Bulletin of the Ministry of Health of the Czech Socialist Republic 1980, pg. 127—131, Section 21--24

14
POLICY OF MEDICAL GENETICS

1. **Contents and tasks**

1.1. Medical genetics is an independent scientific field within the system of medical sciences. It is an extension to the field of internal medicine, paediatric medicine, gynaecology and obstetrics. It is based on the findings of general and experimental genetics. Using its own methods, it analyses the etiological share of genetic and external factors in the occurrence of illnesses and defects. It contributes to the explanation of their formal and causal pathogenesis, and develops new diagnostic tools of molecular-genetic testing and discovery of heterozygotes, carriers of predispositions and disorders. It develops and uses methods that can effectively influence human reproduction and healthy development of the young generation in collaboration with other kinds of preventive medical care, thus improving the quality of population. Medical genetics is importantly characterised by its preventative character, and its principles are used in all areas of preventive medical care.
Focus on health education is used in order to improve health awareness of the population with respect to genetic illnesses and disorders.
Further development of medical genetics and its research depends on interdisciplinary integration of efforts and means, on the creation of specialised institutes of interdisciplinary fields, as well as on international collaboration and integration, especially within the framework of the Council for Mutual Economic Assistance (hereinafter 'Comecon').

1.2 Medical genetics focuses of the following tasks:
a) within the framework of preventive medical care, it aims at early diagnosis, treatment, and prevention of genetic disorders and congenital developmental defects (hereafter 'CDD') in humans. In particular:
— it provides genetic counselling, determines genetic risks in families and population, and recommends suitable preventive measures and treatment;
— it diagnoses chromosomally and metabolically conditioned defects and other congenital defects in various stages of ontogenesis;
— it diagnoses teratogenic, mutagenic, and other genetically hazardous factors during the pre-conception, prenatal, and postnatal stage;
— it collaborates with institutions of hygienic services in protection against known harmful environmental factors;
— it provides counselling, expertise, and expert opinion in its field.
b) It keeps record of genetic disorders and CDD in the population. In particular:
— it develops a register of genetically handicapped families and families with increased risk of CDD and genetic disorders, and actively searches out handicapped individuals and carriers of CDDs and genetic disorders;
— it collects and analyses data on the current state of genetic stress the population is exposed to;
— it develops prognoses of further genetic development of the population and suggests measures leading to a lowering of genetic stress;
c) it aims to meet scientific and research goals of the state plan of technical development and research plan pertinent to its folio.

2. Departments of Medical Genetics

2.1 Departments of medical genetics are integrated into hospitals with a Type III polyclinic (hereinafter 'HwP'). They are established as polyclinic departments. In regions where there is more than one Type III HwP, department of medical genetics is established in one of them. A medical genetics department consists of:
— an office for genetic counselling, which provides genetic counselling to families in which genetic disorders and CDD occur. This office also develops a register of families that carry a genetic risk;
--- office for cytogenetic testing, which provides cytogenetic, chromosomal, eventually also other specialised testing also for other departments of preventive medical care including pathology; it closely collaborates with other specialised and reference departments of medical genetics;
Where needed and subject to a decision of a department of health of the relevant regional national

committee, detached offices of departments of medical genetics which provide genetic counselling and cytogenetic testing may be established in other Type III HwPs, eventually also Type II HwPs. This holds especially in cases of increased incidence of genetic disorders.

2.2 Departments of medical genetics at Type III HwPs in select regions may establish, where needed and subject to a decision of the Ministry of Health of the Czech Socialist Republic, specialised departments of medical genetics serving a larger area. Such departments would include:
--- a specialised section of prenatal genetic diagnostics (app. one department per 2—3 million inhabitants);[1]
--- a specialised section for diagnostics and treatment of congenital metabolic disorders or other CDDs.[2]
If needed, other specialised sections may be established (e.g. for the testing of alpha-fetoproteins at the department of medical genetics of teaching hospitals in Prague and in Brno);
--- a pre-conception section of genetic care at select obstetric departments (clinics) of Type III HwPs of teaching hospitals.
In order to use resources rationally, specialised departments of medical genetics may be -- based on mutual agreement -- located at relevant research institutes of the Ministry of Health of the Czech Socialist Republic or other departments and institutes:
--- a specialised section for the testing of mutagens with the Institute for Hygiene and Epidemiology in Prague;
--- a specialised section for prenatal genetic diagnostics – foetoscopy (and others) with the Institute for Care of Mother and Child in Prague.
In order to provide inpatient care for the diagnostics and treatment of genetically affected families and families with genetic risk, hospital beds are provided by the relevant specialised departments (such as internal medicine, paediatric, obstetric wards of the Type III HwPs).

2.3 Centre for Medical Genetics is established based on decision of the Ministry of Health of the Czech Socialist Republic. Its function is fulfilled by the Department of Medical Genetics of the Motol Teaching Hospital in collaboration with the Institute of Research of Child Development at the Faculty of Paediatric Medicine of the Charles University. In future, the task will be taken over by a hospital department of medical genetics with a capacity of some 25—30 beds. The Centre fulfills the most exacting functions of preventive medical genetic care, especially those related to diagnosis, treatment and screening of CDDs and genetic disorders. It establishes and further administrates a genetic register. It carries out the most important tasks pertaining to the development of genetics, tests existing research results, and assesses their practical application. It closely collaborates with other departments in the field, provides an umbrella for activities in medical genetics by advanced services and further develops medical genetics especially in the following directions: clinical and experimental cytogenetics, biochemical and population genetics, prenatal genetic diagnostics, anthropogenetics, oncogenetics, immunogenetics, pharmacogenetics, teratogenesis, etc. It participates in education and advanced training of medical genetics experts and in postgraduate courses in genetics for doctors from other medical fields in collaboration with the Institute for Further Education of Doctors and Pharmacists in Prague, medical faculties, and the Institute for Further Education of Nurses and Medical Personnel in Brno. It helps in developing guidelines for medical genetics and collaborates with departments of medical genetics and other departments of preventive medical care, with institutes of hygiene, departments of joint diagnostic and treatment sections, eventually also, based on mutual agreement, with specialised institutes of other kinds.

2.4 Reference offices of medical genetics are established as needed and in accordance with directives on establishment and activity of reference laboratories and departments of preventive care and treatment.[3]
Currently, they are gradually founded based on existing specialised departments which performed well in their specialised expertise and whose activity should be further extended in the interest of development of medical genetics.[4]

---

[1] As found at Departments of Medical Genetics of Teaching Hospitals in Prague, Brno, Hradec Králové and Olomouc.
[2] See Methodic Directive No. 15/1975 of Bulletin of the Ministry of Health on dispensary care of children suffering from phenylketonuria.
[3] Binding Directive No. 27/1977 of Bulletin of the Ministry of Health of the Czech Socialist Republic on the establishment and activity of reference laboratories for preventive treatment and care.
[4] This concerns the following departments:
-department of cytogenetic methods with the IIIrd Clinic of Internal Medicine of the Teaching hospital in Prague 2,

Tasks of departments of medical genetics are listed in Appendix No. 1.

3. **Collaboration and Relation Between Medical Genetics and Other Fields**

In screening, diagnostics, treatment, and prevention of CDDs, genetic disorders, and genetic predispositions as well as in the development of new methods of medical genetics, the field of medical genetics collaborates with all other medical fields. In particular:
— it develops extensive collaboration with internal medicine, general medicine, paediatrics, gynaecology and obstetrics, plastic surgery, orthopaedic medicine, neurology, psychiatry, etc.
— in select fields, such as internal medicine, paediatrics, gynaecology and obstetrics, eventually also other fields, it relies on doctors who have extended specialisation in the field of medical genetics or are trained in it, and are designated to collaborate with medical geneticists. Doctors with this training also provide specialised clinical testing for departments of medical genetics.
Collaboration with internal medicine is a key element in the development of comprehensive care for adults with genetic disorders or predispositions including malignant tumours. This is also a precondition of their prevention in future generations. Procedures which are to be followed in actively seeking out persons with genetic disorders and CDDs and their registration is described in Appendix No. 2:
— it uses the services of joint departments for diagnosis and treatment within Type III HwP, especially transfusion departments, departments. of clinical immunology and allergology and departments of clinical biochemistry;
— in assessing the cytogenic impact of harmful environmental factors and the use of human tissue cultures, it collaborates with the department of guidelines and organisation of regional institutes of national health and institutes of hygienic service;
— in assessing the teratogenic effects and mutagenic potential of chemical elements and compounds, it collaborates with research institutes belonging under the ministry of health, ministry of education, and the Institute of Hygiene and Epidemiology. In assessing teratogenic and mutagenic potential of drugs and biopreparations, it collaborates with the State Institute for the Control of Drugs and Medications;
— it collaborates with corresponding institutes in other countries, especially Comecon members.

4. **Experts, Their Training and Further Education**

4.1 Following kinds of workers are employed in this field:
--- medical doctors with further specialisation in the area of medical genetics and doctors currently training in this specialisation;
--- other experts with university education, such as graduates of faculties of natural science or faculties with natural science orientation, graduates of faculties of philosophy in areas of psychology or sociology;
--- paramedical healthcare workers — general, paediatric, obstetric nurses, laboratory workers with long-term training for work in medical genetics; lower and auxilliary medical personnel.

4.2 Medical doctors can obtain advanced specialisation in the field after completing 1st degree attest of specialisation in internal medicine, paediatrics, gynaecology and obstetrics, in exceptional cases also in some other specialisations. Their specialist training is regulated by directive No. 72/1971 Coll. on health workers and other specialised workers in healthcare.

4.3 Other specialised workers with university education who work in the field of medical genetics undergo in addition to continuous further training in their field also a course preparing them for working in the field of medical genetics. They take part in seminars, eventually other training in specialist courses organised by the Institute for Further Education of Medical Doctors and Pharmacists together with other

---

U nemocnice 1, Faculty of Internal Medicine of the Charles University in Prague;
-- department of prenatal genetic diagnostics at the Institute of Research of Child Development of the Faculty of Paediatric Medicine of the Charles University in the Teaching Hospital Prague-Motol;
-- department of pre-conception genetic care at the obstetric clinic of the Teaching Hospital Motol in Prague 2, Londýnská Str.
-- department of diagnostics of congenital disorders of metabolism of amino acids and carbohydrates with the Department of Clinical Biochemistry of the Teaching Hospital in Prague 2, Karlovo Square, Faculty of Internal Medicine, Prague
-- department for congenital metabolic disorders of lipo-proteins with the 3rd Clinic of Internal Medicine of the Teaching Hospital in Prague 2, U nemocnice 1, Faculty of Internal Medicine of the Charles University in Prague.

healthcare workers.

4.4 General nurses, paediatric and obstetric nurses and laboratory workers who work in the area of medical genetics undergo a long-term training, which should prepare them for specialised work. This training will be provided by the section for medical genetics of the Institute for Medical Genetics of Paramedical Personnel in Brno. The same Institute is also in charge of their further education in specialised courses and at training centres. Training seminars for paramedical personnel in the area of medical genetics will be provided by departments of relevant fields of the Institute for Further Education of Paramedical Personnel in collaboration with the sub-department of genetics of the Institute for Further Education of Medical Doctors and Pharmacists.

4.5 Doctors and other medical personnel of other specialisations who collaborate with institutes and departments of medical genetics will further their education in the field of medical genetics within training courses provided by the Institute for Further Education of Medical Doctors and Pharmacists in Prague, the Institute for Further Education of Paramedical Personnel in Brno, policy departments of regional departments of national health, and in collaboration with the chief expert and regional experts in the area of medical genetics.

5. **Supervision and Control of the Field of Medical Genetics**

5.1 Chief expert in the area of medical genetics aids the Ministry of Health of the Czech Socialist Republic in expert management of the field. In particular:
--- it is his task for manage, supervise, and test all activity within the field and propose to the Ministry of Health of the Czech Socialist Republic concrete proposals of measures;
--- he directs the activity of regional experts and of his advisory team;
--- he supervises the effectiveness of operation of departments in the field and the use of laboratory and testing equipment;
--- he proposes a practical regionalisation of departments of medical genetics, their integration in units covering larger area and efficient coordination of activities with the goal of effective use of personnel and technical equipment;
--- supervises the practical application of results of current research.

5.2 Regional expert in the field of medical genetics is responsible for expert management of the field of medical genetics within the region and has the same tasks as the chief expert.[5]

6. **Research in the Field**

is coordinated within the framework of the state plan of technical development and departmental plan of research.

6.1 Research focuses mainly of the following issues:
--- population genetics: analyses current genetic stress on the population and develops a prognosis, takes part in analysing genetic data with the aim to help the establishment of a genetic register;
--- cellular genetics: develops diagnostic methods of chromosomal disorders, studies mechanisms of transfer of genetic information and its disorders, studies risk factors present in the environment (mutagenesis), genetic mechanisms of malignant transformation of cells, develops methods of pre-zygotic, prenatal, and postnatal detection of genetic disorders and illnesses;
--- clinical genetics: develops new methods of diagnosis, prevention, and treatment of genetic disorders;
--- immunogenetics: participates in research of pathological development of the immune system, studies genetic variability of the immune system;
--- biochemical genetics: studies biochemical consequences of genetic disorders, possibilities of phenotypic treatment, and takes part in searching out heterozygotes and people with congenital metabolic disorders; as part of this activity, it develops pharmacogenetic drugs as one of the main tools of effective pharmacotherapy.

---

[5] Responsibilities and activities of the chief and regional experts is set in directive No. 46/1975 of the Bulletin of the Ministry of Health of the Czech Socialist Republic on principles of organisation of institutes of national health.

6.2 In all areas of research, international collaboration, mainly within the framework of Comecon and WHO, is of crucial importance, especially in the area of diagnosis of metabolic disorders, prenatal genetic diagnostic, immunogenetics, teratogenesis, and mutagenesis.

7. **Final Stipulation**

7.1 This policy replaces the policy of medical genetics published under No. 13/1970 of Bulletin of the Czech Socialist Republic.

Minister:
doc. MUDr. J. JIROUŠ, candidate of sciences

Appendix No. 1
Tasks of Departments of Medical Genetics

1. Departments of medical genetics fulfil chiefly the following tasks:
a) Genetic counselling offices:
— provide genetic counselling to families with CDD and genetic disorders and expert advise to healthcare institutions that care for genetically affected individuals; it also provides their genealogic examination;
— determine a genetic prognosis of these families and recommends preventive measures;
— provide anthropogenetic and psychological testing;
— assess genetic risks of planned marriages in marital and pre-marital genetic counselling;
— determine genetic risk of existing pregnancies in genetically affected families;
— provide genetic testing of children suitable for adoption;
— recommend abortion and sterilisation for genetic reasons;
— collaborate in active search for carriers of hereditary illnesses with other departments and healthcare institutions and suggests requisite measures;
They actively search for endangered family members of genetically afflicted families and based on analysis of provided data broaden the scope of registration of genetically affected families and determine their genetic risk. They register genetically affected families and complete their register in collaboration with departments of preventive care. List of individuals posing a genetic risk and such families serves as a basis for the establishment of a genetic register.

b) Offices for cytogenetic testing
--- carry out cytogenetic and chromosomal testing;
--- circumstances permitting, collaborate in biochemical, cytochemical, immunological and other testing;
--- provide genetically indicated testing for other departments of preventive medical care including pathological anatomy; specialised genetic testing is provided in collaboration with other specialised genetic and reference departments.

2. Specialised departments of prenatal genetic diagnostics:
--- provide specialised genetic counselling for potentially genetically risky pregnancies and recommend further prenatal genetic testing;
--- collaborate with select obstetric departments of Type III HwPs, which provide requisite testing and examinations (ultrasound examination of the foetus and placenta, amniotic fluid testing, amniocentesis and other requisite tests);
--- follow the development of the foetus, health of the mother, and in case of unfavourable genetic diagnoses recommend abortion; foetuses aborted for genetically indicated reasons are handed over to the relevant department of pathological anatomy of the Type III HwP;
--- carry out requisite testing of amniotic fluid or testing of cell cultures from other tissues of the foetus and provide necessary genealogic, clinical genetic, cytogenetic, immunogenetic, and histochemical testing and examination of pathological anatomy, etc., in collaboration with requisite specialised departments;
— keep records of tested genetically endangered pregnancies;
— collaborate with gynaecologists experienced in medical genetics who care for genetically endangered pregnancies;
— in collaboration with paediatric doctors and doctors caring for adolescent, they follows the

development of children who were registered in prenatal genetic care;
— in their diagnostic activity, they collaborate with other specialised departments and reference sections of medical genetics in the Czech Socialist Republic, Slovak Socialist Republic, eventually also other countries.

3. Reference departments of pre-conception genetic care
--- develop methods of pre-conception care of persons with genetic risk of polygenic CDDs and care of genetically endangered pregnancies;
--- develop methods of planned conception, methods of artificial insemination or sterilisation for genetic reasons.

4. Specialised sections of pre-conception genetic care
--- use above-listed methods of pre-conception genetic care of persons with genetic risk;
--- care for genetically endangered pregnancies;
--- recommend sterilisation for genetic reasons; their activities are closely lined with departments of medical genetics, specialised and reference departments of prenatal genetic diagnostics, and specialised sections of genetic counselling, mainly marital and pre-marital genetic counselling;
— take part in developing guidelines for workers within its area of activity.

5. Specialised sections for the diagnostics and treatment of congenital disorders
— their tasks are specified in methodic guideline No. 15/1975 Bulletin of the Ministry of Health of the Czech Socialist Republic.

6. Specialised section for the testing of mutagens with the Institute of Hygiene and Epidemiology (IHE) in Prague
--- tests mutagenic effects of new drugs, preservatives, pesticides, and new chemical compounds with a significant potential for broad use within the society; tests mutagenic activity of air both in the general environment and at workplace, eventually mutagenic potential of water sources, in case of existing grounds for suspicion of possible genetic risk;
--- provides assessment of genetic risk of chemical compounds using up-to-date methods (in accordance with internationally accepted methods)

a) of testing of microorganisms;
b) cytogenetic analysis of bone marrow of rodents and peripheral human lymphocytes;
c) using genetic tests that asses degrees of gene and chromosome (in F_1 generation) mutation in mammals;
d) testing of cell cultures (gene mutations, cell transformations);

— based on test results, this specialised section determines the degree of genetic risk of use of tested substances and recommends condition of their practical use (maximum acceptable concentration of compounds in air, ADI);
--- serves as a national reference laboratory for assessing the genetic risk potential of chemical substances and provides guidelines for the operation of regional hygienic stations and district hygienic stations, and pharmaceutical industry whose activities are relevant to these issues;
--- provides information on the seriousness of genetic risk of various parts of the environment to the departments of medical genetics of Type III HwPs and hygienic services (which may be relevant for example for a recommendation of abortion);
--- once a year issues a list of drugs and other chemical substances with mutagenic potential in humans;
--- keeps record on genetic risk of various environmental factors for humans in case of increased occurrence of mutagens (accidents and emergencies), and in collaboration with relevant regional hygienic stations guarantees the sampling of biological material and repeated examination of exposed persons;
--- collaborates methodically with similar departments in socialist countries (especially the Soviet Union and German Democratic Republic).

Appendix No. 2

Search for Persons with Congenital Developmental Disorders and Genetic Disorders and Their Registration

Search for persons with congenital developmental disorders (hereinafter 'CDD') and genetic disorders is carried out by doctors active in preventive medical care in collaboration with departments of medical genetics. Key role in finding them have HwP of Type I and Type II, but mainly healthcare districts where healthcare workers have the broadest contact with patients. The aim is to find and register individuals with genetic disorders and CDDs and individuals where suspicion thereof exists. District healthcare workers develop record of families with genetic disorders and families where such disorders are suspected, and utilise their knowledge of health history of the broader family. District gynaecologists use these records to recommend to women of fertile age a participation in specialised pre-conception care, and to pregnant women participation in specialised prenatal care. In in-patient care, systematic registration of genetic disorders and CDDs is carried out by gynaecology and obstetrics departments of hospitals and their newborn wards. They keep record of all foetuses and stillborn children with obvious malformations or malformations discovered during autopsy. Stillborn children with malformations are recorded using the SEVT 1433i form.

Same kind of records is kept of newborn children born with evidence of genetic disorders or suspicion thereof and of newborn children with CDDs. All live-born children are screened for phenylketonuria, eventually also other metabolic disorders. In collaboration with workers of other specialised services of HwPs, district paediatricians are required to inform the authorities of all children under 1 year of age born with a CDD or a genetic disorder.

Individuals registered in their healthcare districts or with doctors of specialised departments of Type II HwPs because of a genetic disorder or suspicion thereof are sent for specialist examination to select doctors with relevant specialties in Type II HwPs.

Collaboration with institutions of social care, regional national councils and their institutions (such as institutes of special care, marital and premarital counselling centres) is essential in the screening of the population for families with genetic risk.

Depending on the character of the genetic disorder, field sections of departments of medical genetics of Type II HwPs collaborate with relevant specialists in HwPs for more specialised testing on the level of Type III HwP.

Departments of medical genetics assesses the test results of individuals with genetic disorders or handicaps, provide them with further care, and recommend measures suitable from the genetics point of view to relevant doctors of preventive healthcare.

Marital and pre-marital counselling centres also participate in keeping record of individuals for the purpose of genetic prevention. It is intended that in the course of gradual integration of departments of medical genetics within regions, they should become a part of systematic preventive efforts.