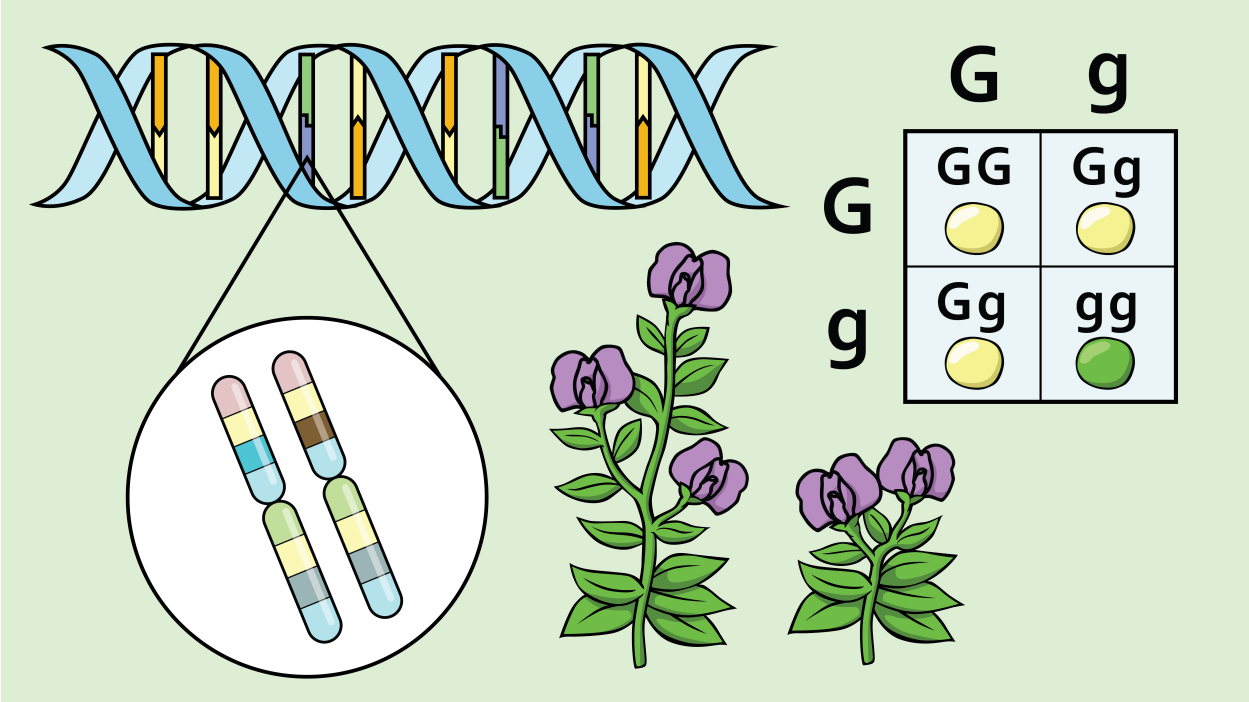


Introduction to Genetics

What makes each and every one of us unique? This important question can be answered by genetics! Genetics is the study of genes and heredity. In simpler language, genetics is the study of how traits get passed from parents to offspring. The subject of genetics can provide insight into evolutionary history, human health and disease transmission, criminal forensics, and our own ancestry.

Mendel's Work

The study of genetics began with the work of an Austrian monk named Gregor Mendel. Mendel was born in Hynčice, in the Austrian Empire in 1822. In 1843, he began studying to become a monk and moved to Brno, Austria to live and learn at the St. Thomas Monastery. The term monastery is used to describe a home for a group of monks all practicing religious principles. In these communities, there are houses and workplaces for monks and nuns who, in this era, chose to live a secluded lifestyle away from the general

public. This separation from society allowed for these religious persons to have a greater focus towards academic and spiritual pursuits. At the St. Thomas Monastery, Mendel had access to a library of academic texts, the teachings and expertise of fellow monks, and laboratory facilities. It was in the vegetable gardens of the monastery where he began conducting his experiments in 1845.

Mendel wanted to observe how traits were passed from parents to offspring and designed a set of experiments using pea plants to test these ideas. Pea plants were an excellent choice for his research as they have a wide variety of traits that Mendel could easily observe and track. These traits include: seed shape, seed color, flower color, pod shape, pod color, flower position, and stem length. Mendel could also self-pollinate the pea plants and observe the resulting patterns of characteristics.

	Seed Color	Seed Shape	Pod Color	Pod Shape	Flower Color	Flower Position	Plant Height
Dominant Traits	 Yellow	 Round	 Green	 Inflated (Full)	 Purple	 Axial	 Tall
Recessive Traits	 Green	 Wrinkled	 Yellow	 Constricted (Flat)	 White	 Terminal	 Short

Prior to Mendel and his pea plants, scientists held the belief that offspring inherited a blended combination of their parent's traits. However, this was a conclusion drawn from outdated research that was often completed quickly, resulting in inaccurate data and conclusions. Mendel however, spent eight years growing generations of pea plants, observing and recording the physical traits they possessed. As a result, he was able to identify trends and patterns that had yet to be discovered up until this point.

Mendel started his experiments by self-crossing plants that had differing physical characteristics. For example, he would cross a tall pea plant with a short one or a plant with green seeds with a yellow seed plant. This crossing was completed by Mendel taking the pollen from one mature pea plant and using it to fertilize another in order to achieve the crosses he desired. Once he crossed two plants he allowed their offspring to grow and once mature, he would then cross two of the offspring plants together, resulting in the next generation. He repeated this process, while simultaneously cataloging and reporting the physical appearance of each generation of offspring, until he had data from many generations to analyze.

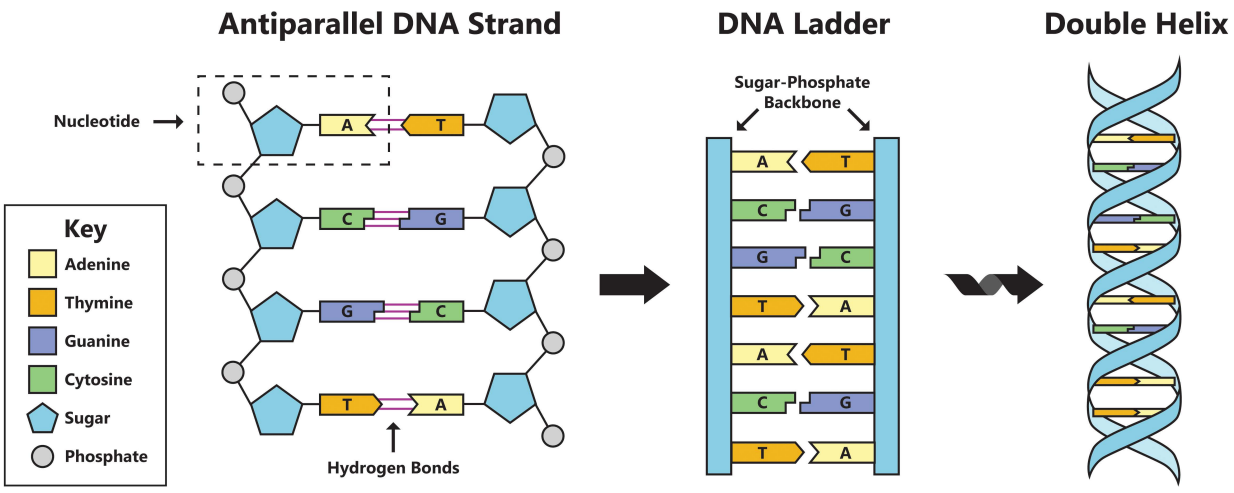
From this experiment, Mendel acknowledged that parental pea plants passed down independent, distinct traits, or “factors”, to their offspring rather than combined, or “blended” traits. For example, rather than the cross of a tall and short pea plant resulting in all medium height offspring, he noticed the offspring were either tall OR short. In his data, he began noticing statistical similarities within his tests. Specifically, in the ratios of certain physical traits observed.

Eventually, it was these patterns, or ratios, Mendel was able to summarize in his three Laws of Heredity. When Mendel first published his results in 1866, the scientific community overlooked the importance of his research and his paper went ignored until 16 years after his death.

Although overlooked at the time, Mendel’s pea plant experiments and his discovery of the Laws of Heredity is one of the most important scientific discoveries of all time. Mendel’s work earned him the nickname, “The Father of Modern Genetics”. Perhaps most impressive about his work, Mendel was able to unravel the mysteries of heredity without any knowledge of DNA and chromosomes which would later show a clear picture of how traits are passed from generation to generation.

DNA: The Hereditary Material

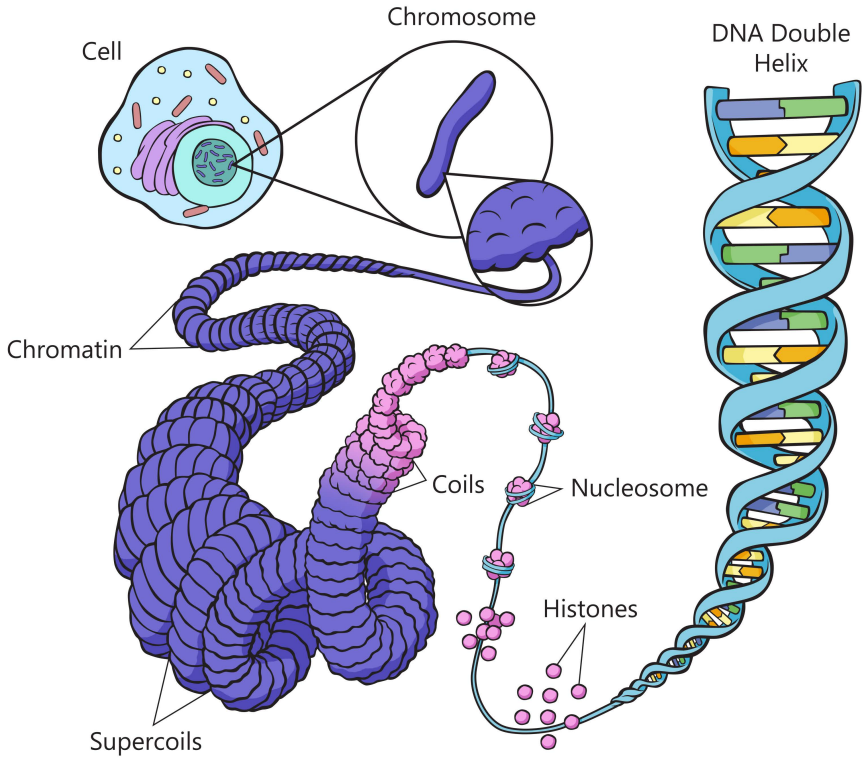
DNA, or deoxyribonucleicacid, is the biological molecule that contains an organism’s genetic information. DNA has a unique structure which serves its function of coding for our physical traits. First, a DNA molecule consists of two long chains that wind around each other to form a double helix. Connecting both sides of the helix to one another are nucleotide subunits, or nucleotides. One nucleotide is made up of a sugar, a phosphate, and one nitrogenous base. There are 4 different nitrogenous bases found in DNA: adenine, guanine, thymine, and cytosine. These nucleotides are held together in the center of the DNA molecule by hydrogen bonds. The backbones of DNA are made up of the sugars and phosphates which are connected using covalent bonds. To clarify, a DNA molecule can be compared to a ladder. The sides of the ladder would be the sugar and phosphate chains and the rungs of the ladder would be the nitrogenous base pairs. Finally, twisting the ladder would give the DNA the correct helical shape!



These nitrogenous bases exhibit a phenomenon called complementary base pairing where the nitrogenous bases always bond in specific pairs. In a normal DNA double helix, an adenine base will always be bonded to a thymine base. Additionally, a guanine base will always be bonded to a cytosine base. Complementary base pairing is one of the unique and most important characteristics of the DNA double helix. Complementary base pairing allows for processes that are integral to life itself, such as DNA transcription and translation, to occur. It is crucial to note that while the nitrogenous bases are bonded to their complements across the DNA helix, the order of nucleotides up and down the DNA helix varies. It is the variation in this linear sequence of bases that allows for each individual's unique physical traits.

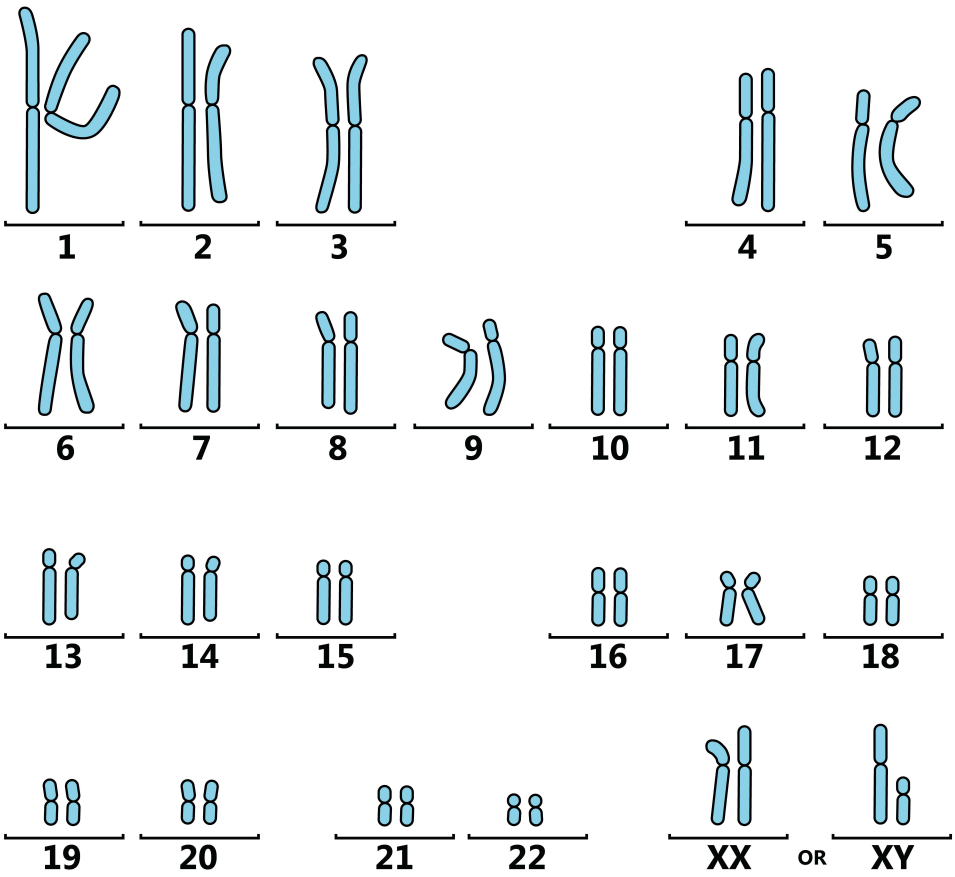
On average, one strand of human DNA can reach lengths up to 5 feet long! Imagine trying to fit a 5 foot long strand inside of a cell-it seems an impossible task! In order to fit inside the nucleus of a cell, DNA must first be condensed. In order to condense, the DNA strand is first wrapped around proteins called histones. Together, the histone and the DNA wrapped around it make up a complex called a nucleosome. Next, nucleosomes further condense by folding together to create chromatin fiber. Chromatin fiber then coils together in tight loops called supercoils. The condensed chromatin fiber forms chromosomes and chromosomes arethe final condensed form of DNA. The majority of the time,

DNA stays in this condensed chromosomal form. However, there are points when DNA must decondense within a cell's life. For example, during cellular division, or mitosis, the DNA within the cell's nucleus must be copied. This requires the DNA to decondense and unwind in order for the sequence of nitrogenous bases to be read and copied to form a new DNA strand.



Molecular inheritance is dependent on the structure of chromosomes. During reproduction, chromosomes are passed from parents to their offspring. Humans have 46 chromosomes in the nucleus of every somatic cell, which are arranged in 23 pairs. In each chromosome pair, one chromosome is passed down from the mother while the other is from the father.

Scientists categorize these chromosomes by their physical size, and number them; the largest pair of chromosomes is labeled Chromosome 1, the next largest Chromosome 2, and so on. One pair of chromosomes, the X and Y chromosomes, are sex chromosomes, which, as the name implies, determine a person's sex. If a person has two X chromosomes, they are genetically female, while a person with one X chromosome and one Y chromosome is genetically male.

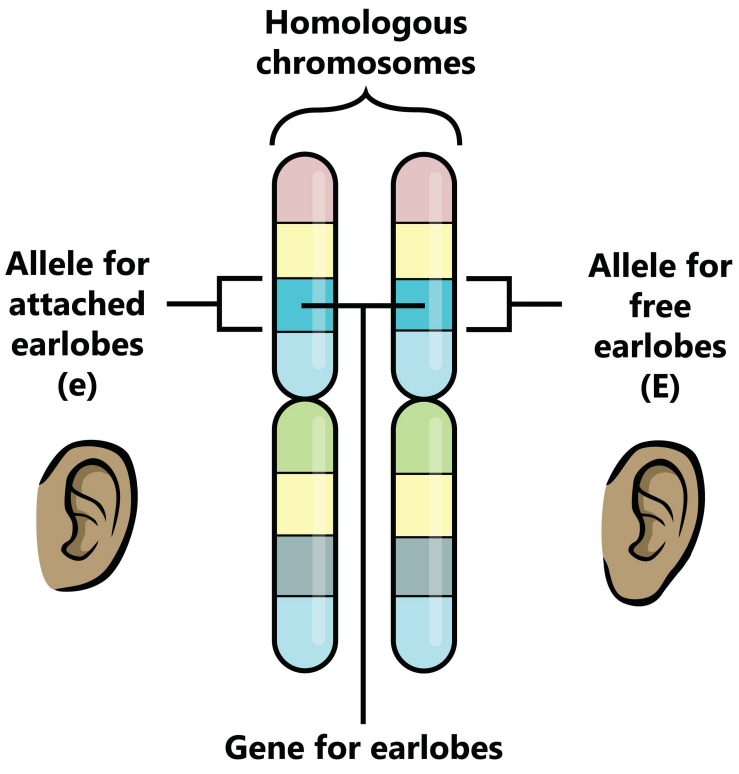


Remember, chromosomes are simply condensed strands of DNA. Specific sections of DNA strands hold specific hereditary information. These specific sections of DNA are called genes. Genes are the physical sequence of DNA base pairs that code for the production of specific proteins in cells. It is these proteins, rather than the DNA itself, that create an individual's physical appearance. Some examples of genes include those that code for widow's peak, hitchhiker's thumb, or even the ability to roll your tongue!

DNA provides a set of instructions that is used to tell the body how to create the proteins it needs. In broad terms, the information stored in DNA is translated into RNA, which then is read by cell organelles called ribosomes. These ribosomes use the information contained in the RNA molecule to produce proteins. Ultimately, it is the production of these proteins that give organisms their unique appearance. Overall, it is important to remember that DNA itself does not directly build proteins. Rather, DNA holds the genetic information that is used by ribosomes to build proteins needed for the body.

Genes are found at specific locations, or loci, on specific chromosomes. These loci are universal for all organisms in a species. For example, the genes that encode for height are located in the same places on the same chromosomes for all humans. In a way, gene loci can be compared to our addresses. Just as we live at our unique address, genes 'live' in one spot on a chromosome. Another example, a gene called CDH11 is always located on the long arm of chromosome 16 in humans. CDH11 codes for proteins that can give the physical appearance of a widow's peak in the hairline.

The constant nature of gene loci is what allowed scientists to map the entire human genome in 2003! A genome is the complete set of genetic material in an organism. Scientists have also created maps of other important animals that are commonly used in research such as mice and worms.



Genes are very complex. In the same gene locant, there can be different variations of the gene called alleles. For example, earlobe attachment is a physical trait controlled by a single gene with two alleles. This gene provides instructions to the body to produce either earlobes that are attached to the sides of the head or earlobes that hang freely and are not attached to the sides of the head. In this example, one "version", or allele, codes for attached earlobes while the other allele codes for detached earlobes. The combination of alleles a person has for their earlobe gene, determines which type of earlobes they have.

Understanding Alleles

Not all alleles have equal power when it comes to determining the expression of a certain trait. For any two genetic alleles, one will be the dominant allele and the other will be the recessive allele. The dominant allele will be expressed in the phenotype, or physical appearance, of the offspring, while the recessive will not. Recessive alleles will not be expressed in appearance unless there are no dominant alleles present. When denoting dominant versus recessive alleles in Punnett Squares, dominant alleles are capitalized while recessive alleles are left lowercase.

Genetic crosses can be used to determine how an allele gets passed down from parents to offspring, just as Mendel did with his pea plants. Crosses are purposeful matings between organisms used to observe what the offspring phenotype will be. For example, in the cross of two pea plants, there is a gene that determines the plant's seed color. In pea plants, the seed color gene has two alleles, green and yellow. The yellow allele is dominant over the green allele. To represent the seed color gene, the letter G is used. To denote the dominant yellow allele, the letter is capitalized, G. The recessive green allele is represented by a lowercase g.

Genotype: **Gg**

Phenotype: 

As discussed earlier, for each gene, a person inherits one copy, or allele, from their mother and one from their father. As a result, there are different combinations of alleles that can be passed down to offspring. Returning to the seed color gene example, let's say a pea plant inherits one dominant allele for seed color (G) from the maternal pea plant and one recessive allele (g) from the paternal plant. The offspring then would have a genotype, or genetic composition, of Gg. Since there is a dominant allele present in the genotype, the offspring plant would produce seeds which are yellow in color. The yellow seeds produced are the physical expression of the plant's genotype. Remember that the physical expression of genetic information in an organism is called a phenotype.

There are special names that are given to specific combinations of alleles in a genotype. A genotype that has two copies of the same allele, for example: GG or gg, is called homozygous. Alternatively, a genotype that has two different alleles, for example: Gg, is called heterozygous. It is important to note that only when an organism has a homozygous recessive genotype, will the recessive phenotype be expressed.

Overall, it is the inheritance of different allele combinations that give organisms different appearances!

Punnett Squares

Thanks to the work of Gregor Mendel, it became clear that genetic traits were passed down in a predictable manner. However, it was not until 1912 that a test was developed to predict potential genotypes and phenotypes based on a hypothetical cross of parental genes.

Reginald Punnett was an English geneticist who studied zoology at Cambridge University. During his time at Cambridge, Punnett was doing research studying nematode worms. It was while he was doing this research that he met a man who changed the course of his career.

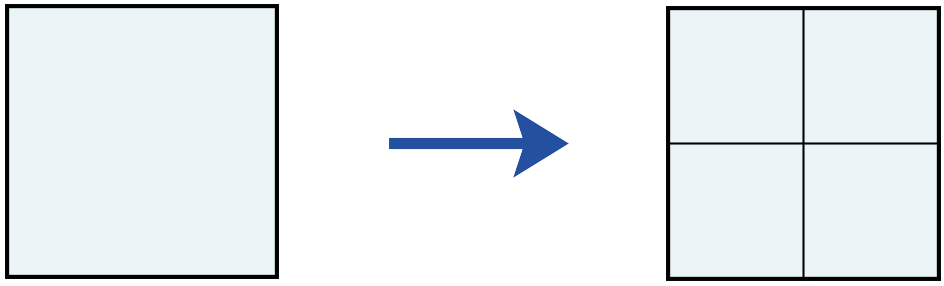
William Bateson, a British biologist, was studying evolution. Bateson learned of Gregor Mendel's work and became fascinated by it, and he himself began breeding plants and animals looking for heritable traits. Due to Bateson and his dedication to Mendel's ideas, he spread the information published by Mendel across Britain and his work reached more people than ever before. One of these people was Reginald Punnett.

With both men finding interest in Mendelian genetics, they began to collaborate with the common purpose of expanding Mendel's ideas. They became focused on determining a graphical way to represent the number and variety of genotypic and phenotypic combinations that can result from a genetic cross. Together, they eventually figured out the solution and called their discovery Punnett squares!

Setting Up the Punnett Square

Punnett squares work by crossing parental genotypes. A monohybrid cross is the simplest version of a Punnett square. A monohybrid cross looks at one specific gene and predicts the possible genotypic and phenotypic combinations that can occur in an offspring. Read the steps below to see how to set up a monohybrid Punnett square. Watch a video showing the process of Setting up a Punnett Square here.

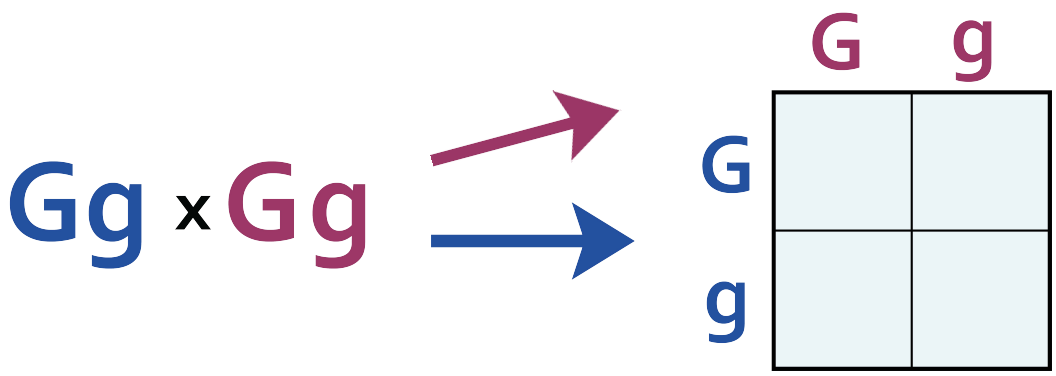
Step 1: Draw the Punnett square. Draw one large square and then draw a plus sign within the box so there are now four equal sized squares. The starting Punnett square should look like a window!



Step 2: Determine the parental genotypes. For this example, the maternal plant has a genotype of Gg and the paternal plant also has a genotype of Gg. To represent the crossing of these two parental plants, we write their genotypes separated by an "x" and place it above the Punnett square. For this example, it would read: Gg X Gg.

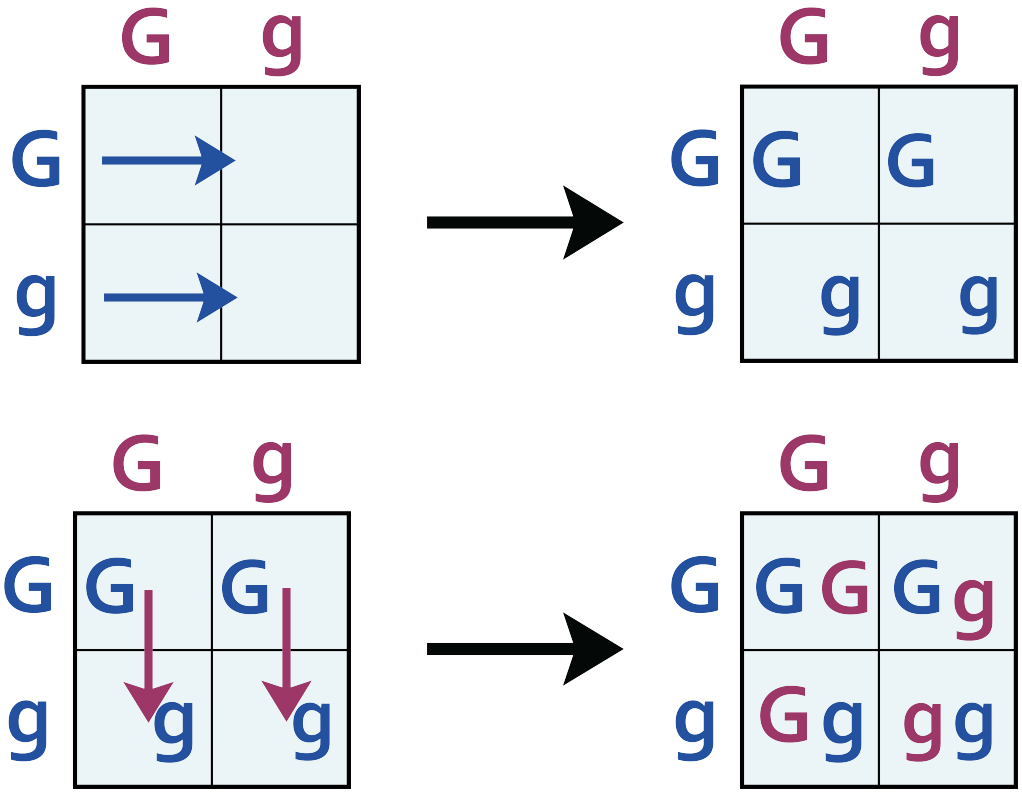
Gg x Gg

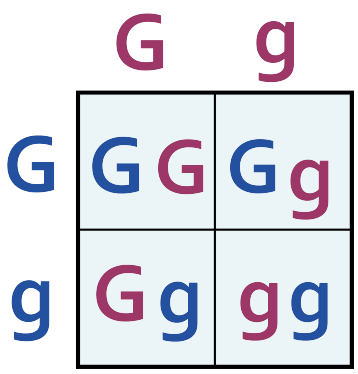
Step 3: Separate the parental genotypes. For the paternal genotype, the first allele will be placed alongside the top left box and the second allele will be placed alongside the bottom left box. For the maternal genotype, the first allele in the genotype will be placed above the top left box and the second allele will be placed above the top right box. In this example, one G is placed above the top left box and one g above the topright box. One G is placed alongside the top left box, and one g alongside the bottom left box.



Completing the Punnett Square

Step 4: Complete the Punnett square by filling in the middle boxes. To fill in these boxes, the alleles on the top and side must be carried down and across to the middle boxes. For the alleles on top, those alleles get passed down the columns they are above. A G should be written in both the top left and bottom left boxes. Next, a g should be written in both the top right and bottom right boxes. For the alleles alongside, those alleles get carried across the rows in the same manner. For example, a G should be written in both the top left and top right boxes. A g should be written in both the bottom left and right boxes. Moving the individual alleles down the columns and across the rows results in two letters, or alleles, inside each box.





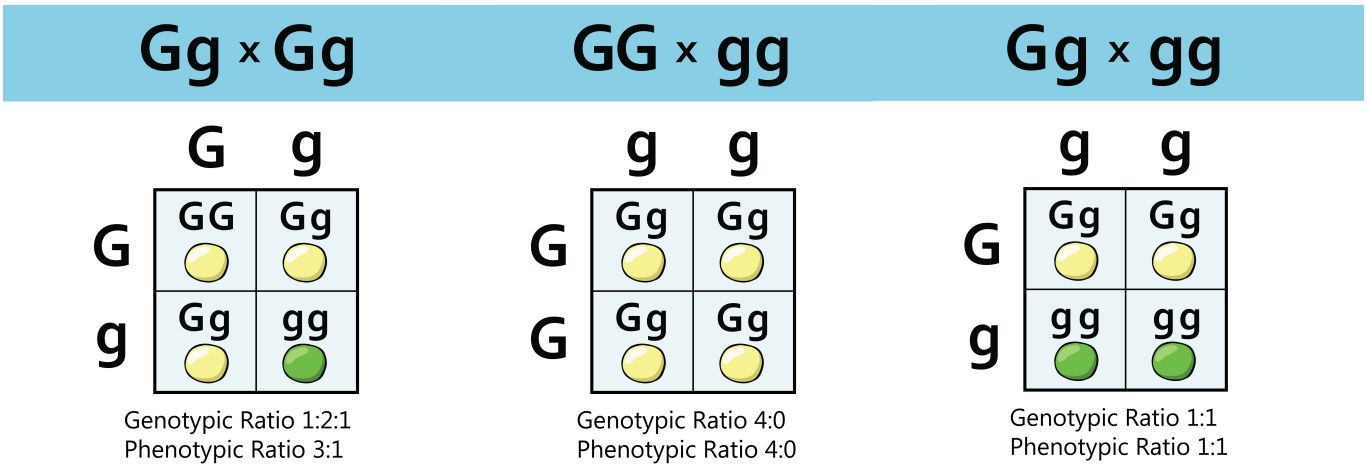
GG:1, Gg:2, gg:1

Step 5: The combination of alleles inside each box of the Punnett square represent the possible genotypes of an offspring from the parental cross. To interpret the results of the Punnett square, the percent chance of an offspring inheriting a specific genotype must be calculated. Using the completed Punnett square above, first write down each possible genotype found inside the boxes. Next, count the number of times each genotype appears. This would result in: . This tally of genotypes is called the genotypic ratio.

Step 6: Using the information from the genotypic ratio, the phenotypic ratio can also be calculated. To determine the phenotypic ratio (the number of each different possible phenotype) simply look at each genotype and determine what phenotype would result from it. For example, the genotype GG would result in a yellow seed color. The genotype Gg would also result in a yellow seed color. Finally, the genotype gg would result in a green seed color. To calculate the phenotypic ratio, simply count the number of times a genotype would result in a particular phenotype. In this example, 3 of the genotypes in the Punnett square would result in a yellow phenotype and only 1 genotype in the Punnett square would result in a green phenotype. This would make the phenotypic ratio 3:1.

The genotypic and phenotypic ratios in monohybrid crosses are often predictable depending on what parental genotypes are being crossed. In other words, specific crosses always result in the same ratios. This can help us determine if we completed a Punnett square correctly, as well as to determine parental genotype if they are unknown. When two heterozygous parents are crossed, there is always a genotypic ratio of 1:2:1 and a phenotypic ratio of 3:1, as seen in the example above. Another cross that always provides the same ratios is a cross between two homozygous parents. This cross always results in a genotypic and phenotypic ratio of 4:0. If the parental phenotypes of a cross are unknown, but there is a 1:2:1 genotypic ratio observed, it can be

concluded that both parents are heterozygous for the trait. This method for determining parental genotypes can be used for any of these known genotypic or phenotypic ratios.



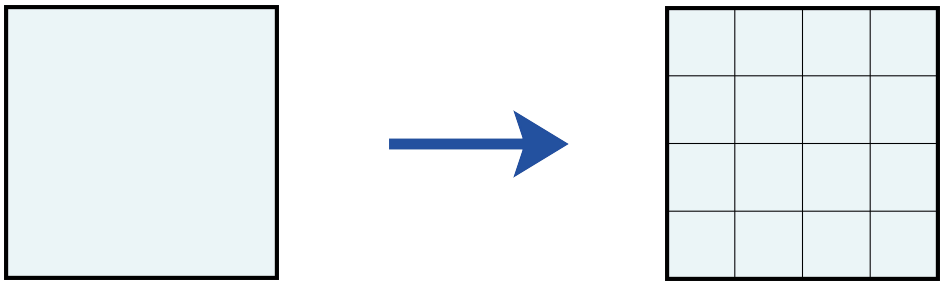
Dihybrid Crosses

Punnett squares can also be used to determine the probability of the genotypes and phenotypes of two traits inherited by an offspring. These crosses, called dihybrid crosses, are very similar to monohybrid crosses. However, the major difference being that they predict the inheritance of two genes rather than one singular gene. For example, rather than just analyzing the probability of inheriting a certain seed color in pea plants, a dihybrid cross can determine the probability of inheriting a certain seed color and seed shape.

While these crosses are completed in a similar manner to monohybrid crosses, there are important differences in the process that must be accounted for. To complete a dihybrid cross, use these steps below! Watch a video showing the process of Setting up a Dihybrid Cross Punnett Square here.

Setting Up A Dihybrid Cross

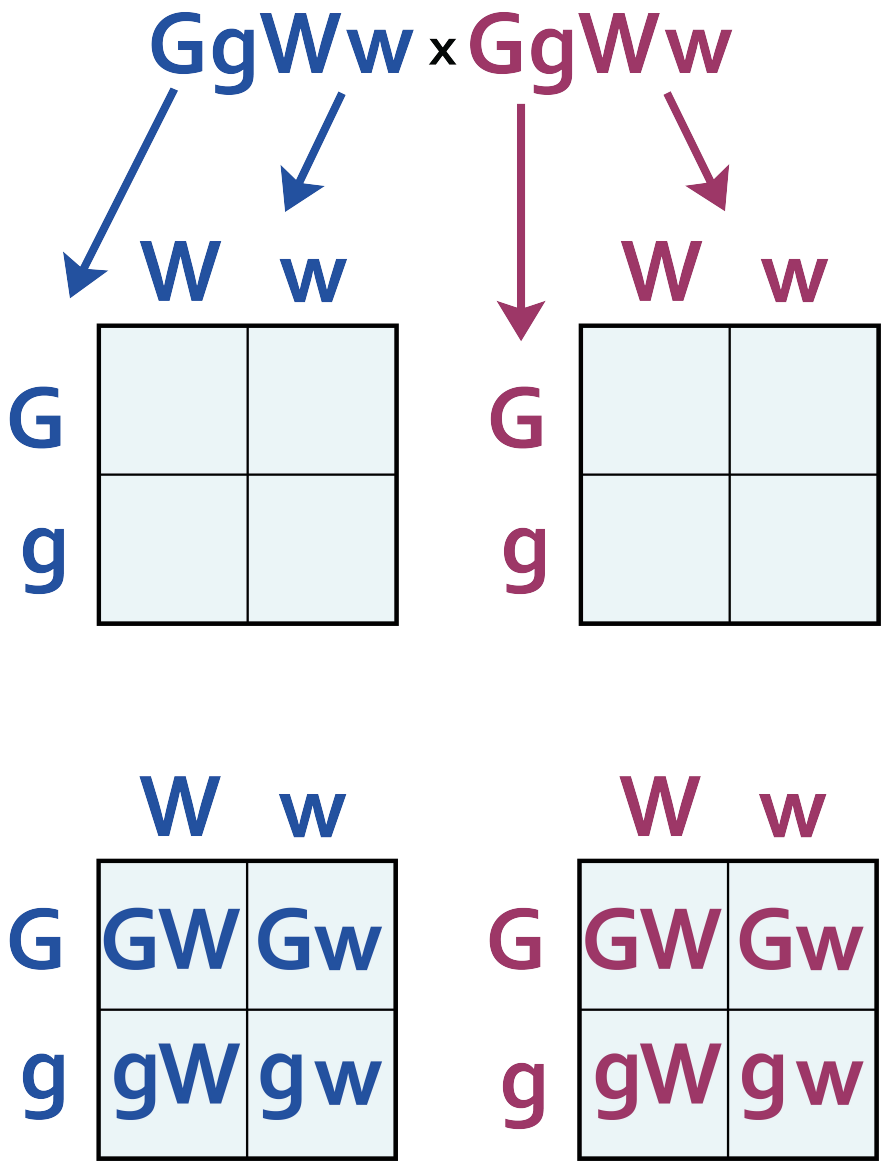
Step 1: Draw the Punnett square. In dihybrid crosses, since two traits are being analyzed, a greater variety of offspring genotypes are possible. For these crosses, draw one large square. Then, split the square vertically into 4 equal columns and 4 equal rows. This should result in a 4x4 grid, with 16 squares total in the middle.



Step 2: Determine the parental genotypes. Since dihybrid crosses analyze two genes, both must be written in the parental genotypes. In this example, we will again analyze seed color, represented by the letter G and also seed shape, denoted with the letter W. The seed color gene has two alleles, yellow (dominant, G) and green (recessive, g). The seed shape gene also has two alleles, one for round seeds (dominant, W) and wrinkled seeds (recessive, w). In this example, both parents are heterozygous for both traits meaning that they have 1 dominant and 1 recessive allele for each individual gene. When completing a dihybrid cross, these allele combinations are written together as one genotype. As such, both parental genotypes would be, GgWw. Because, in this example, both parents have the same genotype, the cross is written as GgWw X GgWw.

GgWw x GgWw

Step 3: Determine allele combinations. Because there are two genes each with two alleles, 4 possible allele combinations can be made from each parent. To determine these allele combinations, two monohybrid Punnett squares can be completed using the parental genotypes. For example, take one parental genotype and split it up into a cross between both genes in their genotype. In this case, the maternal genotype is GgWw so a monohybrid cross of Gg X Ww will determine all of the possible allele combinations the mother could pass to the offspring. The middle boxes then show the genotypes GW, Gw, gW and gw. This should be done for both parental genotypes. In this example, since the paternal genotype is also GgWw, it will also result in the same allele combinations of: GG, Gw, gW, gw.



	GW	Gw	gW	gw
GW				
Gw				
gW				
gw				

Step 4: List parental allele combinations above the Punnett square boxes. Across the top, place each of the 4 maternal allele combinations with one combination above each column. Do the same down the side, placing each of the paternal allele combinations with one combination beside each row.

Completing a Dihybrid Cross

Step 5: Fill in the middle boxes. To complete the dihybrid cross, alleles are moved down each column and across each row, just as in a monohybrid cross. However, unlike in monohybrid crosses, because two alleles from two separate genes are being represented, each box will contain 4 total alleles, when the dihybrid cross is complete.

	GW	Gw	gW	gw
GW	GWGW	GWGw	GWgW	GWgw
Gw	GwGW	GwGw	GwgW	Gwgw
gW	gWGW	gWGw	gWgW	gWgw
gw	gwGW	gwGw	gwgW	gwgw

Interpreting a Dihybrid Cross

Step 6: Determine the phenotypic and genotypic ratios of inheritance for the offspring. To do this, first write down the different genotypes found inside the boxes. Next, count the number of times each genotype appears. In this example, there is 1 GGWW, 2 GGWw, 1 GGww, and so on. The phenotypic ratio can be calculated by interpreting the genotypes in each box and

determining the phenotypes these allele combinations code for. Then, tally the amount of times each phenotype appears. In dihybrid crosses of two heterozygous parents there is a predictable genotypic and phenotypic ratio that is always consistent. The phenotypes of the offspring should appear in a 9:3:3:1 ratio as there are 9 yellow round pea plants, 3 yellow wrinkled pea plants, 3 green round pea plants, and 1 green wrinkled pea plant offspring. The genotypic ratio for this cross is always equal to 1:2:1:2:4:2:1:2:1. If the cross is not between two heterozygous parents, both ratios can be calculated by counting up all the different genotypes and phenotypes, the same process used in the monohybrid cross.

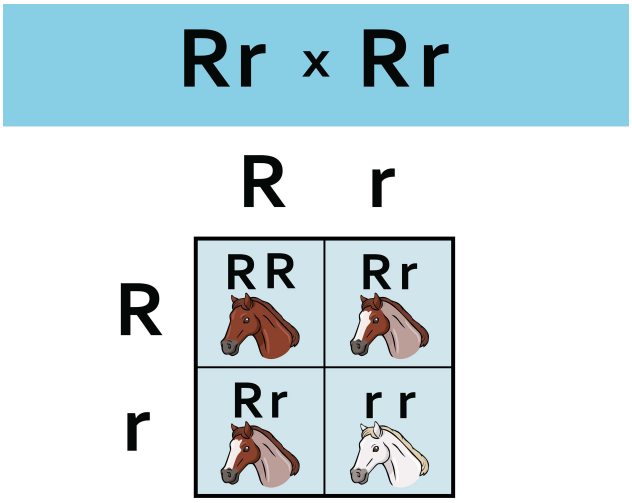
Non-Mendelian Genetics

During Gregor Mendel's pea plant experiments, he worked with plants whose traits were encoded by genes that had only two alleles with specific dominance patterns. In other words, for each of the traits he tested, the dominance he observed always showed one phenotype completely overriding the other. Today, we know that genetic inheritance is not always this simple. Commonly, alleles have more complex dominance relationships than what Mendel observed. We also know today that many genes have more than two allele variations. Overall, traits that do not fit the descriptions of simple dominance or have two alleles per gene, are classified as Non-Mendelian traits. There are three main patterns of Non-Mendelian genetics: codominance, incomplete dominance, and multiple alleles.

Codominance

Codominance is observed when both alleles for a trait are equally expressed in an organism's phenotype. In other words, for heterozygous organisms both the dominant and recessive allele will show independently in the phenotype. For example, roan horses show this pattern of inheritance in their hair colors. Roan horses are generally composed of two hair colors. The gene for hair color in this breed of horse is composed of two alleles, R for red coloring and r for white coloring. However, when a horse has a heterozygous genotype (Rr), they are not completely red colored, as Mendel would have suggested. Because the gene for horse hair color is codominant, a heterozygous horse will grow both red hairs and

white hairs in its coat. Across of two heterozygous roan horses is shown here.



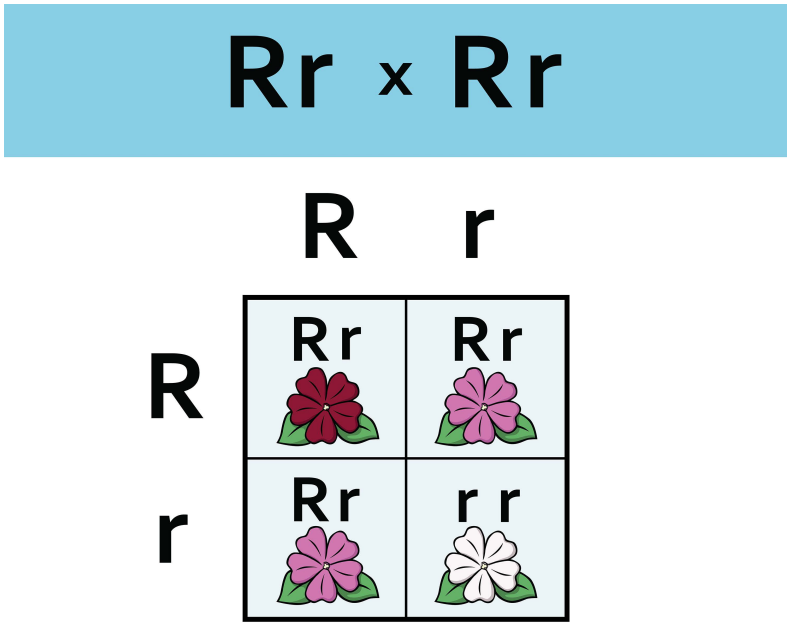
Genotypic Ratio 1:2:1
Phenotypic Ratio 1:2:1

Crosses between two heterozygous individuals with codominant genes always have a predictable ratio similar to the trends observed above. For this type of cross, a genotypic ratio of 1:2:1 will always be observed. This is because there will be 1 homozygous dominant offspring that has solid red color and 1 homozygous recessive offspring will be a solid white color. Then, there will always be heterozygous offspring who exhibit both the red and white colors in their coat. This results in both a genotypic and phenotypic ratio of 1:2:1.

Incomplete Dominance

In instances of incomplete dominance, the dominant allele of a gene is not fully dominant which results in an intermediate phenotype to be expressed in heterozygotes. In these cases, the heterozygous phenotype can be seen as an intermediate, or a “blend”, of the homozygous dominant phenotype and homozygous recessive phenotype. In this example shown below, a gene coding for flower color exhibits incomplete dominance. In this gene, the allele for red flowers is dominant over white flowers. However, since the trait is controlled by incomplete dominance, the heterozygous offspring are a “blended” phenotype. In this case,

the intermediate between red and white is pink. This Punnett square between two heterozygotes (Rr) results in one red flower that is homozygous dominant and one white flower that is homozygous recessive. It also shows two offspring that have pink heterozygous flowers which is the result of the incomplete dominance pattern as pink is the “blended” phenotype.



Genotypic Ratio 1:2:1
Phenotypic Ratio 1:2:1

Incomplete dominance also results in predictable genotypic and phenotypic ratios when two heterozygous individuals are crossed. In these crosses, there will be 1 homozygous dominant offspring, RR, which results in a red flower and 1 homozygous recessive flower, rr, which will be a white flower. Then, 2 Rr genotypes will result in the blended pink phenotype. So, like codominant traits, genes that exhibit incomplete dominance will also have predicted genotypic and phenotypic ratios.

Multiple Alleles

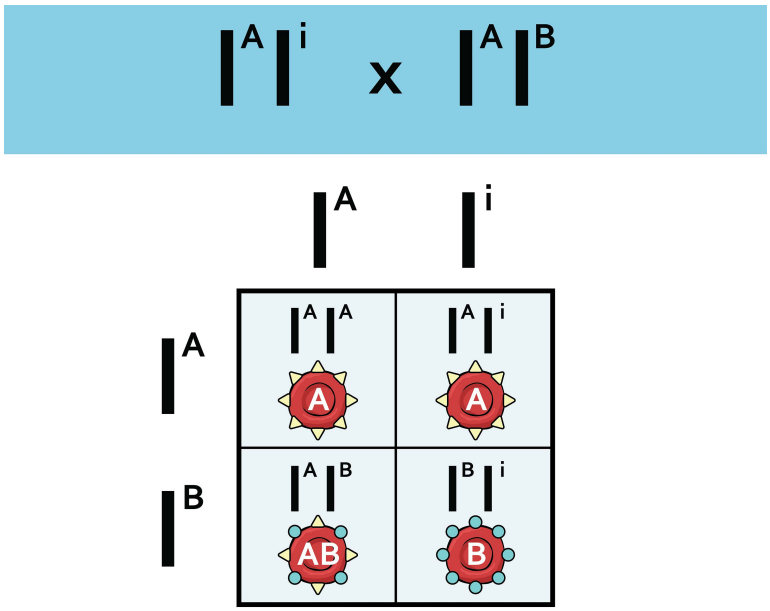
Another Non-Mendelian inheritance pattern occurs when there are more than just two alleles for one gene. When traits are controlled by multiple alleles, there are also different dominance

patterns that can simultaneously occur. For example, the gene coding for human blood types is controlled by three alleles.

Humans can exhibit three different types of red blood cells: Type A, Type B, and Type O. These three types are coded by three different alleles: I^A, $I^{B,}$ and Ii respectively. Type A red blood cells contain A antigens on the cell's surface while Type B red blood cells have B antigens on their surface. Type O red blood cells do not possess any antigens on their surface. Depending on what alleles someone inherits, and therefore the resulting antigens which are present on their red blood cells, determines the person's blood type: A, B, AB, or O!

Between these three different alleles, both codominance and simple dominance are observed. Codominance can be seen when alleles I^A and I^B are inherited together which results in blood Type AB. AB red blood cells have both type A antigens and type B antigens present on their surface. Simple dominance can also be seen as I^A and I^B are both dominant to I^i. For example, someone with the genotypes I^AI^i or I^BI^i will have Type A antigens or Type B antigens on their red blood cells as the recessive i allele, coding for no antigens, does not get expressed in the phenotype. Only if someone inherits two copies of the Ii allele will they have Type O blood which contains no surface antigens.

In the Punnett square below, a cross between a person heterozygous for Type A blood (I^AI^i) is crossed with someone who has Type AB blood (I^AI^B). The potential offspring can then have the genotypes I^AI^A, I^AI^i,I^AI^B, or I^BI^i. Within this example, each resulting offspring has a different genotype so the genotypic ratio would be 1:1:1:1. The phenotypic ratio would be 1:2:1. This is because the genotype I^AI^B results in AB blood, both I^AI^A and I^AI^i result in Type A blood and I^BI^i results in Type B blood. Unlike the other examples of non-Mendelian traits discussed so far, there is not one specific genotypic or phenotypic ratio observed. This is because the dominance patterns are different for every gene with multiple alleles!



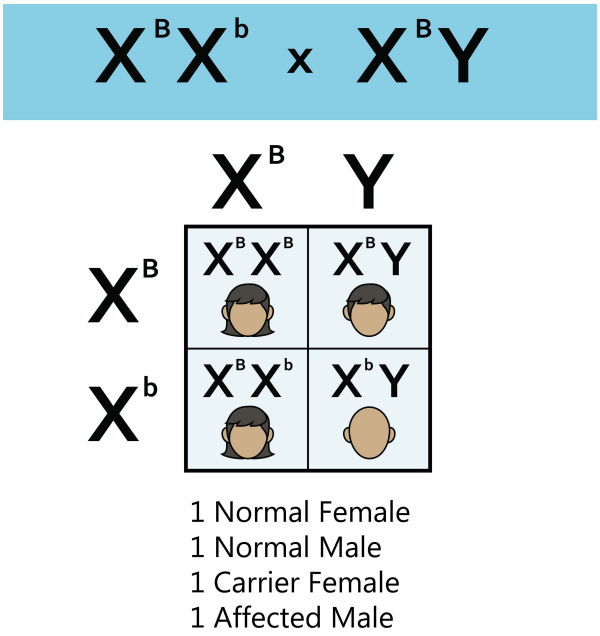
Genotypic Ratio 1:1:1:1
Phenotypic Ratio 1:2:1

Sex-Linked Traits

Genes that are present on one of the two sex chromosomes, either X or Y, are written differently than traits located on autosomal chromosomes. For example, one gene located specifically on the X chromosome is a gene that codes for baldness. The gene for baldness, represented by the letter B, has two alleles. The dominant allele, B, codes for normal hair growth while the recessive allele b, codes for baldness. When writing the genotypes for sex-linked traits, it is crucial to denote the specific chromosome these alleles are on, which means designating the sex of each person in the cross. As stated before, when a person inherits two X chromosomes, they are genetically female and if someone inherits one X and one Y, they are genetically male. Since the gene for baldness is only on the X chromosome, this must be taken into account.

When writing the genotypes of sex-linked traits, the sex of the person is written first then the corresponding alleles are added as superscripts on the sex chromosomes. For example, the cross below shows the offspring between a female carrier for baldness and a male with hair. First, we will start by writing the maternal genotype. Since she is genetically female, XX is

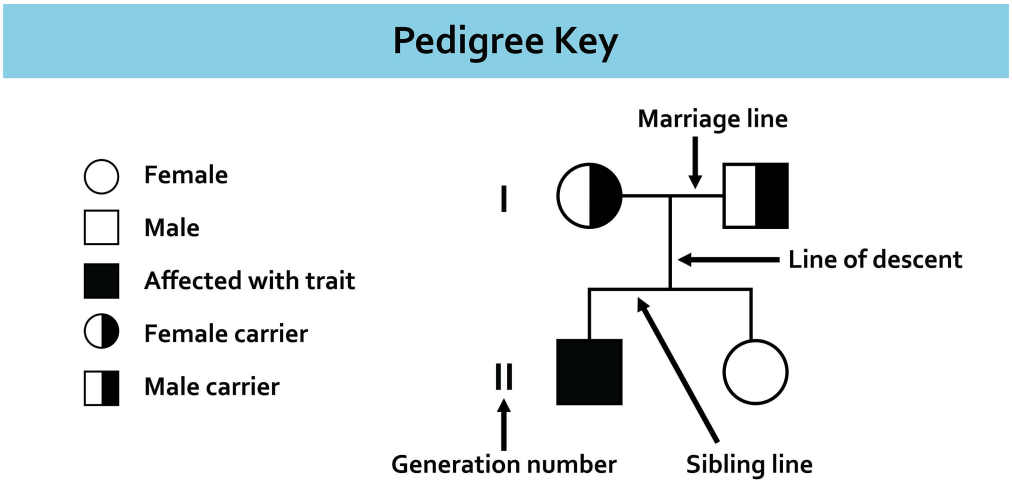
written to show she has two X chromosomes. Then, since she is a carrier for baldness, she has one recessive allele and one dominant allele on her chromosomes. They are written as superscripts as follows: X^BX^b. For the paternal genotype, he has the sex chromosomes XY as he is genetically male. Then, since he has hair, he will carry a dominant allele on his X chromosome. His genotype will be X^BY. Remember, baldness is an X linked trait so, there should be no alleles labeled on the Y chromosome because the gene for baldness is not located on the Y chromosome.



As a result of this cross, the offspring will either be male with hair, male who is bald, a normal female, or a carrier female. Overall, recessive alleles on X linked traits are much more likely to be presented in males rather than females. This is because unlike females who have two X chromosomes, males only have one. This means if they inherit a recessive allele on their X chromosome there is no chance a dominant allele could mask the recessive phenotype. Females with two X chromosomes must inherit two recessive alleles in order to present the recessive phenotype which is unlikely in comparison to a male inheriting only one recessive allele.

Pedigrees

In order to see the genetic history of a family, a pedigree chart can be made. Pedigrees track the genetic inheritance of one gene across multiple generations. Most commonly, these charts track the inheritance of a genetic disease through a family lineage. These charts also help to determine the type of inheritance pattern taking place for the specific gene being analyzed. When reading pedigrees, there is a standardized notation for representing different people.



For example, those who are genetically female are represented by a circle while those who are genetically male are shown as a square. When a pair is married, a line is drawn horizontally connecting the two and if they have children, a vertical line is drawn down from the marriage line to create a new generation. This line is called a line of descent and it is from this line that offspring are drawn on. In the example located in the key above, there are two people in the first generation who are married and they have two children, one boy and one girl.

In order to represent the passing of a specific trait, individuals in a pedigree will be shaded in to represent their genotypes and phenotypes. For example, individuals who are not shaded at all are considered “normal” because they do not reflect the specific trait being examined in their phenotype. If an individual is completely shaded in, this means that they do show the phenotype. Some individuals are carriers which means although they do not show the

phenotype, they possess at least one allele which codes for the phenotype in their genotype, but that recessive allele is masked by adominant allele. So, even though the individual themselves do not show the specific phenotype, they could potentially pass the recessive allele to their offspring.

Autosomal Dominant

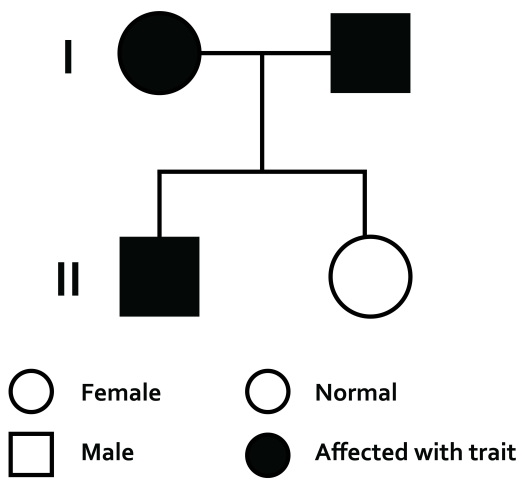
Traits that are considered autosomal dominant are those that are coded by genes on autosomes, or non-sex chromosomes, in which a single copy of a dominant allele will cause expression of the trait in the phenotype. One example of a trait controlled by an autosomal dominant inheritance pattern is Huntington's disease. Huntington's disease causes early breakdown of nerve cells in the brain. In this case, the dominant allele of this gene codes for Huntington's while the recessive allele codes for no disease to be expressed. While a pedigree chart cannot concretely determine the inheritance a trait has, certain patterns can help us narrow down the possibilities and get a good approximation of what the inheritance type is.

For example, the pedigree below shows the inheritance pattern for an autosomal dominant trait. In this chart, we see two parents who are both affected with the trait. These parents then have two children, one affected and one not affected. When determining if a trait is autosomal dominant, look for these patterns within the pedigrees:

- The trait affects both sexes
- Every affected individual has a parent who is affected
- The trait is expressed in every generation
- An affected individual has ½ of their offspring who are affected

The occurrence of these events is not a 100% guarantee however, if this pattern is followed, there is a high probability that the trait is autosomal dominant.

Autosomal Dominant



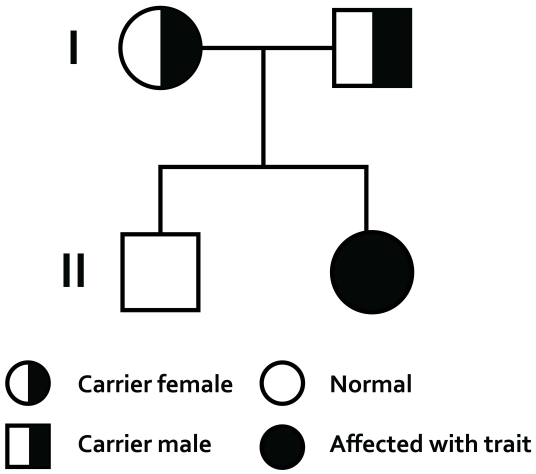
Autosomal Recessive

Autosomal recessive traits are those in which it is the recessive allele that codes for expression of a given phenotype rather than the dominant allele. Similarly to above, these genes are located on autosomal chromosomes, or any chromosome that is not an X or Y. These traits are only expressed in an individual's phenotype when two recessive alleles are inherited together. Cystic Fibrosis (CF) is a common example of an autosomal recessive disease. CF causes a thick mucus to build up in the lungs. Like all autosomal recessive traits, a person can only develop CF if they have a homozygous recessive genotype.

In the example below, the pedigree chart shows the occurrence of a trait that is controlled by an autosomal recessive gene. Here, two individuals, both carriers, have two children. One child is affected by the trait while the other is not affected. Overall, here are some common patterns observed in pedigrees of autosomal recessive traits:

- The trait affects both sexes
- Often, affected individuals have parents who are unaffected but are carriers
- The trait skips generations
- When two carriers are crossed, about ¼ of their offspring will be affected

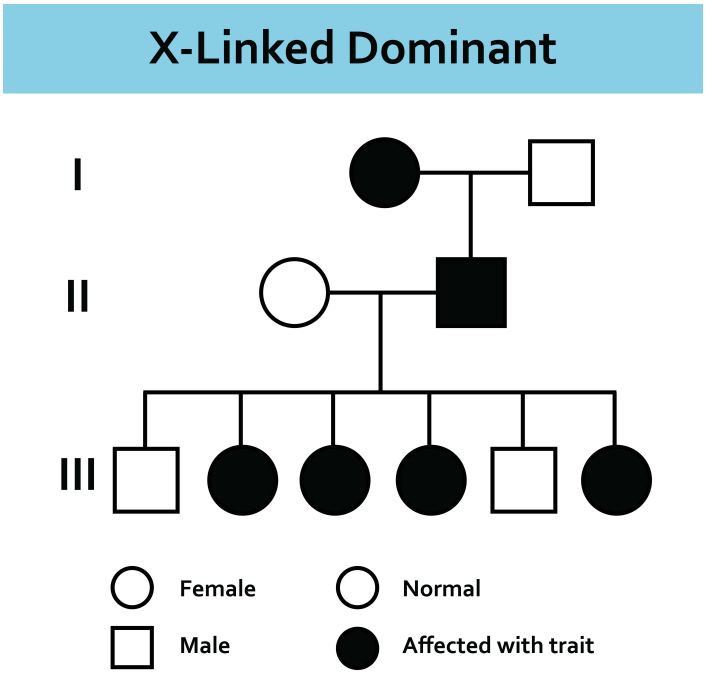
Autosomal Recessive



Remember, if this pattern is represented in a pedigree chart there is a high likelihood that the gene is controlled by an autosomal recessive inheritance type, but it is not guaranteed.

X-Linked Dominant

Unlike the previous two examples, traits that are X-linked are found only on the X sex chromosome rather than on an autosome. In general, sex linked genes can be found on both the X chromosome and the Y chromosome however, since the X chromosome is much larger and contains more genes, X-linked traits are more common than Y-linked traits. X-linked traits can follow either a dominant or recessive inheritance pattern. One specific example of an X-linked trait is the inheritance of a disorder called Rett Syndrome. This disease is controlled by a gene located on the X chromosome and labeled with the letter R. Rett Syndrome causes memory deficiencies primarily in females and is present in an individual's phenotype when they have at least one dominant allele in their genotype: X^RX^R or X^RX^r. Rarely, a man will inherit Rett Syndrome, with the genotype X^RY. Sex linked traits, specifically X-linked dominant traits, have specific pedigree patterns that indicate this pattern of inheritance.



In this pedigree, we see the inheritance of an X-linked dominant trait through three generations. In order to determine if a pedigree shows an X-linked dominant inheritance pattern, look for this major indicator:

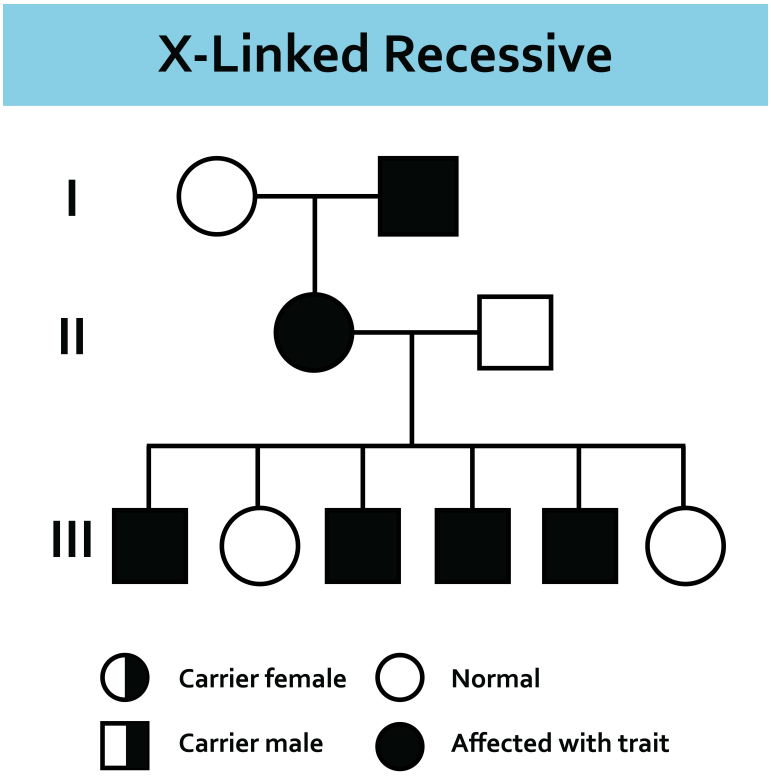
- If a male is affected, so are all of his daughters and his mother
- The trait appears more frequently in females than males

This pattern occurs because if a male expresses an X-linked trait, he has the dominant allele for the trait on his singular X-chromosome. Then, because he only has one X chromosome to pass to his daughters, all the daughters will receive a dominant allele on at least one of their sex chromosomes which leads to expression of the trait.

X-Linked Recessive

Like X-linked dominant traits, X-linked recessive traits are coded for by genes located on the X chromosome. However, X-linked recessive traits are only expressed by the inheritance of a recessive allele rather than a dominant allele. So, for a female to express an X-linked recessive

trait in their phenotype, they must inherit two X chromosomes, both with recessive alleles. A male only needs one copy of a recessive allele to express the trait, as they only have one X chromosome. Red-green color blindness is one trait that is coded for by a gene with X-linked recessive inheritance. Color blindness, specifically red-green color blindness, is a vision deficiency in which individuals have a difficult time distinguishing between the colors red, green, and yellow. The pedigree chart below shows the inheritance pattern for an X-linked recessive trait.



Like the last pedigree chart, this one shows expression of a trait across three generations; however, in this pedigree, patterns specific to X-linked recessive inheritance can be observed, such as:

If a female is affected, so are all of her sons and her father

Trait appears more frequently in males than in females

If a woman is affected with an X-linked recessive trait, both of her X chromosomes must carry the recessive allele. So, her children then will all get a recessive allele X chromosome from her. Since the males will only have that singular X chromosome from their mother, they are guaranteed to express the given trait in their phenotype. Female offspring however, may not express the trait if they inherit an X chromosome with a dominant from their father to mask the recessive allele passed from their mother.

Conclusion

From what started as one experiment by Gregor Mendel, the field of genetics has turned into an expansive, critical field of science! Mendel collected phenotypic observations of inherited traits by crossing different pea plants over multiple generations. However, it is important to remember, Mendel did not have the expansive knowledge about DNA that we have today. Now, we know that DNA codes for the production of specific proteins which influences our physical traits. Similarly, we know now that specific traits are passed from parents to offspring through chromosomes. We also took a closer look at genes throughout the reading as we learned that simple genes have two alleles that can be either dominant or recessive. Depending on which set of alleles you inherit, that will determine the phenotype of the offspring. To determine the results of a genetic cross, or the potential genotypes and phenotypes of offspring, a Punnett square can be made. Another important note to remember is that genetic behaviors are complex and therefore, not all genes exhibit two alleles with simple dominance patterns. Non-Mendelian patterns such as codominance, incomplete dominance, multiple alleles, and sex-linked traits all display greater complexity than what Mendel initially observed. To visualize all of these genetic patterns, specifically across a family line, pedigree charts can be made. These charts show the passing of a trait, most commonly genetic diseases, across multiple generations. If unknown, these charts can give insight on what inheritance pattern the trait displays. Principles and techniques such as Mendelian genetics, Non-Mendelian genetics, Punnett squares, and pedigrees are all fundamental pieces to the study still today.