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MEDGEN

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

CONTENT OF UNDERGRADUATE COURSE OF MEDICAL GENETICS

The aim of new program: improvement of education and training of medical students according to the rapid progress in medical genetics and profound impact of molecular technologies on health care services.

4th or 5th year of undergraduate education

Duration of course: ***2 weeks***

ESTC credits: ***3***

	LECTURES & SEMINARS	PRACTICAL SESSIONS
1	Human Genome and genetic variations. *Architecture of human genome:* *Structure of nucleic acids and genes* *Types of sequences, major types of interspersed repetitive DNA, segmental duplications* *Transcription, splicing of primary transcript. Translation, genetic code* *Dynamic genome and polymorphisms* *Mitochondrial genome*	*Laboratory of molecular genetics:* *Principles of DNA extraction* *PCR* *RT-PCR* *Electrophoresis* *Interpretation of results*
2	Basic laws of genetics. *Mendel laws of inheritance:* *Dominant/recessive segregation and deviations* Introduction to clinical genetics *What can we learn from a family history* *Pedigree interpretation* *Penetrance and expressivity, pleiotropic effect. Rare modes of inheritance* *Genotype-phenotype correlations* *Epigenetics:* *X-inactivation, DNA methilation* *Imprinting: example of disorders*	*Laboratory of molecular genetics:* *Principles of DNA sequencing* *Principles of next generation sequencing*
3	Molecular genetics *Gene mutations* DNA analysis of selected disorders *Nucleic acids hybridization for DNA testing* *Amplifying the sequence of interest* *DNA sequencing* *Genome analysis and DNA microarray*	*Laboratory of cytogenetics:* *Cultivation of lymphocytes* *Karyotyping* *FISH* *Interpretation of results*
4	Clinical cytogenetics *Chromosomes, nomenclature* *Numerical and structural chromosome abnormalities* *Effects of non-disjunction in meiosis* *Copy number variants* *Balanced and unbalanced abnormalities* *Constitutional and mosaic abnormalities* *Chromosomal syndromes* *Recurrent microdeletion and microduplication syndromes*	
5	Congenital malformations or birth defects *Genetic causes* *Clinical features* *Common types* *Clinical care* *Possibilities of treatment* *Prenatal diagnostics*	

6	Hereditary autoinflammatory syndromes *Autoinflammatory and autoimmune disorders* *Clinical and genetic features and diagnostic value of MEFV gene mutations* *FMF: genetic testing and counselling, genotype-phenotype correlations, treatment and colchicine resistance cases* *Management, treatment and surveillance of patients with autoinflammatory syndromes*	*Genetic counselling* *Simulation of genetic counselling cases*
7	Cancer genetics *Features of hereditary cancer syndromes* *Genetic mechanisms of cancer* *Examples of genetic cancer syndromes and mutations in oncogenes and tumor supression genes* *Syndromes of DNA repair defects and cancer* *Genetic and environmental factors that affect cancer risk* *Counselling of families with higher cancer risk* *Genetic basis of hematological cancer (leukaemia): diagnostics and treatment*	
8	Neurogenetics *Premutation, anticipation, hypermethilation and loss of protein function in neurologic disorders* *Neurologic diseases caused by single genes with wide-ranging systemic phenotypes* *Clinical and genetic correlations of congenital myopathies: Duchenne& Becker muscular dystrophy* *Trinucleotide repeat expansion: Fra-X syndrome, Huntington disease and myotonic dystrophy* *Genetics of movement disorders: Parkinson disease, dystonia, etc*	*Practical in genetic counselling*
9	Inborn errors of metabolism *Inborn errors of metabolic pathways* *Types of inheritance and signs and symptoms of IEMs.* *Classification of IEM:* *Metabolism of phenylalanine and tyrosine (metabolic block: albinism, alkaptonuria, phenylketonuria)* *Congenital adrenal dysplasia:* *metabolic block for production of steroid hormones* *Lysosomal storage disorders* *Disorders caused by defects in the metabolism of simple amino acids and monosaccharides.* *Neonatal screening strategy for IEMs*	
10	Genetic screening and prenatal diagnostics *Genetic characteristics in local populations, mutation frequencies and disease prevalence* *Contributing factors for population screening programs for genetic diseases* *Prenatal, neonatal, and postnatal screening* *Principles of prevention and screening* *Ethical, legal, social, and cultural issues associated with genetic screening programs* Basics of Pharmacogenetics and Pharmacogenomics	*Laboratory of biochemistry, immunohistochemistry & clinical laboratory* *Interpretation of results*