Clinical genetics

Clinical or **medical genetics** is a separate medical field. It is based on the knowledge of general and experimental genetics, which it uses to investigate the influence of genetic and external factors on the emergence of various human diseases and defects. Hereditary diseases and congenital developmental defects affect all organ systems, therefore the vast majority of medical fields encounter these groups of diseases. However, it is clinical genetics that plays a central role in approaching these groups of diagnoses. Accurate diagnosis using cytogenetic or molecular-genetic examination, determination of the risk of recurrence of this pathology and possible preventive approaches - these are the main tasks of clinical genetics.

Genetic consultation is an indispensable part of clinical-genetic practice. The purpose is to provide the patient with all **important information** about his disease, to explain the results of the tests carried out so far, to analyze with him the possible causes of his disease, the treatment options, but mainly to **determine the risks** of recurrence of this disease in his descendants, or to propose measures that would reduce this risk. Genetics is not an entirely simple matter - therefore a careful and understandable explanation of the necessary information to the patient is extremely important in this case.

The basic method of clinical genetics remains a genealogical examination with an analysis of the family history. After the basic assessment of the case, further examinations may be indicated (karyotype examination - see Indications for chromosomal examination, molecular-genetic examinations).

It is important to mention that clinical genetics is a **strictly non-directive** field. All decisions (for example, the decision to artificially terminate a pregnancy in case of detection of a serious developmental defect of the fetus) are completely voluntary, and the clinical geneticist is only supposed to provide enough information for the patient's free decision. He should definitely not force the patient to make any decision.

Tasks of clinical genetics

Prevention

Despite some advances in gene therapy, it is currently not possible to routinely treat genetically determined diseases in a causal way. Therefore, **prevention** is still the most important task of medical genetics. Prevention is related to identifying the genetic risk of various defects or diseases. The risks of possible external factors that could have an influence on the genetic information (risks of the working environment, etc.) are also identified. As part of the prevention of birth defects, cooperation with other disciplines is also important. Based on the established facts, an optimal solution to the situation is sought.

In the case of congenital developmental defects, **family planning** is a good prevention. Prophylactic administration of **folic acid** can have a preventive effect on congenital neural tube defects.

Diagnostics

The task of **diagnostics** is to detect congenital developmental defects or genetically determined diseases and classify them accurately. **Prenatal diagnosis** refers to the examination of an unborn individual. Conversely, postnatal diagnosis refers to individuals already born. There are various biochemical, cytogenetic, molecular genetic or imaging tests that can be used to diagnose defects or diseases. The carrier of certain diseases is also investigated, when the individual is not affected by the disease himself, but can pass this hereditary assumption on to his descendants. Various defects, diseases or syndromes may not be the first to be diagnosed by a geneticist (usually a pediatrician or an internist), but genetic testing is often required to confirm the diagnosis.

Early prenatal diagnosis of congenital developmental defects also has a great therapeutic benefit. Although only a minimum of conditions can be solved prenatally today, intensive and targeted perinatal therapy can be decisive for the survival or future life of a child with a congenital defect.

Treatment of defects and diseases

The treatment of genetically determined diseases at the molecular level (that is, at the level of DNA or RNA) is still in the development stage. Therefore, even today, it is still only about symptomatic care, where we try to limit the manifestations of the disease without treating its cause (which is a mutation in DNA). The actual symptomatic treatment is already the task of specialized fields. A number of structural congenital defects can be solved surgically, and early diagnosis is often decisive (for example, in the case of congenital heart defects). In addition to surgery, other fields are also used in the treatment, a number of metabolic diseases can be compensated very well with a special diet, etc.

Registration and monitoring

The incidence of congenital developmental defects is registered in the Czech Republic, as in most developed countries, for the purpose of better awareness of the state of the population and the success of prenatal diagnosis, as well as for the purpose of discovering new factors for the development of these defects.

Spectrum of cases

Clinical genetics deals with a wide range of cases and different diagnoses. However, some groups are dealt with more often. These are mainly the following cases:

- Newborns and children with suspected congenital developmental defects.
- Children or adolescents with a psychiatric finding, raising suspicion of chromosomal or other genetic etiology (inborn metabolic disorders).
- Children and adolescents with a growth disorder or with a disorder of sexual development.
- Children and adults with suspected genetic diseases (for example, suspected thrombophilic mutations or cystic fibrosis).
- Persons with a family history of frequent occurrence of cancer at a younger age, in whom the family occurrence of genetically determined cancer syndrome is suspected.
- Persons or couples suffering from sterility (long-term unsuccessful attempt to conceive) or reproductive losses (repeated abortion).
- Couples coming to a preconception consultation, for example due to the presence of a birth defect or a genetically determined disease in the family history.
- Pregnant women in whom a screening examination (biochemical or ultrasound) has indicated an increased risk of congenital developmental defects, or in whom this risk is increased due to age (generally over 35 years).

Cooperation

Due to its position, clinical genetics cooperates with almost all medical and diagnostic fields. Nevertheless, it is possible to find several fields in which cooperation is most common.

- Cytogenetic laboratories and molecular-genetic laboratories.
- Gynecology and Obstetrics (including Fetal Medicine and Ultrasound Diagnostics).
- Pediatrics and neonatology.
- Clinical Biochemistry.
- Oncology.
- Neurology and Psychiatry.
- Plastic surgery.

Links

Related articles

- Prenatal diagnosis
- Indications for karyotype examination
- Congenital developmental defects
- Hereditary tumor syndromes
- Hereditary metabolic disorders
- Chromosomal aberrations

Sources

- ŠÍPEK, Antonín. *Genetika* [online]. [cit. 1. 6. 2009]. <http://www.genetika-biologie.cz/geneticke-poradenstvi>.