzyme that



helps convert sugar to starch, while wrinkled peas result from a mutated form of this gene, producing an enzyme that does not convert sugar to starch. Mendel also discovered that tall pea plants were dominant over short plants. Tall pea plants result from a normal gene that produces a hormone necessary to assist in the growth of the plant, while a mutated gene does not assist in plant growth, resulting in short plants. This demonstrates that many (though not all) dominant and recessive traits are simply the effects of a functional gene versus a mutated gene in an individual. Mendel's pea plant work and our subsequent understanding of gene function clearly show that mutations are a major source of variation in pea plants and, as we shall see, in all organisms—including humans.