3.1: Mendel’s First Law

Character Traits Exist in Pairs that Segregate at Meiosis

Through careful study of patterns of inheritance, Mendel recognized that a single trait could exist in different versions, or alleles, even within an individual plant or animal. For example, he found two allelic forms of a gene for seed color: one allele gave green seeds, and the other gave yellow seeds. Mendel also observed that although different alleles could influence a single trait, they remained indivisible and could be inherited separately. This is the basis of Mendel’s First Law, also called The Law of Equal Segregation, which states: during gamete formation, the two alleles at a gene locus segregate from each other; each gamete has an equal probability of containing either allele.

Hetero-, Homo-, Hemizygosity

Mendel’s First Law is especially remarkable because he made his observations and conclusions (1865) without knowing about the relationships between genes, chromosomes, and DNA. We now know the reason why more than one allele of a gene can be present in an individual: most eukaryotic organisms have at least two sets of homologous chromosomes.
For organisms that are predominantly diploid, such as humans or Mendel’s peas, chromosomes exist as pairs, with one homolog inherited from each parent. Diploid cells therefore contain two different alleles of each gene, with one allele on each member of a pair of homologous chromosomes. If both alleles of a particular gene are identical, the individual is said to be homozygous for that gene. On the other hand, if the alleles are different from each other, the genotype is heterozygous. In cases where there is only one copy of a gene present, for example if there is a deletion on the homologous chromosome, we use the term hemizygous.

Although a typical diploid individual can have at most two different alleles of a particular gene, many more than two different alleles can exist in a population of individuals. In a natural population the most common allelic form is usually called the wild-type allele. However, in many populations there can be multiple variants at the DNA sequence level that are visibly indistinguishable as all exhibit a normal, wild type appearance. There can also be various mutant alleles (in wild populations and in lab strains) that vary from wild type in their appearance, each with a different change at the DNA sequence level. Such collections of mutations are known as an allelic series.