

# Single Gene Disorders - Autosomal Dominant Inheritance

<https://pubmed.ncbi.nlm.nih.gov/32491444/>

The field of genetics was born through meticulous studies in a monastery garden by a 19th-century monk, Gregor Mendel. His proposed laws explained the modes of inheritance of characteristic traits passed on through generations, such as the flower color of a pea plant. Though it would be many years before the term gene was introduced and much has been learned since his initial observations, the laws have withstood our advances and understanding of biology, with some interesting exceptions. Gregor Mendel proposed three laws explaining the inheritance of traits visible through generations - the characteristic of pea skin - wrinkled or smooth, the color of a pea plant flower - white, pink, red - among other features. We now understand that these traits are encoded in our instruction manual or our DNA. These simple changes to the phenotype, or the trait displayed in an organism, can be explained through changes in our genes. Mendel's laws include the Law of Dominance and Uniformity, the Law of Segregation, and the Law of Independent Assortment.

First, the Law of Dominance and Uniformity states that some alleles, which are variants of a particular gene found at the same chromosomal locus or location, are dominant over the other alleles for a given gene. Those traits that are not dominant are termed recessive. If an organism inherits at least one dominant variant, then it will display the effect, or phenotype, of the dominant allele. Second, the Law of Segregation states that the two alleles for each gene separate from each other during gametogenesis so that the parent may only pass off one allele; thus, the offspring can only inherit one allele from each parent. Third, the Law of Independent Assortment (Law of Reassortment) states that the alleles of different genes segregate independently of one another

during gametogenesis and are distributed independently of one another in the next generation. This concept was later verified with chromosomes, though also disproven in some instances. For example, when genes occur on the same chromosome, they can be linked and not follow this law.