

# 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.