Russian - Armenian University

Program for continuing professional education course/ Fellowship in Genetic Counselling

The Program Overview:

The fellowship program in medical genetics and genetic counselling follows by education and training for fellows. Training will occur at the Center of medical genetics and primary health care (basic University educational center in medical genetics and genetic counselling).

Eligibility requirements: The training program is available for BA degree biologists and psychologists, medical school undergraduates who have completed full undergraduate University education.

Fellows/students assessment: *Test-based and oral examinations.*

CME Credits:90

The curricula during 1,5 years of continuing education in medical genetics/genetic counseling should be focused on:

∗0.5-year education in medical genetics (theoretical studies, clinical/laboratory training).

∗1-year education in genetic counselling (lectures, seminars, practical lessons).

Fellows will attend lectures, seminars, conferences, counselling of geneticists, clinical laboratories, laboratories of cytogenetics and molecular genetics. They will spend their time for training in genetics counselling.

0.5 ACADEMIC YEAR: SPECIAL COURSE OF MEDICAL GENETICS

Academic Credit Points: 30

Semester: 1st

Weekly: 30 h.

Total: 16 weeks.

Lectures: 16 weeks x 2 lectures = 32 h.

Seminars: 16 weeks x 6 = 96 h.

Practical lessons: 16 weeks x 2 lectures = 32 h.

Course background:

The training program for complete knowledge in medical genetics and additional medical disciplines is structured to include an initial 0.5 academic year in clinical and laboratory training in medical genetics, including dysmorphology, chromosomal and gene disorders, inborn errors of metabolism, cancer genetics, interpretation of cytogenetic, molecular genetic, immunological, biochemical, clinical laboratory results.

PROGRAM OF LECTURES, SEMINARS, PRACTICAL LESSONS

Human chromosomes

Lecture:

- Chromosomes and their abnormalities: nomenclature. Balanced and unbalanced abnormalities. Main reproductive outcomes for carriers of translocations/inversions. Copy number variants**.**

Seminar

- How can a patient's chromosomes be studied? Mechanism of X-inactivation.

Practical lessons in cytogenetics:

- Why clinicians need to know about chromosomes? Clinical consequences of abnormal karyotype.
- Case studies.

Human molecular genetics:

Lectures:

- Human genome, looking at noncoding DNA.
- Human genes structure. Regulation of gene expression in human cells.

Practical lessons:

- Gene mutations, de novo mutations. How can we check the DNA for gene mutations?
- DNA-based testing for mutation detection and linkage to the diseases.

Patterns of genetic transmission:

Lecture:

- Modes of inheritance: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked, mitochondrial, atypical patterns. Consanguinity.

Seminars:

- Signs and symptoms of genetic diseases, appropriate clinical measurements.
- Penetrance and expressivity, variable expressivity, reduced penetrance, somatic or gonadal mosaicism.
- Principles of epigenetics.
- Mechanism of imprinting and triplet repeat mutations. Correlation of trinucleotide repeat expansion size with disease penetrance and severity.

Practical lesson: Case study.

Dysmorphic syndromes and craniofacial disorders

Lectures:

- Genetics of dysmorphology and craniofacial disorders. Prevalence, penetrance, testing options, inheritance patterns. Normal developmental parameters, developmental delay, different clinical situations (deformation, malformation, disruption and dysplasia).
- Embriological abnormalities of congenital malformations.

Seminar:

- Phenotypes and diagnosis: use of databases for diagnostics of dysmorphology and craniofacial disorders.

Practical lesson: Case study.

Neurogenetics

Learning Objective: Assess and multidisciplinary management of patients with neurogenetic disorders.

Lectures:

- Genetic basis and clinical presentation of neurogenetic disorders. Different features of neuromuscular diseases in adults, children, infants, and neonates.
- Management of patients with neuromuscular and neurodegenerative disorders, epilepsy, mental and behavioral disorders, neurocutaneous and neurosensoral disorders.

Practical lessons:

- Clinical presentations of neurogenetic conditions, such as:
  - ✓ hypotonic neonate/infant ("floppy baby")
  - ✓ developmental delay (motor, cognitive and/or speech; global)
  - ✓ progressive weakness and/or ataxia
  - ✓ other movement disorders (dystonia, dyskinesia, tics, etc.)
  - ✓ dementia/cognitive decline
  - ✓ intellectual disabilities

Seminars:

- Appropriate investigations for suspected neurogenetic disorders, such as:
  - ✓ indications for serum biochemistry (CK, lactate, etc.)
  - ✓ electroneuromyography (incl. nerve conduction studies)
  - ✓ neuroimaging (MRI, CT)
  - ✓ muscle biopsy
  - ✓ molecular genetic analyses, including predictive testing (such as Huntington's disease)
- Management options available for childhood and adult onset genetic neuromuscular conditions.
- New approaches of gene therapy: example: Duchenne muscular Dystrophy and other disorders.

Autoinflammatory syndromes

Learning Objective: Assess and multidisciplinary management of patients with autoinflammatory periodic fever syndromes.

Lectures:

- Autoinflammatory and autoimmune disorders. Genetics of autoinflammatory periodic fevers. Phenotype-genotype correlation and clinical manifestation.

- Hereditary Periodic Fever Syndromes, Familial Mediterranean fever (FMF), TNF receptor-associated periodic syndrome (TRAPS), Hyperimmunoglobulinemia D with periodic fever syndrome (HIDS), Cryopyrin-associated periodic syndromes (CAPS), other monogenic autoinflammatory syndromes, multifactorial autoinflammatory disorders.

Seminars:

- Genetic factors influencing the risk of developing renal amyloidosis**.**
- MEFV gene mutations as the genetic factor for development of FMF**.** Diagnostic significance of MEFV mutations.
- Typical and atypical course and complications of FMF.
- Management, treatment and surveillance of patients with FMF and other autoinflammatory syndromes.
- Genotype-phenotype correlations and the effect of other possible modifiers such as other genes, gender and/or environmental factors.

Practical lessons:

- Interpret clinical and genetic test results.
- Be able to manage patients with FMF and refer to subspecialists
- Cases study.
- Treatmenet of FMF patients depending on the spectrum of MEFV mutations and clinical features.

Cancer Genetics

Learning Objective: Assess and contribute to the multidisciplinary management of patients with genetic cancer syndromes.

Lectures:

- Genetic mechanisms in cancer. Gene mutations and environmental factors of cancer risk.
- Clinical features of genetic cancer syndromes. Medical and family history features of inherited cancers.
- Breast and ovarian cancer arising from BRCA1 and BRCA2 mutations, colorectal cancer arising from familial adenomatous polyposis (FAP) or hereditary non-polyposis

colorectal cancer (HNPCC/Lynch syndrome), multiple endocrine neoplasia type 1 and 2 (MEN1 and MEN2), melanoma arising from CDKN2A or CDK4 mutations, Neurofibromatosis types 1&2.

Seminar:

- Monogenic predisposition to familial cancer.

Hematological and Immunodeficiency Disorders

Learning Objective: Assess and contribute to the multidisciplinary management of patients with hematological and immunodeficiency disorders

Lecture:

- Clinical features and genetic mechanisms of hematological and immunodeficiency disorders.Environmental factors that affect risk of developing hematological disorders.

Seminars:

- Use a hematological disease registers and other sources to verify diagnoses, to support follow-up of affected and at-risk patients.
- Discussion of strategy for cases: pregnant women - carriers of mutations responsible for hemophilia.

Practical lesson:

Case study.

Reproductive and prenatal genetics

Learning objective: Assess and management of patients with reproductive system disorders.

Lectures:

- Genetic basis of reproductive system disorders in children. Genetic abnormalities in adult patients.
- Role of genetics in the development and evaluation of infertility. Genetic causes of recurrent pregnancy loss.

Seminars:

- Cytogenetic and molecular testing, including clinical laboratory analyses for diagnostics of disorders of reproductive system.

- Indications for prenatal diagnosis. Prenatal care including preconception counselling regarding substance abuse, nutritional supplementation (particularly folic acid), screening for infection, dating of pregnancy with monitoring of fetal growth, maternal serum marker screening and fetal ultrasound.
- NIPT.
- Differences between and appropriate uses of prenatal genetic screening and prenatal genetic testing.
- Techniques for prenatal diagnosis: chorionic villus sampling (at 8-10 weeks of pregnancy), fetal sonography, amniocentesis (at 14-15 weeks of pregnancy), cordocentesis (at 20-30 weeks of pregnancy).
- Reproductive technologies for lowering genetic risks including sex selection by sperm fractionation or preimplantation analysis and *in vitro* fertilization with preimplantation genetic diagnosis (PGD).

Practical lesson:

- Cases study: obtain a pertinent history and relevant medical information needed for case management.

Inborn Errors of Metabolism

Learning objectives: Assessment and multidisciplinary management of patients with Inborn Errors of Metabolism.

Lectures:

- Molecular and biochemical consequences of a primary enzyme block in a metabolic pathway. Genetic basis of IEM, types of inheritance of IEMs and early clinical sign.
- Types of IEM: organic acidemias, fatty acid oxidation defects, primary lactic acidosis, aminoacidopathies (phenylketonuria, tyrosinemia, nonketotic hyperglycinemia, maple syrup urine disease, homocystinuria), disorders of carbohydrate metabolism (galactosemia, hereditary fructose intolerance, fructose 1,6-diphosphatase deficiency and glycogen storage diseases), lysosomal storage disorders (mucopolysaccharidosis, Tay-Sachs disesase, Niemann-Pick disease, Gaucher disease).
- Neonatal screening strategy for IEMs

Seminars:

- Appropriate investigations for suspected IEM. Genetic testing and counselling in clinical practice of IEM.
- Recognize the different features of IEM.

Other Genetic Disorders

Learning objectives: Assessment and multidisciplinary management of patients with other genetic disorders and diseases.

Lectures:

- Genetics, pathogenesis and clinical features of mitochondrial diseases (excluding mitochondrial myopathies).
- Inherited cardio-vascular disorders (arrythmogenic right ventricular dysplasia (ARVD), familial dilated and hypertrophic cardiomyopathy, familial heart disease, long QT syndrome, Loeyz-Dietz syndrome).

Seminars:

- ✓ Inherited renal disorders (e.g. polycystic kidney disease)
- ✓ Inherited skin disorders (e.g. ichthyosis, ectodermal dysplasia, Ehlers-Danlos syndrome, albinism, epidermolysis bullosa, incontinentia oigmenti, tuberous sclerosis)
- ✓ Inherited endocrinology disorders (diabetes insipitus, diabetes mellitus).
- ✓ Inherited sensory disorders (including deafness and ophthalmological disorders).
- ✓ Inherited amyloidosis
- ✓ Metabolic syndrome
- ✓ Inherited dyslipidemias
- ✓ Marfan syndrome

SPECIAL COURSE OF GENETIC COUNSELLING

Academic Credit Points: 60

Weekly hours:30

Semesters: 2,3

Total: 32 weeks.

Lectures: 32 x 2 lectures = 64 h.

Seminars: 76 x 2 h.

Practical lessons: 32 x2 h.

Course background:

The training program for full qualification in genetic counselling is structured to include one academic year of training in interpretation of pedigrees, risk assessment and communication skills, counselling topics to prenatal invasive and non-invasive testing, care, management of patients and their families. The purpose of the genetic counselling is to respond to the persons and families wishing to know about the risk of developing or transmitting genetic disorder, information on possible preventive measures and social and psychological consequences for disable patients and family members. Education should be provided to allow the fellows to be able to communicate with probands and family members.

Lectures:

- Basics of genetic counselling and principles of communication.
- Role and functions of a genetic counselor.
- Prenatal genetic counselling and screening.
- Psychological impact of genetic tests.
- Practical aspects of consent, confidentiality and disclosure.
- Multicultural education and genetic counselling. Challenges of cross-cultural interpretations.
- Clinical judgement, timing, and professional language when informing parents of an infant with serious malformation or developmental delay.
- Communication strategies to: facilitate and engage in realistic discussion with respect, empathy, and sensitivity. Assist patients to consider their available choices and to make decisions, identify role of family with genetic diagnosis.
- Family history and pedigree drawing.
- Counselling techniques in genetics evaluation and case management.
- Educate patients about their condition, use of lifestyle modifications and psychosocial impact of the disease.
- Principles of presimptomatic genetic testing.

- Pre-test and post-test disclosure and counselling
- Taking a relevant bleeding history (previous challenges and family history) with a focused clinical examination.
- Genetic counselling in reproductive pathologies.
- Practical genetic counselling in dysmorphology.
- Conditions with prenatal findings (spina bifida, hydrocephalus, cerebral palsy, Down syndrome).
- Genetic counselling for patients with FMF, carriers and at-risk asymptomatic family members.
- Impact of consanguinity in developing FMF and other genetic disorders.
- Counselling for predisposition to familial cancer.
- Discussion of possibilities of multidisciplinary rehabilitation, management, and care for neuromuscular disorders.
- Genetic counselling in mental retardation and learning disorders.
- Describe current recommendations concerning cardiac surveillance in families with inherited heart diseases.
- Recognize the medical presentations and family histories that indicate a risk of IEM.
- Counsel patient and their family about specific conditions and their impact on long term prognosis of IEM.
- Discuss possibilities of multidisciplinary rehabilitation, management, and care for IEM.
- Identify population-based genetic registers and their practical issues.
- Describe the principles of prevention and screening.
- Genetic counselling and carrier testing in children
- Counselling of patients with disorders with non-traditional inheritance.

Seminars:

- Presentation and discussion cases with colleagues.
- Assess patient's psychological state.
- Decision-making in a non-directive manner
- Confidentiality and security of written and verbal information.

- Issues regarding pregnancy, chronicity, death and loss, as well as the impact of cultural issues.
- Describe the genetic characteristics in local populations, including mutation frequencies and disease prevalence.
- Predictive genetic testing.
- Prenatal/pre-implantation diagnosis.
- Prepare patient and family for testing and offer post-testing counselling and support.
- Describe contributing factors for the establishment of population screening programs for genetic diseases.
- Discuss the public health outcomes and benefits of screening programs, including the costs.
- Identify population-based genetic registers and their practical issues.
- Describe the principles of prevention and screening.
- Discuss ethical, legal, social, and cultural issues associated with genetic screening programs.
- Patients` interpretation of risks offered in genetic counselling.
- Educate patients about their condition, use of lifestyle modifications and psychosocial impact of the disease.
- Counselling of pair about specific conditions and impact on long term prognosis of reproductive system disorder. Information on reproductive and prenatal diagnosis options.
- Prepare patient and family for testing and offer post-testing counselling and support. Help patients to deal with uncertainty regarding the diagnosis and prognosis of the fetus.
- Comprehend principal qualitative, quantitative, bio-statistical and epidemiological research methods utilized in genetic counselling and describe their use.
- Social and emotional aspects of genetic counselling. The emotional effects of genetic diseases.
- Outcomes of mutation positive test result.
- Outcomes of mutation negative test result.
- Passing on genetic information from parents to children.

Practical lessons:

- Draw a pedigree using correct symbols.
- Pedigree interpretation and use a pedigree to support diagnosis and a genetic risk assessment.
- Inheritance patterns in pedigrees.
- Describe genetic diagnosis, clinical features, prognosis of the disease.
- Provide genetic counselling of proband and/or family members.
- Identify the mode of inheritance and define the penetrance and expressivity
- Provide the indications for genetic testing.
- Genetic testing of children.
- Counselling of pregnant women at late termination of pregnancy.
- Prenatal diagnosis counselling scenarios.
- Perform risk calculation for a genetic condition in fetus**.** Interpret indicated tests of specific disorder in a pediatric and adult patient for case management.
- Provide information on IEM prenatal diagnosis options.
- Population screening for genetic diseases.
- Medical presentations and family histories that indicate a risk of familial neurological disease.
- Differential diagnosis, appropriate genetic testing (including prenatal testing) of neurological disorders.
- Measures that can be used to prevent or ameliorate cancer in individuals with increased inherited risk. Clinical screening measures that might be offered to detect cancer at an early stage.
- Screening protocols for at-risk relatives of cancer patient.
- Familial cancer counselling scenarios.
- Taking a relevant bleeding history of cancer/hematological disease (previous challenges and family history) with a focused clinical examination.
- Explain the molecular and cytogenetic testing results.
- Communicate genetic information.
- Interpretation of laboratory analyses.

- Interpret medical records, laboratory and instrumental exams of children with developmental delay and/or dysmorphic syndromes.
- Assess an individual patient's risk factors.
- Ethical situations.
- Counselling scenarios in Assisted Reproductive Technologies.

Professional exercises:

1.Giving news:

Think of a time when you gave someone bad news.

Think of a time when you received some bad news.

2.Breaking news scenarios.

3. Discussions of counseling skills: skill practice by role play.

4. Discussions of difficult cases.

5. Skill practice by role play on prenatal with uncertain outcome and group discussion.

6. Clinical and laboratory testing, reporting, disclosing news.

7.Risk-based decision making.

8.Transactional analysis.

Dealing with Medical Uncertainty

- **Learning Objective: Identify and discuss factors contributing to uncertainty in medical practice**

Lecture:

- Uncertainty as an integral part of all medical practice, including clinical genetics, genetic counselling. Uncertainty is a limitation of current knowledge and not a professional failure

Seminars:

- Disclose and openly acknowledge areas of uncertainty to patients. Reframe uncertainty by breaking it down into manageable parts.
- Assist in the patient's decision making process to reach an acceptable conclusion.
- Develop a solid basis of accurate knowledge, including literature search and consultations with colleagues

- Recognize that uncertainty in current clinical genetic knowledge can provoke anxiety in the patient and the clinician
- Discuss many types of uncertainty in medical practice, including:
  -Antenatal detection of abnormality with uncertain consequences for the fetus
  -Lack of diagnosis, particularly in person with intellectual disability, malformations or dysmorphism leading to uncertainty about cause, prognosis and recurrence risks
  -Known diagnosis but uncertain prognosis
  -Absence of prenatal tests leading to uncertain outcome for current or future pregnancy.

Bioethics

- **Learning Objective: Identify, understand and apply appropriately the principles, guidance and laws regarding medical ethics and confidentiality.**

  Lecture
- Ethical issues in reproductive medicine including parental-fetal conflict of autonomy and beneficence, timing of fetal humanity, confidentiality versus rights to informed choices, and equal access to expensive medical resources

  Seminars/discussions:
- Discuss relevant strategies to ensure confidentiality. Identify situations where confidentiality might be broken
- Discuss different cultural attitudes to genetic disorders and genetic testing and their impact on individuals, families and cultural groups.
- Recognize the factors influencing ethical decision making: including religion, personal and moral beliefs, and cultural practices
- Discuss privacy, consent and access issues in relation to the creation and use of genetic registers.
- Discuss the importance of genetic registers in research and management of genetic conditions.
- Differentiate between the legal and personal requirements of the patient's medical record- and that of the genetic register.

- Summarize the diversity of public opinion on ethical and moral aspects of the practice of clinical genetics.
- Consult with patients before distribution of information within members of the immediate healthcare team
- Communicate the value of genetic registers to colleagues, patients and their families. Provide patient details to the appropriate genetic registers.
- Know the principles of gaining regulatory approvals for genetic counselling and the role of research ethics committees.
- Discuss the importance of ethical approval and patient consent in medical practice.
- Follow guidelines on ethical conduct in research and consent for genetic counselling.

Informed Consent

- **Learning Objective: Ensure the trainee has the knowledge and skills to deal appropriately with ethical and legal issues that arise during the management of patients with genetic disorders.**

Seminars:

- Disseminate appropriate information in lay terms and gain informed consent from patients. Discuss the process for gaining informed consent
- Understand process of consent for tissue/sample storage and use
- Understand the process of consent for non-invasive activities, such as sharing information with relatives
- Understand how to gain consent in a research context.

Practical lessons:

- Present all information to patients (and careers) in a format they understand and allow time for reflection on the decision to give consent
- Respect a patient's rights of autonomy
- Show willingness and ability to obtain a second opinion, senior opinion, and legal advice in difficult situations of consent or capacity

LEARNING OUTCOMES FOR GENETIC COUNSELLING

Four broad learning outcome areas in genetics, in which it is proposed that a graduate genetic counsellor should attain by the end of their education, are shown below:

1: Communication skills

2: Professional skills

3: Counselling skills

4: Professional ethical values and skills.

1: Communication Skills

After completion of the programme, the graduate can:

- Establish a mutually agreed upon genetic counselling agenda with the client
- Establish empathetic relationship with the client and identify client's concerns and needs
- Obtain and record an appropriate and complete family history
- Support client to make informed choices concerning actions they may take, relevant to the implications of their family history
- Obtain relevant medical information including, but not limited to, pregnancy, developmental, medical and psychosocial histories
- Explain the technical and medical aspects of diagnostic and screening methods, as well as treatment/management of genetic conditions and/or birth defects to clients with different educational, socioeconomic and ethno cultural backgrounds
- Manage a genetic counselling case in a culturally responsive manner.
- Document and present case information clearly and concisely, in both oral and written formats.

2: Professional skills

After completion of the programme, the graduate:

- Has a deep understanding and knowledge of genetics and genomics concepts and principles
- Has an understanding of counselling theory in relation to genetic healthcare.
- Can identify and integrate modern genetic testing options used in genetic counselling.
- Can understand and explain the patterns of inheritance and the underlying mechanisms by which a genetic condition occurs.
- Can understand and provide clinical information as well as treatment options to the clients and their families.

- Have knowledge of reproductive organs and systems and general human anatomy.
- Makes appropriate and accurate genetic risk assessment.
- Can construct relevant and comprehensive personal and family histories and pedigrees
- Can assess client's understanding guide counselling sessions as needed and flexibly response to the provided information.
- Can critically assess genetic, medical literature and records.

3: Counselling skills

After completion of the programme, the graduate can:

- Use a variety of consultation skills
- Use counselling techniques in relation to genetic health state in an appropriate manner
- Provide client-centred counselling and mental assistance
- Support client's decision-making in a non-directive, non-coercive manner
- Evaluate and act by referral to another healthcare practitioner if indicated
- Be engaged constructively in peer supervision
- Establish and maintain interdisciplinary professional relationships and function as part of a healthcare team.

4: Professional ethical skills and values

After completion of the programme, the graduate:

- Can act in accordance with the ethical, legal, and philosophical principles and values of the genetic counsellor profession.
- Has an understanding of ethical principles relevant to genetic healthcare (WMA Helsinki Declaration).
- Can integrate ethical principles into counselling practice.
- Can maintain confidentiality and safety of client and obtained sensitive information.
- Should demonstrate initiative for continued professional development.

References:

- Core competences for genetic counsellors, European Board of medical genetics, June 2010
- European Core Curriculum for the Master programme in Genetic Counselling
- Code of professional practice for genetic counsellors in Europe, European Board of medical genetics, June 3013
- The American Board of Genetic Counseling , Genetic Counselors' Scope of Practice, June 2007