

<b>Higher education institution:</b> <i>Slovak Medical University in Bratislava</i>					
<b>Faculty:</b> <i>Faculty of Medicine</i>					
<b>Course code:</b> <i>GM 046</i>			<b>Course title:</b> <i>Clinical genetics</i>		
<b>Type, extent and method of educational activity:</b> <i>Number of hours per semester:</i> <i>Lectures: 14/1 hours per week</i> <i>Practices: 0 hours per week</i>					
<b>Number of credits:</b> <i>1 credits</i>					
<b>Recommended semester/trimester study:</b> <i>9. th semester</i>					
<b>Level of higher education study:</b> <i>1. + 2. level</i>					
<b>Prerequisite courses:</b>					
<b>Requirements for completion of the course:</b> <i>1 Final test: minimum threshold of success: 60%. Evaluation: Evaluation: A: 95% -100%, B: 88% -94% C: 77% -87%, D: 66% -76%, E: 60% -65%. Active participation in the course – continuous assessment- (credit)</i> <i>Student workload is 11 hours.</i>					
<b>Learning outcomes:</b> <i>Acquisition of theoretical knowledge and practical skills from the field..</i>					
<b>Brief content of the course (syllabus):</b> <i>Genetic diseases, classification, distribution, occurrence.</i> <i>Developmental disorders and Birth defects,</i> <i>Clinical cytogenetics – Disorders of the Autosome and Gonosome</i> <i>Clinical genetic approach to dysmorphic patient</i> <i>Basic of dysmorphology and syndromologic diagnostics,</i> <i>Types of genetic tests and their laboratory diagnostics.</i> <i>Basic and advanced methods of DNA diagnostics</i> <i>Selected frequent monogenic diseases and their molecular basis,</i> <i>Hereditary metabolic diseases</i> <i>Rare genetic diseases and syndromes</i> <i>Cancer Genetics- acquired chromosomal abnormalitie, hemoblastosis and tumor diseases</i> <i>Neurogenetic and behavioral diseases</i> <i>Reproductive genetics, infertility and sterility disorders,</i> <i>Prenatal genetic diagnosis procedures and methods.</i> <i>Genetic Counseling and Risk Assesment in selected cases</i> <i>Genomic and personalised medicine, ethical and social issues in the field.</i> <i>Treatment and prevention of genetic disorders, interdisciplinary cooperation</i>					
<b>Recommended literature:</b> <i>Thompson a Thompson's : Genetics in Medicine, Seventh Edition, Saunders Elsevier-Philadelphia, 2007, p.600, ISBN-10 1416030808</i> <i>Sršeň, Š. - Sršňová, K.: Základy klinickej genetiky a jej molekulárna podstata, 4. prepracované vydanie, Osveta, Martin, 2005, 446 s. ISBN 80-8063-185-9.</i>					
<b>Language requirements:-</b>					
<b>Notes:</b> <i>The course runs in Slovak and English language.</i>					
<b>Course assessment</b> Assessed students in total: 0					
A	B	C	D	E	FX
0%	0%	0%	0%	0%	0%
<b>Lecturers:</b> <i>doc. MUDr. Darina Ďurovčíková, CSc.,</i> <i>RNDr. Katarína Závodná, PhD.,</i> <i>RNDr. Regina Behulová, PhD.,</i>					
<b>Date of last modification:</b> <i>01.09.2014</i>					

**Supervised by:** *prof. MUDr. Peter Šimko, CSc.*