MEDGEN

Developing Medical Genetics Education Through Curriculum Reforms And Establishment Of Postgraduate Training Programs

THE PROFESSIONAL CRITERIA OF THE CLINICAL GENETICIST IN ARMENIA

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General Information

The clinical geneticist carries out the professional activity in the field of Medical Genetics in hospitals, out-patient facilities and in the primary health care facilities.

Educational Requirements

The requirements for the clinical geneticist are the following:

- MD degree in General Medicine
- Residency in one of the following departments: Family Medicine, Internal Medicine, Pediatrics or being General Practitioner (Internatura until 1994)
- Additional fellowship in Medical Genetics

The possibilities of the official functions

The clinical geneticist could take the following positions:

1. Clinical Geneticist in the hospitals
2. Clinical Geneticist in the outpatient facilities
3. Head of the Medical Genetics Department
4. Director of the organization in the field of Medical Genetics
5. Member in the Disability Assessment Commission
6. Member in the Adoption Commettee

There is also possible to participate in the various activities and events conducted by the Ministry of Health (MOH) and other Social-Medical organizations.

General Knowledge

The knowledge in the constitution of RA, the health care legislation, working security rules, prevention and management principles of the hospital infections, first aid skills, and general awareness in the core/targeted programs of MOH.

General Skills

The skills in equipment safety
Support the first aid
The principles of the prevention and management of hospital infections

Profesional knowledge and skills

Clinical and genetical examination for the genetical diagnosis

Necessary skills	Collection of documentation related to the patients' health status and treatment, including: conducted interventions, prescribed medications, contraception and pregnancy histories, participation in the screenings and prevention activities, patients' socio-economic status, family status etc.
	Register the genetic history of the family using the standard genealogical tree method
	Written concept form for all procedures
	Taking photos for the clinical use
	Physical examination including differential analysis of the syndromes
	Additional clinical examinations or refer to the narrow specialist
Nessecary skills	Use the European and International guidelines for the practical work and improvement of the skills of medical geneticist
	Explore various dismorphological pecularities
	Formulate the correct clinical diagnosis and if necessary conduct additional examinations for the validation of the diagnosis
	Analyze the existing family, medical and personal history for the assessment of the genetic risk
	Substantiate all the modes and mechanisms of inheritance of the given genetic disorder
	Collect, assess and use the existing information on the given genetic disorder
	Create and register the complicated pedigree (or genealogical tree)
	Assess the validation of the primary source of the information, calculate the empiric risk or repetition risk of the genetic disorder
	Formulate the range of the differential diagnosis of genetic disorders
	Share and discuss unexplored cases with the colleges
	Ensure professional consultations to other clinics and hospitals
	Use the informational resources including the scientific websites, the genetic databases and statistical software
Necessary knowledge	The terminology of the Medical Genetics
	The known or likely genetic cause of the disease
	The modes of inheritance, Mendelian and atypical, the different modes of inheritance in the pedigree
	The assessment of the genetic risk
	The mechanisms of imprinting and repetitive triplet mutations, the molecular bases of the somatic mutations and the consequences of the somatic chromosomal abnormalities
	The physiological and pathophysiological bases of the genetic disorders
	The typical symptoms and signs of the genetic disorders, the genetic bases of the multietiologic diseases
	The ideasof the variable expression, penetration, pleiotropy, somatic and sexual mosaicism and *de novo*mutations
	The natural course of the genetic disorders
	The role of heredity in the development of the rare genetic disorders
	The role of the family history in the diagnosis of the rare genetic disorders
	The basic pediatric expressions and signs of the genetic disorders
Other characteristics	To respect the medical ethics during work with children and their parents (or their guardians), adults and their relatives
	The abilities to communicate with the patients with verbal, physical or mental disabilities
	To create an environment of mutual understanding and trust during the work with

	patients and their family members
	To ensure the protection of the patients' and their family members' personal data

Genetic testing and genetic diagnosis

Working activities	The written concept for the genetic testing
	Sampling of blood, oral mucosal cells and hair bulb, biopsy of the skin
	Using the general methods for the biochemical diagnosis of the inborn abnormalities
	The genetic testing and diagnosis of the cases with infertility, repeated spontaneous abortions and complicated obstetrics history
	Prenatal genetic testing
Necessary skills	To substantiate the choice of the particular method of the genetic testing for the given genetic disorder
	To provide to the patients the information about the benefits and risks of the genetic testing methods before getting the written concept for the mentioned manipulations
	To substantiate the best strategy of the genetic testing for the genetic disorder based on the existing medical and family history and results of the physical examinations
	To keep the working safety rules
	To analyze and explain the results of the genetic testing by taking into consideration the range of the sensitivity of the test
	To use the international guidelines of the cytogenetic and standard classification of the mutations
	To explain the clinical consequences of non normal karyotype, non normal nucleotide variability and enzyme deficiency
	To analyze and explain the results of the genomic tests
	To explain the results of the pharmacogenetic testing
	To analyze the clinical significance of the findings and if necessary to discuss with the specialists from the molecular and cytogenetic laboratories
	To implement Bayesian calculation for the assessment of the genetic risk using the genetic, genomic and non genetic data
	To substantiate the benefits of the new DNA-technologies and use them correctly
	To assess the significance of the genetic variability with uncertain significance
	To conduct the differential diagnosis based on the personal medical and family history, physical examination and laboratory findings by excluding the related diseases and phenocopies
	To explain the uncertainty and limitations of the genetic tests used during the screenings and for the diagnostic purposes
	To explain and discuss the results of the test to the proband and family members
	To use the European and International guidelines for the practical work of the medical-geneticist
Necessary knowledge	The conditions of the conducted genetic tests
	The difference between screenings and diagnostic tests
	The technical tools, risks, limitations and benefits, analytical and clinical accuracy and application of the conducted genetic tests
	The chromosomal analysis technique in the various tissues
	The standard and molecular cytogenetic testing technique
	The general molecular genetics technique
	The application and clinical significance of the new DNA technologies
	The differentiation between genetic and genomic tests
	The possibilities of the testing for the genetic predisposition for the

	multietiological diseases
	The implementation of the pharmacogenetic testing including the bases of the personal medical care based on the genomic peculiarities of the person
Other characteristics	To raise the awareness about genetic services and the existing methods related to the genetic health
	To take into consideration the ethical, legal and social aspects of the genetic testing

The primary genetic consultation after the genetic testing

Working activities	To plan the genetic consultation
	To present the results of the genetic tests and differential diagnosis
	To provide the results of the genetic/genomic tests, including the information about genetic disease, analysis of the pedigree and assessment of the genetic risk
Necessary skills	To conduct genetic consultation according to the plan
	To explain to the proband or to the person under the risk the clinical picture of the genetic diseases, the genetic bases of the disease the modes of inheritance, the results of the test and the assessment of the genetic risk
	To explain to the proband and to his/her family members the genetic and clinical information and the associated health risks
	To explain the importance of the secondary (not targeted) results of the genetic or genomic tests
	To ensure psychological assistance to the patients and their family members
	To use the European and International guidelines for the practical work of the medical-geneticist
Necessary knowledge	The principles of non-directly, non-guided genetic consultation
	The retrospective and prospective principles of the genetic consultation
	The factors affecting the process of genetic consultation, including the doubts and fears of patients and their family members, cultural, religious, ethnic factors, consanguineous marriages and prenatal diagnosis
	The identification of the family's role in the diagnosis of the genetic disorders
Other characteristics	To put attention on self-consultation methods and improve the quality in case of need
	To assist to the proband to share the results with the family members
	To work out the socio-psychological assistance plan for proband and his/her family members and solve the possible conflicts
	To raise public awareness in understanding of genetic diseases, tests and modes of inheritance

The follow up of the proband and the preventive testing of the family members

Working activities	To formulate the genetic inference about the further follow up of the proband and the expedience of the screening of family members, about the prognosis, the prescriptions, the modern approaches of the treatment and the further family planning
	To refer patients to the other specialist for the consultations
	To cooperate with the specialists for the follow up of children born with the metabolic abnormalities
	To conduct preventive testing for the family members, to explain the results and to formulate the further plan of actions
	To explain the results of prenatal tests results and to formulate the preventive plan
Necessary skills	To substantiate the further plans for patients follow up, as well as to plan the probable modes of screenings and follow up for family members in case of the

	following reproductive, prenatal, children and adult genetic diseases: Neuromuscular diseases Neurodegenerative diseases Mental and behavioral disorders Neurodermal disorders Inborn abnormalities Chromosomal syndromes Skeletal dysplasia Autoinflammatory syndromes Metabolic inborn disorders Genetic cancer syndromes Hematological disorders Renal diseases Cardiovascular diseases Hypercholesterolemia Endocrinologic diseases Mucoviscidosis Reproductive function diseases Multifactorial diseases Pharmacogenetic diseases
	To form and discuss the follow up plan for the proband with proband and his/her family members and with other people involved in the care
	To explain the importance of the early diagnosis of the genetic risk of the diseases and the influence of the environmental and behavioral factors on the genetic tests and genetic predisposition
	To formulate the mandatory conditions of treatment of metabolic inborn diseases for follow up (treatment directed to the biochemical changes) and medical care (including symptomatic treatment, other medical and surgical interventions)
	To formulate and plan the preventive prediagnostic examinations for the late developing neurodegenerative diseases
	To explain the role of germinal and somatic mutations (also in case of cancer) and the prescription of the necessary genetic test
	To discuss with the proband and his/her family members the details on the genetic disease, the risk of repetition and the possible ways of decreasing the risk
	To provide the genetic consultation to the patients with diagnosed genetic diseases and his/her family members and offer necessary care and follow up
	To substantiate the need of prenatal testing and the guidelines and restrictions of the tests
	To substantiate the possible ways of solution of the reproductive health issues through genetic counseling, prenatal testing or auxiliary reproductive technologies
	To interpret pregnant woman's blood test results and ultrasound examinations and to provide consultations on the possible normal and non normal results
	To interpret and simplify the existing medical errors caused by a lack of scientific evidence.
	To apply the European and International guidelines for the practical work of the medical-geneticist
Necessary knowledge	The factors that may have an impact on the process of genetic counseling, including fears of proband and family members, cultural, ethnic and religious factors, consanguineous marriages, prenatal diagnosis
	The direct and indirect principles of the genetic counseling
	The clinical and reproductive problems of the adult patients with inborn metabolic disorders

	The methods and technologies of prenatal genetic testing
	The conditions of special preventive genetic testing
	The various types of medical errors in the medical genetics
Other characteristics	To provide professional consultations to the colleagues involved in the treatment of the patient

The organization of activities of medical stuff, colleagues and other medical institutions

The working activities	The preparation of the individual working plan and reports
	The analysis of disability and mortality status among the attached patients
	The maintenance of accounting-reporting medical records
	The preparation of the necessary documents for the submission to the medico-social committee of assessment the disability status of the patients
	The management and follow up of the patient with the genetic disease in continuing contact with the consultant specialists
Necessary skills	To analyze the morbidity, disability and mortality status of the attached patients
	To fill the reporting documents and to supervise the quality of the medical documentation in the medical institutions covering the field of medical genetics
	To prepare the documents for the medico-social expertise for determining disability
	To present medico-statistical data for the report of the medical institution
	To work in the information-analytical system (The State Integrated Health Information System)
Necessary knowledge	The roles and responsibilities of the partners
	The role of the multi-profile work in the field of the clinical genetics
	The principles of the inter-professional cooperation in the process of the follow up and management of the patients and family members
Other characteristics	To ensure the safety of the medical data despite of the type of the medical supervision

The special knowledge and skills

The implementation and recording of the genetic screening

The working activities	The implementation of the screenings among the carriers of the common genetic disorders
	The recording of the personal and medical data in the registers
Necessary skills	To analyze and assess the individual risks of the patients
	To explain the sensitivity, specificity and preventive value of the screening tests
	To substantiate the results and benefits of the screening tests
Necessary knowledge	The principles of the genetic screenings and the prevention of the genetic diseases
	WHO guidelines and restrictions towards the implementation of the screening programs
	The possible application of the screenings for the diseases corresponding to the approved standards
	The genetic features and the epidemiology of the population, including the specter of the mutations, the spread and frequency of the diseases

	The existing international screening programs, including pre- and post-natal programs
	The national screening programs of the genetic diseases, the current results and benefits for the public health
	The practical use of the genetic register, methods and restrictions of the data collection
	The other factors that may have an impact on the implementation of the genetic programs
Other characteristics	The ethical, legal, social and cultural issues related to the genetic screening and pre-natal diagnosis

The differentiated application of the principles of individual data safety and ethics of medical genetics

The working activites	The process of receiving the consent form from proband or guardian (in case of children or persons with mental disabilities)
	The application of the safety mechanisms in recording the personal data in genetic registers
Necessary skills	To discuss with the proband until sharing the medical information among medical team members involved in the treatment
	To use and share the confidential information carefully
Necessary knowledge	The aspects of the medical genetics secrecy (legal, social, working, etc) and conditions of disclosure
	The consent form for the medical genetics data for using in the genetic registers and for scientific purposes
	The issues associated with post mortem study
	The ethical aspects of the consent form
	The main sources of counseling and legal aspects in criminal cases in the field of medical genetics
	The equality of services in medical genetics
Other characteristics	The discussion of the ethical issues with the proband, family members, colleagues and community