

Polymorphism

Polymorphism is a very broad term in general use. Polymorphism crosses several discipline boundaries, including ecology, genetics, evolutionary theory, taxonomy, cytology and biochemistry. In biology, polymorphism has been given a specific meaning, being distinguishable from mono-morphism (having only one form). In other terms, polymorphism is the occurrence of two or more clearly different morphs of forms, also referred to as alternative phenotypes, in the population of a species. More simply, polymorphism is when there are two or more possibilities of a trait on a gene. For example, there is more than one possible trait in terms of a human eye colouring; they can be of different colour in humans.

Polymorphism (also referred to genetic polymorphism) is used somewhat differently by geneticists and molecular biologists to describe certain mutations in the genotype, such as single nucleotide polymorphisms (SNPs) that may not always correspond to a phenotype, but always corresponds to a branch in the genetic tree.

A gene is said to be polymorphic if more than one allele occupies that gene's locus within a population. In addition to having more than one allele at a specific locus, each allele must also occur in the population at a rate of at least 1% to generally be considered polymorphic. Gene polymorphisms can occur in any region of the genome. The majority of polymorphisms are silent, meaning they do not alter the function or expression of a gene. For example, ABO genes responsible for blood grouping in humans. Some polymorphism is visible. For example, in dogs the E locus, can have any of five different alleles, known as E, E^m, E^g, E^h, and e. Varying combinations of these alleles contribute to the pigmentation and patterns seen in dog coats.

A polymorphic variant of a gene can lead to the abnormal expression or to the production of an abnormal form of the protein; this abnormality may cause or be associated with disease. For example, in humans, HbA allele in hemoglobin gene is mutated to HbS allele which leads to condition commonly known as sickle cell anemia.