Polymorphism

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A combination of the Greek words *poly*(meaning multiple) and *morph* (meaning form), polymorphism is a term used in genetics to describe multiple forms of a single gene that exists in an individual or among a group of individuals.

Genetic Polymorphism Defined

Where monomorphism means having only one form and dimorphism means there are only two forms, the term polymorphism is a very specific term in genetics and biology, relating to the multiple forms of a gene that can exist. The term does not extend to character traits with a continuous variation such as height (even though this may be a heritable aspect).

Instead, polymorphism refers to forms that are discontinuous (have discrete variation), bimodal (having or involving two modes), or polymodal (multiple modes). For example, earlobes are either attached, or they are not, it is an either/or situation and not like height, which is not a set number or another.

Polymorphism was originally used to describe visible forms of genes, but the term is now used to include cryptic modes such as blood types, which require a blood test to decipher. Also, the term is sometimes used incorrectly to describe visibly different geographical races or variants, but polymorphism refers to the fact that the multiple forms of a single gene must occupy the same habitat at the same time (which excludes geographical, race or seasonal morphs.)

Genetic polymorphism refers to the occurrence of two or more genetically determined phenotypes in a certain population (in proportions that the rarest of the characteristics cannot be maintained just by recurrent mutation). Polymorphism promotes diversity and persists over many generations because no single form has an overall advantage or disadvantage over the others in terms of natural selection.

Polymorphism and Mutation

Mutations by themselves do not classify as polymorphisms. A polymorphism is a DNA sequence variation that is common in the

population. A mutation, on the other hand, is any change in a DNA sequence away from normal (implying that there is a normal allele running through the population and that the mutation changes this normal allele to a rare and abnormal variant.)In polymorphisms, there are two or more equally acceptable alternatives and to be classified as a polymorphism, the least common allele must have a frequency of 1 percent or more in the population. If the frequency is lower than this, the allele is regarded as a mutation.

Polymorphism and Enzymes

Gene sequencing studies, like that done for the human genome project, have revealed that at the nucleotide level, the gene encoding a specific protein can have a number of differences in sequence.These differences don't alter the overall product significantly enough to produce a different protein but may have an effect of substrate specificity and specific activity (for enzymes), binding efficiencies (for transcription factors, membrane proteins, etc.) or other features and functions. For example, within the human race, there are many different polymorphisms of CYP 1A1, one of many cytochrome P450 enzymes of the liver. Although the enzymes are basically the same sequence and structure, polymorphisms in this enzyme can influence how humans metabolize drugs. CYP 1A1 polymorphisms in humans, where, in exon 7 the Isoleucine amino acid is replaced by Valine, has been linked to smoking-related lung cancer. The use of genetic polymorphisms was one of the strengths of deCODE Genetics, a company that focused on determining genetic risk factors for various diseases.