MEDGEN

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

Student Learning Outcomes for MSc Program in Medical Genetics
And Graduate Profile for Residency Training Program in Clinical Genetics
at Yerevan State Medical University after Mkhitar Heratsi

Policy

Rapid progress made in the field of medical genetics has led to an increased demand for genetic services. There is a lack of trained medical geneticists and health care professionals to deliver the education and training in Armenia.

In 1997, a comparative study of genetic services in Europe by the Concerted Action of Genetic Services in Europe found genetic service practices and facilities of different countries varied considerably. In the same year, the European Society of Human Genetics (ESHG) embarked on a project to develop professional guidelines on various aspects of human genetics. In 2001, the European Society of Human Genetics recommended a formal recognition of medical genetics as a medical speciality in Europe. One aspect of the work was the production of guidelines for genetic services in Europe, published in the European Journal of Human Genetics in 2003 which involved the definition and the aims of genetic services, their organisation, their quality and the role of public education. A European Union funded EuroGentest project addressed some of the challenges of the genomic era by encouraging the creation of a European Network of Excellence on genetic testing. The main objective was to harmonise and improve the quality of European genetic services, covering all aspects of genetic testing, including education. In 2010, the Expert group of the EuroGentest project described and agreed a set of core competences that could apply to health professionals in Europe, whatever their national setting which could provide an appropriate framework for establishing minimum standards of preparation for health care professionals in genetics across national boundaries. In 2010, the Committee of Ministers of the EC developed

recommendations to member states on the impact of genetics on the organisation of health care services and training of health professionals. Armenia joined and signed this memorandum in 2010.

As a result of these recommendations, YSMU have restructured its teaching and regardless of which forms of teaching they have introduced and have reconsidered the essential core elements required by all Medical Graduates.

Background

This 2014 document has recently been updated by a new version of "Core Competences in Genetics for Health Professionals in Europe" published in 2010, and "Recommendation CM/Rec(2010)11 of the Committee of Ministers to member states on the impact of genetics on the organisation of health care services and training of health professionals" also published in 2010. The main recommendations in this document which relate to the teaching of Medical Genetics include:

- Appropriate education and training should be provided for all healthcare professionals. From the results of a comparative study of genetic services in Europe it became clear that comparison of genetic education was complicated by the variety of health care systems, organisational structures in health professional education and health professionals involved at first patient contact. One of the main recommendations of the study was the provision of a joint education and training programme to promote teaching and training programmes in medical genetics to medical and other students in related fields with assessment for specialists, medical geneticists and other health care workers. Besides the scientific and medical education, social, legal and ethical aspects of medical genetics should also be taught. Medical genetics should not be regarded as one of the sub-specialities which might not be assimilated by the health care professional but as a core component of training of all health care professionals. In 2001, the European Society of Human Genetics recommended a formal recognition of medical genetics as a medical speciality in Europe.

- During preclinical education both lectures and practical courses of genetics should be delivered by an instructor who is a specialist in medical genetics. The basic training in genetics should be directed towards the application of genetics in medicine. The clinical education should include lectures and training in small groups of students that allow for interaction between the teacher and the student.

- The essential knowledge and skills required by the medical geneticist include the ability to:
  establish a diagnosis;
  - ✓ interpret the role of the family history and assess the mode of inheritance;
  - ✓ know the indications for and how to interpret the results of genetic tests;
  - ✓ evaluate risk for the individuals and family relatives;
  - ✓ provide information on possible procreation choices;
  - ✓ discuss long term disease outcome.

- Education and training should be provided in order to allow the medical geneticist to be able to communicate genetic information to enable patients to make informed decisions about themselves and where appropriate to raise awareness about possible implications for family members.

- Education and training should be delivered in order to allow the primary care provider (general practitioner) to have knowledge of the most common chromosome abnormalities and monogenic disorders that are prevalent in the population concerned. This should include risk assessment, patient communication, a basic knowledge of pharmacogenomic developments, and the capacity to interface with other levels of care relevant for genetics, i.e. genetic labs, tertiary care. The general practitioner should also be aware of the indications and availability of predictive and prenatal diagnostics in order to adequately refer patients and their relatives to more specialised

services when appropriate. Courses on basic medical genetics should be accessible to those in primary care physician training.

- Institutions with medical geneticists and with a sufficient density of cases should offer education programmes in medical genetics. Curricula should be harmonised within Europe and preferably be of 5 years duration. Specialists in medical genetics must be competent in genetic diagnosis but must also be proficient in genetic counselling, and in the management and care of patients and their families.

- The genetic laboratory scientist is responsible for genetic testing and for producing results of laboratory studies for the clinician to examine. The genetic laboratory scientist should be aware of the significance and the implications of the results of genetic tests. His/her curriculum should include basic medical genetics in parallel with specific technological education.

- The curriculum of the genetic counsellor should include basic genetics, interpretation of pedigrees, risk assessment and communication skills.

- The curriculum of specialists in other medical fields, for example internal medicine, paediatrics, neurology and ophthalmology, should ensure familiarity with clinical diagnosis and management of genetic disorders relevant for the respective speciality. During the specialist's training there should be an opportunity to spend some time, e.g. six months, under the supervision of a medical geneticist in order to be acquainted with principles of the genetic disorders related to the medical speciality.

- Specialist nurses may play an important role if trained in medical genetics. Training should be available to nurses who are able to provide basic genetic counselling and support management for the affected individuals and their families.

- Practising clinical geneticists, genetic scientists and other health care professionals should receive continuing education in medical genetics. Continuing education should also equip primary care clinicians to provide genetic services and to enable these clinicians to recognise when referral to a medical geneticist is warranted. There is a need to secure a sufficient number of teaching personnel to enable medical geneticists and health care professionals to keep abreast with this rapidly expanding field.

This document takes these recommendations into account as well as needs of national stakeholders when considering the most appropriate material for inclusion in a core curriculum for Medical Genetics for undergraduate students and Clinical Genetics for medical residents for training in Clinical Genetics.
This paper provides a recommended core basic science and clinical curriculum for the teaching of Medical genetics to medical students.

As a subsequence, many departments of YSMU related to the subject of Medical Genetics have been asked to revise their core curriculum and skills deemed to be appropriate for the teaching of Medical Genetics to students.

The learning outcomes are based on the reconstruction of the undergraduate and postgraduate medical curriculum into a core curriculum comprising essential knowledge, skill, and attitudes to be acquired by all medical students together with a series of special study modules, which allow students to study medical genetics. The aim of this document is also to promote self-learning and improve the educational effectiveness of the undergraduate course of Medical Genetics and residency program of Clinical Genetics.

Learning Outcomes for the two-week course of Medical Genetics for all undergraduate medical students

Upon completion of a two-week program of Medical Genetics, undergraduate students will have acquired the following competencies relevant to Medical Genetics:

1. To be able to take a family history and construct and interpret a family tree from a verbal description.

2. To be able to recognize all forms of Mendelian inheritance, consanguinity and founder effects.

3. To have a clinical knowledge of the most important Mendelian and chromosomal conditions, stating usual mode of inheritance, major features, complications, and usual diagnostic tests. To be able to understand the implications of balanced and unbalanced chromosomal translocations and mircodeletions and the methods that can be used to detect them.

4. To have a clinical knowledge of the genetic factors associated with cancer predisposition by identifying characteristics of a family history which suggest the presence of a familial cancer syndrome.

5. To be able to recognize the genetic and environmental contribution to multi-factorial conditions and the relevant management and population health issues.

6. To be able to understand approaches which can be used for the diagnosis of genetic disease and carrier detection, including family history, ethnic background, clinical phenotype and the role of laboratory testing.

7. To understand different forms of DNA testing including prenatal, predictive, diagnostic, and pharmacogenetic testing.

8. To have a knowledge of molecular genetic and cytogenetics testing techniques and to be able to interpret genetic test reports.

9. To be familiar with the practice of the genetic counselling clinic, its aims and methods including the principals of non-directive, non-judgmental counselling and impact of genetic diagnosis on the extended family.

10. To know when and where to get help and information from clinical geneticist, and where to find further information.

11. To know what are considered as major ethical issues relating to the use of genetic information and procedures.

Learning Outcomes for the two-year residency training program of Clinical Genetics

The practice of Medical Genetics is based on an in-depth knowledge of basic genetic principles, knowledge of genetic disease as it affects all body systems and individuals of all ages, and a clear understanding of the principles of genetic counselling. Upon completion of a two-year residency program of Clinical Genetics, residents will have acquired the following competencies and will function effectively as a clinical geneticist, integrating medical knowledge, clinical skills, and professional attitudes in their provision of patient-centered care.

Effective, ethical and patient-centered medical care

1. Performing a consultation, including the presentation of well-documented assessments and recommendations from another health care professional

2. Recognizing and appropriately responding to relevant ethical issues arising in the care of patients and their family members

3. Demonstrating patient- and family-centered care

Clinical knowledge and skills appropriate to Medical Genetics

4. Appling clinical and biomedical knowledge relevant to Medical Genetics:

- Principles and application of cytogenetic, molecular genetic, and genomic techniques
- Principles of biochemistry applicable to Medical Genetics
- Monogenic and complex inheritance
- Developmental biology related to normal and abnormal human morphogenesis
- Principles of biostatistics, genetic epidemiology, population genetics
- Indications, limitations and risks of techniques of prenatal assessment and options for reproductive intervention
- Genetic and non-genetic factors predisposing to fetal loss, infertility, and abnormalities of morphogenesis
- Teratogenic agents and their effects
- Methods of syndrome identification and diagnosis, including the use of computer diagnostic programs
- Clinical features, etiology, diagnosis, management, natural history, and prognosis of well-defined genetic syndromes and diseases
- Principles and approaches of screening for genetic disorders
- Distinction between genetic testing for the diagnosis of disease and predictive testing to assess risk for predisposition to monogenic or complex genetic diseases

5. Contributing to the enhancement of quality care and patient safety by integrating the available best evidence and best practices

Complete and appropriate assessment of patients

6. Eliciting a comprehensive medical and family history

7. Performing a comprehensive physical examination relevant for prevention, diagnosis and/or management of genetic conditions, including application of special expertise in dysmorphology

8. Demonstrating effective clinical problem solving and judgment to address patient problems

9. Taking the appropriate steps for syndrome identification, including the interpretation of laboratory and imaging findings relevant to genetic diseases, available databases, electronic and Internet sources.

Effective preventive and therapeutic interventions

11. Implementing an appropriate management plan

12. Demonstrating appropriate application of primary, secondary and tertiary preventive interventions relevant to Medical Genetics

13. Providing appropriate application of therapeutic interventions relevant to Medical Genetics

Appropriate consultation from other health professionals

14. Providing effective and appropriate consultation of another health professional for optimal patient care

15. Arranging appropriate follow-up care services for patients and their families with other health professionals

The Student Learning Outcomes for Master and Residency Training Programs at Yerevan State Medical University after Mkhitar Heratsi (YSMU) are prepared by the Department of Medical Genetics of Yerevan State Medical University after Mkhitar Heratsi (YSMU) in collaboration with relevant specialists from University of Bologna (Italy), University of Plymouth (UK), Charles University in Prague (Czech Republic), Medical University of Graz (Austria), and University Paris Descartes (France).

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This project has been funded with support from the European Commission:
Grant Agreement 544331-TEMPUS-1-2013-1-AM-TEMPUS-JPCR
This publication reflects only the author's view and the Agency and the Commission are not responsible for any use that may be made of the information it contains.