

MEDGEN

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

Student Starting Profile for Undergraduate
Medical/Biological Students
for Learning of Medical Genetics



TEMPUS

The developed student starting profile focuses on the restructuring the undergraduate medical curriculum into a core curriculum comprising essential knowledge, skills and attitudes to be acquired by all undergraduate students to allow them to continue education in Medical Genetics during graduate education. The aim of the student starting profile is to improve the effectiveness of undergraduate medical/biological education being as a prerequisite and core elements for further teaching of Medical Genetics to all graduates. Much of this material should be covered in the early years of undergraduate education to facilitate an understanding and place of genetics in modern medical practice.

Rapid development in molecular biology and medicine emphasizes the importance of ensuring that undergraduate medical students are familiar with the principles of human genetics and their applications in preventive medicine. There is an unambiguous and pressing need for the establishment of careful coordinated and clearly delineated teaching of basic genetics and all relevant disciplines in medical schools.

The aim of the current student starting profile is:

- ✓ to identify an essential core of genetic material to be taught to undergraduate medical students;
- ✓ to select a core of material which would be required by every student to take a graduate course of Medical Genetics;
- ✓ to select material for inclusion where lack of the knowledge or relevant skills would lead to inability to have an appropriate education in Medical Genetics;
- ✓ to select material for teaching which will allow and facilitate future professional development.

BASIC GENETICS

Four learning outcomes in basic genetics are proposed which medical students should attain by the end of their undergraduate education consisting the student starting profile for the teaching of Medical Genetics to graduates.

By the end of undergraduate education, the medical student will:

Understand and describe the mechanisms that underpin human inheritance

Be able to describe the structure of DNA and outline of its function as genetic material (replication, transcription and translation);

Be able to describe gene structure, expression and regulation;

Be able to describe general features of the human genome (amount of DNA, number of genes, organisation of chromosomes, repetitive DNA, amount of inter-individual variation);

Be able to describe the chromosomal basis of inheritance and how alterations in chromosome number or structure may arise during mitosis and meiosis;

Be able to describe modes of inheritance (Mendelian and non-Mendelian);

Describe the basic organisation of the cell

Understand the basic structure and functions of the cell

Be able to describe cellular processes: growth (cell cycle), metabolism, cell specialization;

Be aware of two distinct types of cell division: mitosis, meiosis, and gametogenesis;

Have an understanding of the role of genetic factors in health and disease development

Basic understanding of genetics (genotype, phenotype, allele, heterozygosity, homozygosity, penetrance, expressivity, pleiotropy etc.);

Understand how mutations can affect gene dosage and function;

Understand how mutations cause partial or complete loss of function or gain of function;

Understand the use of polymorphisms as genetic markers;

Be aware of the role of genetic and environmental factors in multifactorial conditions;

Be aware of gene frequencies of common recessive mutations;

Understand the main principles of genetic processes in the population, founder effect, gene drift, inbreeding;

Be aware that population ancestry may affect the frequency of susceptibility alleles and of Mendelian diseases;

Identify patients with or at risk of a genetic condition

Be able to take a family history and construct and interpret a genetic pedigree using standard symbols

Be able to recognize inheritance patterns, consanguinity, and founder effects based on the family history of disease and the family tree

OTHER BIOMEDICAL DISCIPLINES

Here we summarize the main issues which undergraduate students need to know or have an understanding from different biomedical disciplines which are interconnected with the subject and teaching of Medical Genetics:

Biochemistry

Main biochemical criteria in normal and pathological states
Modern methods of biochemical research and their use in the clinic

Embryology

The critical stages of organogenesis
Role of teratogens and their effects

Immunology

Abnormalities of the innate and adaptive immune systems
HLA system
Mechanisms of the development of inflammation, autoinflammation and allergy

Oncology

Molecular and hereditary bases of cancer and cancerogens

Pharmacology

Bases of pharmacokinetics and pharmacodynamics
Cell and Molecular Pharmacology
Cellular Basis of Drug Dependence

Cardiology

Specificities of the structure and the function of cardiovascular system in children
Cardiomyopathy and rhythm disturbance
Congenital abnormalities of cardiovascular system
Abnormalities of lipidic metabolism and atherosclerosis
Multifactorial diseases such as ischemic disorder, metabolic syndrome, hypertension
Modern diagnostic methods of cardiovascular diseases and their use in clinic

Nephrology and Urology

Specificities of the structure and the function of urogenital organ system in children

Congenital abnormalities of urogenital organ-system

Nephrotic syndrome (including amyloidosis) and nephritic syndrome (including glomerulonephritis)

Modern diagnostic methods of urogenital diseases and their use in clinic

Obstetrics and Gynecology

Normal and pathological courses of pregnancy

Pregnancy and newborn screening programs

Invasive and non-invasive methods of examination of the fetus

Normal and pathological courses of the delivery

Pediatrics

Stages of psychomotor and physical development of children

Specificities of physical and sexual development of teenagers

Endocrinology

Congenital and evoked abnormalities of the function of the thyroid gland, disorders of the parathyroid gland, and the calcium metabolism

Diabetes mellitus and diabetes insipidus

Congenital and evoked adrenal diseases

Abnormalities during the sexual development

Modern diagnostic methods of endocrine diseases and their use in clinic

Ophthalmology

Changes of the eye grounds during different disorders

Affections of the optical nerve

Common structural and functional abnormalities of different parts of the eye

Skin disorders

Types of skin lesions and their mechanisms of development

Histology

Modern examination methods of tissues and their use in clinic

Hematology

Specificities of the blood elements in children
Bone marrow and hemopoiesis
Blood groups
Anemias, leukemias, thalassaemias,
Abnormalities of coagulation mechanisms, and thrombophilia
Modern methods of blood analysis and their use in clinic

Stomatology

Congenital abnormalities of the organs of the buccal cavity

Orthopedics

Common structural changes and congenital diseases of the skeletal system
Modern methods of examination of skeletal system and their use in clinic

Pulmonology

Specificities of the structure and function of the respiratory system in children
Congenital abnormalities of the respiratory system
Bronchial asthma

Gastroenterology

Specificities of the structure and function of the gastrointestinal tract in children
Common congenital abnormalities of the gastrointestinal tract
Abnormalities of the absorption function in children
Cystic fibrosis
Jaundices
Modern methods of examination of gastrointestinal system and their use in clinic

Psychiatry

Bases of medical psychology
Schizophrenia
Delay of intellectual development
Intellectual disability

Neurology

Specificities of the nervous system in children
Main types of neuromuscular disorders
Seizure syndrome, epilepsy, ataxias
Congenital abnormalities of the Central Nervous System
Modern methods of examination of the nervous system

Otolaryngology

Hearing impairment
Modern methods of examination of the nose, the throat, and the ear

Required Skills

Requesting of the anamnesis

Physical examination, including anthropometric examination, palpation, auscultation, and testing of reflexes

CONCLUSION

This paper provides a recommended core medical and genetic science issues for teaching basics of genetics and several biomedical disciplines to undergraduate medical students, and it serves as a student starting profile to upgrade the knowledge and skills of students during future learning of Medical Genetics.

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.