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CONCEPT OF MEDICAL GENETICS

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In the medical sciences system, medical genetics is a discrete scientific discipline. Using its general methods, medical genetics analyses etiological involvement of genetic and external factors in the origin of diseases and disorders. Medical genetics contributes to explaining their formal and causal pathogenesis, offers new diagnostic possibilities, including cytogenetic testing and detecting heterozygotes, carriers, dispositions and patients, uses effective preventive procedures, such as choosing a healthy child, selecting the sex of the child, family precaution regime, etc.

Medical genetics also brings new aspects and develops methods to regulate population development thus helping to solve problems with population quality. The reviewing, counselling and expert activities of medical genetics assist healthcare and social institutions. Medical genetics is involved in establishing correct and timely diagnosis for genetically determined diseases as well as in their screening, prevention and therapy.

Effective use of medical genetics in treatment and preventive care requires

1. deepening of medical genetics knowledge of physicians as stipulated in the Government Resolution No. 59/1966,
2. supporting research,
3. supporting practical use of medical genetics in all fields of treatment and preventive care,
4. establishing a network of medical genetics sites that would centrally provide general tasks of the field on local basis spanning the territory of one to two regions (hereinafter only “the medical genetics department”).

Medical genetics departments are to be set up in hospitals with type III outpatient clinic, especially in university hospitals.

In a university hospital, medical genetics department may be located in the building of institute of biology.

Qualification for the head of the department is in the first stage contingent on a sufficiently long duration of practical training in medical genetics and theoretical training in a training facility.

Medical genetics department shall carry out the following tasks:

a) Genetic assessment for diagnostic and differential diagnostic purposes for local inpatient and outpatient facilities, in particular genealogic, cytogenetic and anthropogenetic assessments. In association with clinical biochemistry department and transfusion department, medical genetics department also provides necessary biochemical and immunobiological assessments.

In order to provide large-scale and other specific biochemical assessments for medical genetics department, it is useful to single out the required number of staff members (e.g. a biochemist and medical laboratory technicians) from medical genetics department who would perform these tasks in the department of clinical biochemistry.

b) Record keeping and screening of individuals and families with serious genetically determined diseases for which effective and feasible form of prevention is known. Medical genetics department provides preventive measures for the registered families.

c) Providing counselling to actively sought out families where genetically determined diseases occur. If necessary, performing genetic testing for such families, establishing familial and individual prognosis, proposing methods of prevention and providing treatment and preventive care.

d) In association with department for organization and methods participating in monitoring of demographic and population development, working on analysing it and establishing its prognosis and on proposing regulations.

e) Collaborating with regional experts on practical use of medical genetics, i.e. in prevention, capture and treatment of genetically determined diseases and diseases with genetically conditioned disposition (so-called special modifications).

f) Expertly supervising local physicians in medical genetics issues.

Stages of Implementation

The first stage follows from the current situation and aims to enforce and differentiate the existing sites focusing on medical genetics. It is anticipated that this stage will be accomplished in 1972 and consists of:

a) finishing the principal medical genetics department in collaboration with university hospitals in Prague. An organization model of an independent medical genetics department will be reviewed, methods not yet used in this country will be implemented and staff will be trained.

b) appointing sites for future independent medical genetics departments with local or regional coverage and their gradual material equipping and staffing. The sites will gradually fulfil medical genetics tasks according to their abilities.

The second stage anticipates finishing the independent medical genetics departments with local or regional coverage so that the entire territory of the country is covered and at least minimal equipment is secured (in years 1973-1980).

Medical Genetics Department Scope of Work and Work Methods of the Discipline

1. **Family precaution regime** is a generally applicable method for hereditary diseases (so-called special modifications). These conditions and deviations affect more than half of population. There is a great probability that relatives will be affected with these conditions and deviations and based on genetic knowledge individuals can be identified within the families who may not yet show clinical symptoms but there is a high probability that they are going to become ill. Family precaution regime is therefore a method for individualized and targeted prevention and should routinely constitute part of treatment preventive care associated with these conditions.
2. **Differential diagnostic assessments:** About 1 % of all new-born babies are affected with a disorder caused by chromosomal aberrations. Adequate diagnostic tests for this are sex chromatin identification or chromosomal testing that have not been routinely done here yet. About 2 % of all new-born babies are affected with metabolic deviations (various types of oligophrenia, congenital defects and disorders) that require complex genetic testing, i.e. biochemical genetics and genealogical testing methods. In most diseases with genetic component, specialized genealogic and anthropogenetic assessments that could specify the individual and familial prognosis are not being done yet.
3. **Seeking out heterozygotes, carries, dispositions and patients**: A number of specific methods of medical genetics allow for identification of the appropriate type of individual within a family or population, which fact has an extraordinary importance in practice. Either a patient or dispositions for a condition are detected in time (e.g. phenylketonuria and galactosaemia), or diagnosis is specified in more detail before the baby is born and the prognosis is established, a prognosis for a specific type of marriage is established.
4. **Genetic counselling** provides a family with true information based on family testing using genetic methods. Proposes and recommends preventive measures within the family. Besides the already listed preventive methods, this may be for instance a recommendation to carry out sterilization or abortion for genetic reasons if there is a high probability of the children in the family being born with a serious disorder. The counsel shall also establish disease prognosis for the individual and the family.
5. **Choosing the sex of the child:** Some serious diseases show so-called sex-linked and sex-limited inheritance. These diseases affect about ¼ % of the population. The more familiar of these diseases include for instance A and B haemophilia. Transmission in a family is of such nature that it can often be determined if male or female child is going to be affected. For example in dominant inheritance, an affected man's daughters will all have the disease but none of his sons will. In this case, the sex of the foetus can be found out in week 11 of pregnancy (by testing sex-chromatin in amniotic fluid epithelium) and if a sex that is at hazard is detected, abortion is performed. In Denmark this method has become a routine already. The incidence of these diseases may in this way be significantly decreased over a short period of time.

6. **Choosing a healthy child:** This is a similar concept. In indicated cases the foetus is tested for presence of metabolic or chromosomal deviation. (This method has not been elaborated for broad range of use yet.)

7. **Genetic register and statistics** is an important part of genetic practice, collecting verified data about a family and population, helping with active search for family members at hazard by using genetic knowledge and registering family relationships. At the same time, a register allows for rational evaluation of population development and provides data for population development estimates.

8. **Population genetics** elaborates population models that allow for analyses of qualitative population development thereby contributing to scientifically supported population policies. The major criterion is a qualitative development of the gene pool (the sum of genetic information in a population), the aim is to positively affect the population quality by social and healthcare policies (e.g. salary policies, family allowance system and other social measures may have counterselective effects, just as the abortion act and co-education in special schools).

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