Institute of Microbiology and Molecular Genetics
Faculty of Life Sciences
University of the Punjab, Lahore
Course Outline

Programme	BS	Course Code	MMG303	Credit Hours	3(2+1)
Course Title	**HUMAN GENETICS**				

COURSE INTRODUCTION

This course explores the principles of human genetics with a focus on molecular mechanisms underlying genetic variation, inheritance patterns, and the implications for human health and disease. Topics will include genetic mapping, gene expression, genomic technologies, and the ethical, legal, and social implications of genetic research.

LEARNING OUTCOMES

On the completion of the course, students will be able to

1: Understand the basics of human genetics

2: Recognize inheritance patterns and influence/ linkage of gender on various human traits

3: Evaluate the effect of the environment on genes

4: Understand certain in-born errors in metabolism

5: Understand the importance of euploidy and diseases caused by abnormal numbers or structures of chromosomes

COURSE CONTENT

Introduction to Human Genetics: Overview of human genetics and its relevance. Genetic terminology and concepts; Chromosomal Basis of Inheritance: Structure and function of chromosomes, Chromosomal numerical and structural defects and abnormalities, Techniques for chromosomal analysis (karyotyping, FISH); Molecular Genetics of Human Disease: Single-gene disorders, Inheritance patterns (autosomal dominant, autosomal recessive, X-linked), Mutation types and their effects on gene function; Human Genetic Variation: Types of genetic variation (SNPs, CNVs, in-dels), Population genetics and genetic diversity; Gene Mapping and Linkage Analysis: Methods for mapping genes (linkage analysis, association studies), Gene mapping in complex traits; Gene Expression and Regulation: Mechanisms of gene regulation (transcriptional, post-transcriptional), epigenetics and its role in gene expression; Genetic Basis of Complex Diseases: Multifactorial and polygenic inheritance, examples of complex diseases (e.g., cancer, cardiovascular diseases), Genetic and environmental interactions; Ethical, Legal, and Social Issues: Genetic privacy and discrimination, Informed consent and genetic testing, Ethical issues in genetic research and therapy, genetic counseling; Current Topics and Advances in Human Genetics: Recent advancements in human genetics research, Emerging technologies and their impact.

PRACTICALS

Problems relating to pedigree analysis of various human genetic diseases. Problems related to sex linkage, inherited variations, deletion and duplication mapping, study of chromosomal analysis techniques and problems related to chromosomal numerical/ structural defects

TEXTBOOKS AND READING MATERIAL

1: Lweis, R. (2018). *Human Genetics, Concepts, and Applications*. 12th Edition. McGraw Hill, U.S.A.

2: Dennis, C., Gallagher, R. and Watson, J. (2000). *The Human Genome*, 1st edition, Palgrave, U.K.

3: Lewein, B. (2004). *Gene VII*. Oxford University Press, U.K.

4: Strachan, T. and Read, A. P.(1999). *Human Molecular Genetics*, 2nd Edition, Wiley-Liss, N.Y.

5. Gardner, R. M., Sutherland, G. R., & Shaffer, L. G. (2012). *Chromosome Abnormalities and Genetic Counseling* . Oxford University Press, USA.

ASSESSMENT

Sr. No.	Elements	Weightage	Details
1.	Midterm Assessment	35%	Written Assessment at the mid-point of the semester.
2.	Formative Assessment	25%	Continuous assessment includes Classroom participation, assignments, presentations, viva voce, attitude and behavior, hands-on activities, short tests, projects, practicals, reflections, readings, quizzes etc.
3.	Final Assessment	40%	Written Examination at the end of the semester. It is mostly in the form of a test, but owing to the nature of the course the teacher may assess their students based on term paper, research proposal development, fieldwork , report writing etc.