Protein Polymorphism

Polymorphism is a term for a condition where there are *at least 2 genetic variants* (alleles) of some sign in the population. At the same time its frequency of occurrence in the population exceed 1%. If the percentage of occurrence is smaller, we're talking about a random occurrence – mutation.

Polymorphisms have their bases in the DNA structure. Due to the variability of the genetic code, individual polymorphisms may not always been shown. Especially when they are present in non-coding regions – *introns*.

With the knowledge of localization of polymorphisms we are able to create *genetic maps*. **Short repetitive sequences**a (*microsatellites*) are highly specific to the individual and can therefore be used for example in determining parenthood. Offspring inherit different alleles from their parents.

Types of Polymorphism

Spot polymorphism

- Arises due to *point mutations* in DNA. Typically, single nucleotide substitution that leads to the inclusion of other amino acids. This condition is detected using the **RFLP** – Restriction Fragment Length Polymorphism.
- Sometimes this type of polymorphism is also known as **SNPs** – Single Nucleotide Polymorphism. In some studies it has been found a link between certain forms of the SNP and multifactorial diseases – such as *hypertension*, *diabetes* or *heart disease*.

Repetetive sequences

- Sections of nucleotides in DNA that occur in many copies. Their length and number varies in *individual from person to person*. Its inheritance is corresponds to Mendelian Inheritance Rules.

RFLP

Replacement of even a **single nucleotide** can lead to change of the DNA information, thanks to which there is lost a section for breaking down by **restriction enzymes**. Currently, there is a proliferation of strands of DNA examined by **PCR** (polymerase chain reaction). On the basis of different function of restriction enzymes, fragments of various lengths are formed. They have different rates of migration during the electrophoresis. Longer fragments move more slowly.

Polymorphisms in Biochemistry

Also in biochemistry, we see the existence of several variants that may come from the **same locus**. *For example*: the different types of **hemoglobin** - α, β, γ, δ. Each of them is determined by its own structural gene. For each type is typical a small difference, that occurs in the chain - which often means only one amino acid substitution.

We know also many **pathological polymorphisms**. The change of the DNA sequence can also lead to incorrect transcription of information and the subsequent *lack of an enzyme*.

Why Does Polymorphism Exist?

There are several theories of its origin. Obvious is the effort of the organism **to benefit** a particular genotype. The most common form of change is **mutation**. This can lead (for example) to change the produced quantity or efficiency of a protein. Polymorphisms may also occur randomly – by **genetic drift**.

The frequency of polymorphisms differ considerably, according to various authors. Their incidence is approximately equivalent to **1:500–1000** pairs of bases[1]. In non–coding regions, polymorphisms can occur even at every 50^{th} nucleotide, but they have no clinical significance.

Links

Related articles

- Allele

- Mutation
- Introns
- Genetic Mapping
- RFLP

Sources

- Wikipedie, otevřená encyklopedie. *Genetický polymorfismus* [online]. ©2011. The last revision 2011-07-23, [cit. 2011-07-31]. <http://cs.wikipedia.org/wiki/Genetick%C3%BD_polymorfismus>.

References

1. KUMA, VINAY – ABBAS, ABUL K – FAUSTO, NELSON, ET AL,. *Robbins Basic Pathology.* 8th edition edition. 2007. ISBN 1416029737.