

# Genetic heterogeneity and protein polymorphisms

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**Protein polymorphism** is the existence of several different variants of a certain type of protein

## Genetic heterogeneity

**Genetic heterogeneity** is the presence of multiple forms of a certain trait that occurs more often than 1% of cases in the population. If this value is lower, we are talking about mutation. Heterogeneity is often determined already at the DNA level. Polymorphisms in DNA are detected using molecular genetics.

As mentioned below, different mutations creating genetic heterogeneity can lead to an individual advantage. However, these changes can also harm their wearer. Diseases caused by heterogeneity of alleles or loci include, for example:

- CFTR gene for cystic fibrosis (1000 forms of this gene are known),
- retinitis pigmentosa.

## Protein polymorphism

**Protein polymorphism** is the existence of several different variants of a certain type of protein. Example: enzyme - carboxylase in the population. Sometimes we also talk about biochemical polymorphism. The reason for the diversity of individual proteins is the different primary structure of the proteins. It can often be the replacement of individual amino acids, changes in charge, changes in the size or arrangement of the molecule. Changes in the primary structure can sometimes lead to the loss of protein function and result in many genetic diseases. We most often encounter with **codominance**. An example is human hemoglobin:

### Hemoglobin polymorphism

According to the primary structure, we distinguish 4 basic chains of human hemoglobin - each of them is encoded by a different gene. By **polymorphic system** we mean all forms of protein - i.e. hemoglobin (Hb). Individual **variants** are then denoted by capital letters - HbA, HbB. The polymorphic *type* refers to the combination of individual variants and is a manifestation of the genotype, i.e. the phenotype - HbAA, HbAB, HbBB.

### Meaning of polymorphism

Protein polymorphism probably served to preserve a certain random mutation that favored its bearer. A typical example is the existence of hemoglobin polymorphism and its relation to malaria. Carriers of the HbA/HbS polymorphic type have been shown to be resistant to malaria and provide them with a distinct advantage. It is actually a form of selection.

## Links

### Related articles

- Polymorphism
- Cystic fibrosis
- Selection
- Mutation
- Malaria

### External links

Genetický polymorfismus (<http://ucebnice.euromise.cz/index.php?conn=0&section=biostat2&node=38>)

Genetic heterogeneity ([https://en.wikipedia.org/wiki/Genetic\\_heterogeneity](https://en.wikipedia.org/wiki/Genetic_heterogeneity))