

## Mendel and the Gene Idea

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Gregor Mendel performed his ground-breaking genetics experiments on pea plants. He removed the male stamens from flowers, leaving the female carpel intact. He then collected pollen from other flowers and transferred the pollen to the modified flowers. In this way, he was assured of which plants mated with which other plants. He performed seven sets of experiments, with each experiment concerned with one characteristic. In each experiment, he mated two true-breeding parents (the P generation) having different traits from one another. Such a mating is called a monohybrid cross. The resulting offspring made up the F<sub>1</sub> generation. He then interbred members of the F<sub>1</sub> generation to produce the F<sub>2</sub> generation.

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Mendel crossed a true-breeding, purple flowered plant with a true-breeding, white flowered plant, and he repeated the experiment hundreds of times. Every one of the F<sub>1</sub> offspring was purple flowered, and the flowers were just as purple as the true-breeding purple flowered parent. When he performed hundreds of crosses between F<sub>1</sub> individuals to create the F<sub>2</sub> generation, he obtained both purple flowered individuals and white flowered individuals, with a ratio of 3 purple plants to 1 white plant. These results indicated that the purple trait dominates over the white trait, so purple is dominant and white is recessive.

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A characteristic (like flower color) can have multiple values (like purple and white), called traits. For the control of a characteristic to operate according to Mendelian inheritance, two requirements must be fulfilled:

- The characteristic must have exactly two traits.
- One of the traits (the dominant trait) must completely dominate over the other (the recessive trait).

In each of Mendel's experiments, a monohybrid cross was performed between a parent with one trait for the characteristic under study and one parent with the other trait of that same characteristic. Each time, the F<sub>1</sub> generation was 100% dominant, and the F<sub>2</sub> was made up of 75% dominant individuals and 25% recessive individuals (an F<sub>2</sub> ratio of 3:1).

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A diploid cell contains two of each kind of chromosome. The two make up a homologous pair. Each chromosome contains a long sequence of DNA nucleotides. A gene is a shorter sequence that is part of that long sequence. Each chromosome contains many genes, and each gene corresponds to a characteristic. The position on a chromosome at which a gene is found is called its locus. A diploid cell, having two of each kind of chromosome, contains two of each kind of gene (one on each homolog). A gene can have multiple versions. The different versions of a gene are called alleles. Whereas genes correspond to characteristics, alleles correspond to traits. For a given gene, a diploid organism can have both alleles be the same, or the two alleles can be different. An organism having two identical alleles is homozygous. If both of the alleles are the dominant version, the organism is homozygous dominant. If both alleles are the recessive version, the organism is homozygous recessive. If the two alleles are different (one is dominant, and the other is recessive), the organism is heterozygous.

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For a given characteristic, an organism's observable trait is called the phenotype. A genotype is a symbolic representation of the organism's alleles for the gene that controls that characteristic. A genotype for a single gene in a diploid organism contains two symbols (one for each allele). A genotype for a haploid gamete contains just one symbol. Fertilization can be represented by a Punnett square, in which the haploid genotypes of the gametes of the two parents are listed to the side of the square (for one parent) and above the square (for the other parent). Boxes within the square contain combinations of the gametic genotypes. These combinations are obtained by matching up row with column. The genotypes in the boxes are therefore diploid, and the boxes represent zygotes. The genotypes explain Mendel's results. Each true-breeding (homozygous) parent can produce only one kind of gamete, so the only possible  $F_1$  genotype is heterozygous. Because of dominance, all the  $F_1$  individuals expressed the dominant phenotype. The Punnett square predicts  $F_2$  offspring in the ratio of three dominant individuals for each recessive individual, in agreement with Mendel's results.

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The  $F_2$  phenotypic ratio (3:1) for a monohybrid cross involving Mendelian inheritance is different from the genotypic ratio (1:2:1), because there are two ways to express the dominant phenotype (homozygous dominant and heterozygous).

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A test cross is performed to determine the genotype of an individual with the dominant phenotype. A dominant individual can be either homozygous or heterozygous. Each of the two possibilities corresponds to its own Punnett square when that dominant individual is crossed with a recessive individual. The actual cross is performed, and whichever Punnett square agrees with the results for the real offspring reveals the genotype of the dominant individual in question.

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A dihybrid cross is much like a monohybrid cross, except that two different characteristics are tracked at the same time. If there were no independent assortment, then the alleles for the two genes from one parent would always remain together. This would result in offspring that are entirely like one or the other parent. Independent assortment does occur, however, so it's possible for offspring to be either of the parental type (like one of the parents) or of the recombinant type (with one parent's trait for one characteristic and with the other parent's trait for the other characteristic).

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The same ratios that a Punnett square predicts can also be computed by multiplying values for individual probabilities.

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Non-Mendelian inheritance involves exceptions to Mendelian inheritance, so the normal Mendelian ratios are skewed. One exception is called incomplete dominance. One allele is dominant over the other, but not completely. As a result, a heterozygote, which has only one dominant allele, does not express the dominant trait as fully as a homozygous dominant individual does. Therefore, a heterozygote has a phenotype that is intermediate between that of a homozygous dominant individual and that of a homozygous recessive individual.

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The ABO blood group exemplifies two different exceptions to Mendelian inheritance:

- Multiple alleles - there are three alleles for the gene that determines ABO blood type, but any number of alleles greater than two violates one of the assumptions of Mendelian inheritance. Since multiple alleles allow for multiple traits, there is no longer a simple dominance/recessivity relationship between alleles.
- Codominance - two different alleles act as dominant alleles and each of the corresponding phenotypes is fully expressed at the same time. In the ABO example. A type AB person (a heterozygote with an A allele and a B allele) has blood cells that are just as much type A as a fully type A person and that are just as much type B as a fully type B person.

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The control of coat color in rats is complicated by an exception to Mendelian inheritance called epistasis. Epistasis is interaction between two different genes. In the case of rats, the protein product of the C gene acts to turn on the gene controlling the production of pigment (gene B). For the B gene, black (B) is dominant over brown (b). However, neither color of pigment is produced unless expression of the C allele of the C gene turns on the B gene. If the C genotype is cc, there is no protein made that turns on the B gene, so the coat is white.

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Another exception to Mendelian inheritance is polygenic inheritance, in which several different genes make individual contributions to a single phenotype. In such a case, there are many possible traits for the characteristic, and the phenotype within a population varies along a continuum. Human skin color is an example of a characteristic that is controlled by polygenic inheritance.

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Even genetically identical individuals can express different phenotypes because of environmental influences.

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Pedigree analysis is useful in determining the genetics of humans, because humans cannot be bred for genetic experiments. Not only is such breeding legally and ethically prohibited, but it would take decades to get any results for humans, and even then those results would not be useful, because an individual woman does not produce a great enough number of offspring for statistically useful purposes.

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A pregnant female can undergo either amniocentesis or chorionic villus sampling to test whether a fetus she is carrying has genetic defects.