Molecular basis of hereditary diseases

General

Hereditary diseases represent an important group of human diseases, including, for example, various metabolic defects, immune disorders, hemoglobinopathy or selected congenital malformations. The cause of hereditary diseases is generally a *disruption* of a series of reactions leading from the genetic information (genotype) to the corresponding bodily function or characteristic (phenotype).

The general scheme showing the path from gene (or from DNA) to protein is referred to in molecular biology as the **central dogma . *A*** *Mutation* at the DNA level can significantly disrupt the normal sequence of reactions and often leads to the development of a pathological phenotype - i.e. *to the development of the relevant disease*.

Normal state:

gene (DNA) → mRNA with standard sequence → functional protein → normal function/trait

Pathological condition:

Mutated gene (DNA) → mRNA with non-standard sequence → non-functional protein → damaged function/trait → disease

Of course, such a scheme is only a rough guide, in certain cases the transcription process itself is disturbed as a result of the mutation, and the disturbed function may not be caused only by a non-functional protein, but also by its lack.

Examples

defect type	examples of disabilities
enzyme defect	PKU, galactosemia, adenosine deaminase deficiency
receptor defect	testicular feminization, hypercholesterolemia
molecular transport defect	cystic fibrosis, hypertension
cell structure defect	Duchenne and Becker muscular dystrophy
defect homeostasis	antihemophilic globulin, immunoglobulins
growth and differentiation regulation defect	sex determination, X chromosome inactivation, tumor suppressors
intercellular communication defect	insulin, growth hormone, sex differentiation
defect mitochondria	Leber optic atrophy

Links

Related Articles

- Mutation
- Principles of therapy for hereditary diseases
- Primary immunodeficiency
- Inherited metabolic disorders

Source

- ŠTEFÁNEK, Jiří. *Medicína, nemoci, studium na 1. LF UK* [online]. [cit. 11.02.2010]. <https://www.stefajir.cz/>.