4.1: Mutation and Polymorphism

We have previously noted that an important property of DNA is its fidelity: most of the time it accurately passes the same information from one generation to the next. However, DNA sequences can also change. Changes in DNA sequences are called mutations. If a mutation changes the phenotype of an individual, the individual is said to be a mutant. Naturally occurring, but rare, sequence variants that are clearly different from a normal, wild-type sequence are also called mutations. On the other hand, many naturally occurring variants exist for traits for which no clearly normal type can be defined; thus, we use the term polymorphism to refer to variants of DNA sequences (and other phenotypes) that co-exist in a population at relatively high frequencies (>1%).
Polymorphisms and mutations arise through similar biochemical processes, but the use of the word “polymorphism” avoids implying that any particular allele is more normal or abnormal. For example, a change in a person’s DNA sequence that leads to a disease such as cancer is appropriately called a mutation, but a difference in DNA sequence that explains whether a person has red hair rather than brown or black hair is an example of polymorphism. Molecular markers, which we will discuss in Chapter 9, are a particularly useful type of polymorphism for some areas of genetics research.

Contributors and Attributions

- Dr. Todd Nickle and Isabelle Barrette-Ng (Mount Royal University) The content on this page is licensed under CC SA 3.0 licensing guidelines.