Extending Mendelian Genetics

Mendel's use of pea plants ensured that he would be able to follow easily predictable dominant and recessive patterns of inheritance. We now know that most phenotype expression is much more complex. Very few human traits follow the dominant and recessive relationship, or "Mendelian" rules of inheritance.

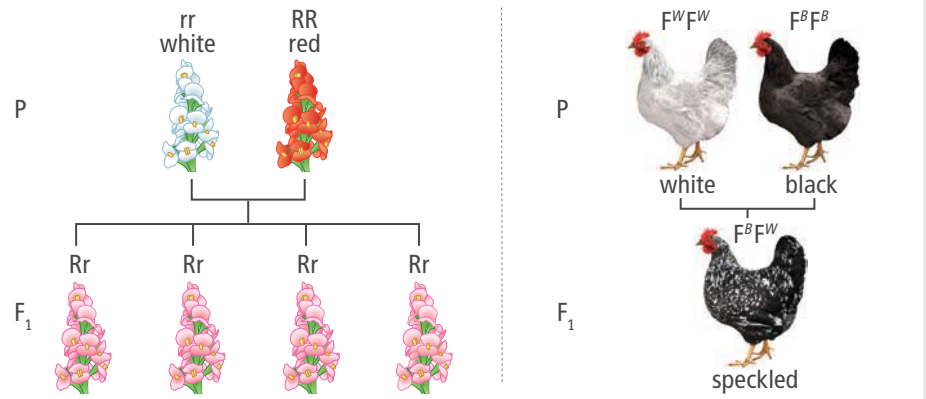
Complex Patterns of Inheritance

Mendel's basic theory of heredity was correct, but his research could not have explained all of the continuous variations for many traits. Many traits result from alleles with a range of dominance, rather than a strict dominant and recessive relationship.

Incomplete Dominance and Codominance

Sometimes alleles show incomplete dominance, in which a heterozygous phenotype is somewhere between the two homozygous phenotypes. This yields a blended result. For example, a cross between a snapdragon with white flowers and a snapdragon with red flowers results in offspring with pink flowers. Sometimes, both alleles of a gene are equally expressed and appear in the phenotype. These alleles show codominance, and both traits are fully and separately expressed. For example, when a certain breed of white-feathered chicken is crossed with the black-feathered phenotype of the same breed, their offspring have feathers that are speckled black and white.

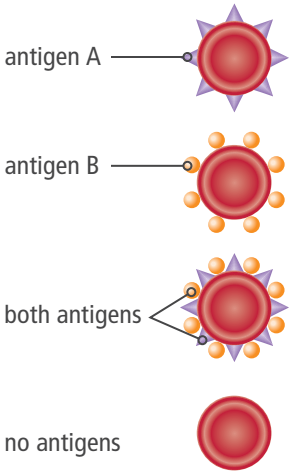
FIGURE 11: Incomplete Dominance and Codominance



Multiple Alleles

In some cases there are more than two alleles possible in a population. Human blood type is an example of multiple alleles. The three alleles are called I^A, I^B, and *i*. Both I^A and I^B result in a protein, called an antigen, on the surface of red blood cells. Allele *i* is recessive and does not result in an antigen. Someone with a genotype of I^Ai will have type A blood, and someone with a genotype of I^Bi will have type B blood. I^A and I^B alleles are also codominant. That means someone with a genotype of I^AI^B will have type AB blood. People with an *ii* genotype have red blood cells without an antigen, and they have type O blood.

FIGURE 12: Human blood type is controlled by multiple alleles, two of which are codominant.



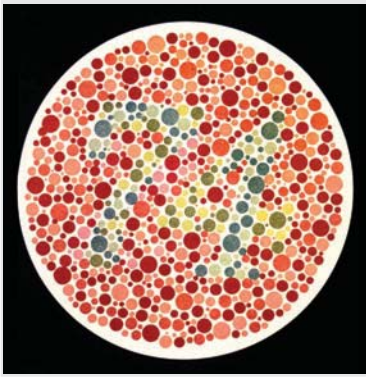
Sex-Linked Traits

Recall that humans have 23 pairs of chromosomes and that the last pair is referred to as sex chromosomes. These chromosomes—X and Y—contain different genes, which make a unique pattern of inheritance. Many of the genes seen on the X chromosome do not have corresponding genes on the Y chromosome, simply because the Y chromosome is so much smaller. Males only have one copy of the Y chromosome, so any recessive gene on a Y chromosome will be expressed. Any recessive gene on an X chromosome also will be expressed in males, because there is no second X chromosome to mask the recessive allele's expression. The genes located on an X or Y chromosome are referred to as sex-linked genes. Red-green colorblindness is an example of a trait caused by a sex-linked gene that occurs more often in males.

Females have double the number of genes located on an X chromosome, but they do not need double the number of their associated proteins. A process known as X inactivation solves this dilemma. Only one X chromosome is active, while the other is silenced or has very few active genes. X inactivation results in more balanced gene expression between males and females.

Explain The gene for red-green colorblindness is located on the X chromosome. Does the mother or father pass the gene for colorblindness to sons? Explain your answer.

FIGURE 13: People with red-green colorblindness cannot distinguish between the colors red and green.



Polygenic Traits

In contrast to the traits studied by Mendel, most plant and animal traits are actually the product of multiple genes. Very few traits in humans are controlled by a single gene. Your height is an example of a polygenic trait, in which multiple genes contribute to the overall phenotype observed. The height genes you inherit from your mother and father accumulate, and the final height that you are likely to reach is due in part to the cumulative effect of these genes. Scientists have discovered over 600 genes that affect height. These complex traits show a continuous range of phenotypes from very short to very tall. Polygenic traits often show a bell-shaped curve when graphed. Many people fall around the average, and very few show one extreme or the other.

Epistasis

Another polygenic trait is fur color in mice and in other mammals. In mice, at least five different genes interact to produce the phenotype. Two genes give the mouse its general color. One gene affects the shading of the color, and another gene determines whether the mouse will have spots. But the fifth gene involved in mouse fur color can overshadow all of the others. In cases such as this, one gene, called an epistatic gene, can interfere with the expression of other genes. Genes that modify the expression of another gene are said to show epistasis.

In albinism, a single epistatic gene interferes with the expression of other genes. Albinism is characterized by a lack of pigment in skin, hair, and eyes. A mouse that is homozygous for the alleles that prevent the coloration of fur will be white, regardless of the phenotypes that would normally come from the other four genes. A person with two recessive alleles for albinism will have very light skin, hair, and eyes, regardless of the other genes he or she has inherited.

FIGURE 14: Albinism in this wallaby is caused by an epistatic gene that blocks the production of pigment.

Cause and Effect

FIGURE 15: Several different genes interact to produce the range of human eye colors.

Genes for Eye Color

Another example of epistasis occurs in human eye color. Two genes thought to be responsible for eye color are called *OCA2* and *HERC2*, both located on chromosome 15. The *OCA2* gene codes for a protein involved in storing pigment in the iris. This protein helps cells store melanin, the pigment that affects eye coloration. More of the protein leads to darker eyes, which may appear brown. Less of the protein leads to lighter eyes, which may appear blue. The expression of the *OCA2* gene, however, can be turned on or off by a mutation in another gene. This gene, called *HERC2*, can reduce the expression of *OCA2*, leading to less melanin being stored in the iris and resulting in blue eyes. Several other genes are known to contribute to eye color, including those that lead to green eyes.

Analyze Draw a simple diagram to model the scenario described in each question. Use your diagram as evidence for your explanations.

1. A child inherits a functional copy of the *OCA2* gene from his mother but a mutated version of this gene from his father. Predict his eye color. Explain your answer.
2. Another child inherits two functional copies of the *OCA2* gene but also inherits two copies of the *HERC2* gene that suppresses the expression of the *OCA2* genes. What would you predict about the color of this child's eyes? Explain your answer.

Genes and the Environment

The environment also interacts with genes and affects their expression. Environmental influences, such as temperature, diet, light, and even pH, all play a role in the expression of countless traits in plants and animals. For example, the sex of sea turtles depends both on genes and on their environment. Female turtles make nests on beaches and bury their eggs in the sand. Eggs that mature in warmer temperatures develop into female turtles. Eggs that mature in cooler temperatures develop into male turtles.

Genes and the environment also interact to determine certain human traits. For example, a person's height is determined by genes, but environmental factors, such as lifestyle and nutrition also affect height. Studies of identical twins have shown that the environment during early development can have long-lasting effects. One twin might get more nutrients than the other because of its position in the mother's uterus. This difference can result in height and size differences that last throughout the twins' lives. Also, twins raised in environments with different diets and health care often differ in height as well as other physical traits.

Gather Evidence How might patterns of inheritance influence taste preferences? What environmental factors might affect this trait?