Monogenically inherited diseases

Monogenically inherited diseases arise as a result of a mutation in a single gene and their infuluence by the external environment is minimal. More than 10 000 monogenically conditioned disorders are currently known.

Basic characteristics

The nature of the disease depends on the function of the modified gene. In the case of metabolic disorders, a mutation can cause insuffcient expression of a gene and thus a deficiency of the enzyme it codes for, which results in a disorder of the metabolic pathway in which the given enzyme participates. There is also a disorder in the synthesis of structural proteins, which in most cases has consequences for the entire organ systems.The disorder is not necessarily caused by insufficient expression, but often a product with altered function is synthesized based on the mutation, or the expression of the given gene is increased. In the vast majority of cases, these disorders have much more serious consequences for the sufferer.

We divide monogenic diseases according to the type of inheritance into:

- Autosomal dominant (AD)
- Autosomal recessive (AR)
- X-linked

Diagnosis of monogenic diseases can be direct or indirect. It depends if we know the casual mutation or the polymorphism that is linked to the gene.

The treatment of some monogenic diseases is possible at the phenotypic level, when it is a symptomatic treatment. Unfortunately, modern medicine cannot deal with most of them. Gene therapy, which is curretnly helpful for example in cystic fibrosis, is a hope for the affected in the future, but it is not permanent for the time being. In principle, it is an attempt to replace a non functional gene or to inhibit its excessive expression.

Examples of monogenically inherited diseases

Galactosemia

In this disease, the organism is unable to metabolize galactose (lack of enzyme galactose-1-phospaturidyltransferase), which leads to its accumulation in the tissues. The liver, eyesight, kidneys and nervous system can be damaged. Inheritance is autosomal recessive.

Tay-Sachs disease

🔎 *For more information see Tay-Sachs disease.*

Tay-Sachs syndrome is a fatal genetic disorder of metabolism, in which gangliosides accumulate in nerve tissue. The cause of the disease is insufficient expression of the gene for the enyzem Hexoaminidase A, which has the task of breaking down gangliosides. Inheritance is autosomal recessive.

Marfan syndrome

🔎 *For more information see Marfan syndrome.*

It is an autosomal dominant connective tissue disease that affects the skeleton, heart, eyes, blood vessels and lungs. The cause is a defect of fibrilin formation. Clinical signs include excessive growth, chest excavation, aortic dilatation, long and thin fingers.

Links

References

- http://www.who.int/genomics/public/geneticdiseases/en/ (http://www.who.int/genomics/public/geneticdiseases/en/)
- ŠÍPEK, Antonín. *Genetika* [online]. [cit. 2011]. <http://www.genetika-biologie.cz/geneticky-podminene-choroby>.