Course Title	Medical Genetics				
Course Code	BMS224				
Course Type	Compulsory				
Level	Bachelor (1st Cycle)				
Year / Semester	2nd Year / 4th Semester				
Teacher's Name	TBA				
ECTS	8	Lectures / week	3 Hours	Laboratories / week	2 Hours
Course Purpose and Objectives	The objective of the course is to familiarize students with: • The fundamentals of human genetics and their manifestation at the cellular, organ, individual and population levels • Classical as well as modern Genetics and its applications in diagnosis and research • The expression of genetic diseases and their significance for clinical medicine, diagnosis and treatment				
Learning Outcomes	Upon successful completion of this course students should be able to: ▪ Discuss the basic concepts of human genetics. ▪ Identify the genetic components of polygenic and multifactorial diseases. ▪ Describe the relationship between gene structure and function and its implication in the development of genetically-based diseases. ▪ Identify the clinical presentation and etiology of genetic disorders including: single gene disorders, disorders of chromosome abnormalities, inborn errors of metabolism, multifactorial genetic disorders and cancer genetics. ▪ Account for the occurrence, causes, pathophysiology, diagnostic principles, and ethical considerations of the most common genetic disorders (i.e. muscular dystrophies, cystic fibrosis, thalassaemias, hemophillia, genetic deafness, Huntington's disease, color blindness, hereditary cancer). ▪ Assess and appraise the importance, usefulness and limitations of genetic tests including: cytogenetic testing, molecular testing, pre-natal testing, genome scanning, new born screen and biochemical genetics testing. Determine which test(s) are most appropriate for a given clinical scenario.				

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<tr><td></td><td colspan="3">Record family-history in relation to the inheritance of a certain genetic trait (pedigree analysis), and predict inheritance pattern.
Debate the contribution of current advances in molecular genetic research and its implementation in clinical practice.
Explain the importance of the study of population genetics and pharmacogenetics in the study of genetic diseases.</td></tr>
<tr><td>Prerequisites</td><td>None</td><td>Co-requisites</td><td>None</td></tr>
<tr><td>Course Content</td><td colspan="3"><u>Theory</u>:
Fundamentals of human genetics.
The chromosome and the molecular basis of monogenic, polygenic and multifactorial diseases.
Gene structure and function
Chromosomal theory of inheritance
Genetically-based diseases and Mendel’s laws
Mutations-Genotype and environment. DNA repair mechanisms
Cytogenetics.
Causes, pathophysiology, and diagnosis of the most common genetic disorders (i.e. muscular dystrophies, cystic fibrosis, thalassaemias, hemophillia, genetic deafness, Huntington’s disease, color blindness, hereditary cancer).
Molecular genetic testing, pre-natal testing and newborn testing.
Extranuclear inheritance and mitochondrial disorders
Gene therapy.
Population genetics.
Current advances in molecular genetic research and their implementation in clinical practice. Pharmacogenetics and pharmacogenomics.
<u>Laboratory exercises:</u>
Cytogenetics-Karyotype
Monohybrid and dihybrid crosses in corn (Corn Genetics)-Chi square test
DNA isolation from plants
Blood typing
Detection of genetically modified organisms using lateral flow strips
Pedigree analysis
Mutation analysis using ARMS (Amplification-Refractory Mutation System) method or method to assess whether sunscreen protects DNA from UV-induced mutations (using yeast as a model organism).</td></tr>
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- Study of monogenic diseases using RFLPs (Restriction Fragment Length Polymorphisms).

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<tr><td>Teaching Methodology</td><td>Face- to- face</td></tr>
<tr><td>Bibliography</td><td>Medical Genetics: An Integrated Approach
by G. Bradley Schaefer, James N. Thompson, Jr.; McGraw Hill Medical; ISBN 978-0-07-166438-7

Lewin's Genes X; Jocelyn E. Krebs; 10th; 978-0763779924; Jones and Bartlett Publishers, Inc; 2009

Genetics: From Genes to Genomes, by Hartwell LH., Hood L., Goldberg ML., Reynolds AE., and Silver LM., 2014

ADDITIONAL RECOMMENDED TEXTBOOKS:

Essential Medical Genetics:; Connor, M. / Ferguson, M.; 978-1405169745; Wiley-Blackwell; 2011

Thompson and Thompson Genetics in Medicine; Nussbaum, R.; 7th ; 978-1416030805; Saunders; 2007</td></tr>
<tr><td>Assessment</td><td>

Mid - Term Examination	30%
Final Examination	40%
Assignments/Lab	20%
Class Participation	10%
	100%

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<tr><td>Language</td><td>English</td></tr>
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