

CENTRAL UNIVERSITY OF PUNJAB



Ph.D in Human Genetics

Batch 2026-27

**Department of Human Genetics and
Molecular Medicine**

Graduate Attributes

The students graduating in PhD (Human Genetics) will contribute to teaching and research at local, regional, national and international levels. This programme will enrich students with fundamental knowledge of research ethics, research methodology in the field of human genetics. On successful completion of this Programme, the students will be able to:

- Integrate multidisciplinary approaches to analyze the role of genetic and molecular factors in health and disease
- Design and perform molecular genetics and epidemiological studies for health research
- Compete at national and global level to pursue research and teaching in any field of life sciences
- Perform best practices in research in the field
- Contribute to the scientific workforce that will transform molecular diagnostics and healthcare sectors employing higher order thinking skills and capabilities.

Course Structure of the Programme

Ph.D in Human Genetics

Course Code	Course Title	Course Type	Hours			Credit
			L	T	P	
MHGE.701	Research Methodology and Biostatistics	Core course	4	0	0	4
MHGE.702 [#]	Advanced Human Genomics and Epigenetics	Core course	4	0	0	4
MHGE.703 [#]	Essential of Research in Human Genetics	Core course	4	0	0	4
MHGE.751	Research and Publication Ethics	Core course	4	0	0	2
MHGE.752	Teaching Assistantship	Skill Based	0	0	2	1
UNIV.753	Curriculum, Pedagogy and Evaluation	Core course	1	0	0	1
TOTAL						12

[#]Any one course need to be opted between MHGE.702 and MHGE.703.

Details of syllabus

Course Code: HGE.701
Course Title: Research Methodology and Biostatistics
Total Hours: 60

L	T	P	C
4	0	0	4

Learning Outcomes:

On successful completion of the course the student will be able to:

1. Analyze and evaluate wide variety of statistical data and research writing
2. Perform research and develop knowledge on Intellectual Property Rights
3. Perform statistical data analysis
4. Gather basic knowledge on computer systems and utilize them for research

Unit 1 General principles of research: Meaning and importance of research, Critical thinking, Formulating hypothesis and development of research plan, Review of literature, Interpretation of results and discussion. Technical writing: Technical and scientific writing: thesis, technical papers, reviews, electronic communication, research papers, etc. Poster preparation and Presentations and Dissertation. Reference management using various softwares: Endnote, reference manager, refwork, etc. Communication skills – defining communication, types of communication, techniques of communication, etc.	15 Hours	CLO1
Unit 2 Intellectual Property Rights: Intellectual Property, intellectual property protection (IPP) and intellectual property rights (IPR), WTO (World Trade Organization), WIPO (World Intellectual Property Organization), GATT (General Agreement on Tariff and Trade), TRIPs (Trade Related Intellectual Property Rights), TRIMS (Trade Related Investment Measures) and GATS (General Agreement on Trades in Services), Nuts and Bolts of Patenting, Technology Development/Transfer Commercialization Related Aspects, Ethics and Values in IP.	15 Hours	
Unit 3 Biostatistics: Difference between parametric and non-parametric statistics, Univariant and multivariant analysis, Confidence interval, Errors, Levels of significance, Hypothesis testing. Measures of central tendency and dispersal, Histograms, Probability distributions (Binomial, Poisson and Normal), Sampling distribution, Kurtosis and skewness Comparative Statistics: Comparing means of two or more groups: Student’s t-test, Paired t-test, Mann-Whitney U-test, Wilcoxon signed-rank, One-way and two-way analysis of variance (ANOVA), Critical difference (CD), Fisher’s LSD (Least significant difference), Kruskal–Wallis one-way ANOVA by ranks, Friedman two-way ANOVA by ranks, Chi-square test Regression and correlation: Standard errors of regression coefficients, Comparing two regression lines, Pearson Product - Moment Correlation Coefficient, Spearman Rank correlation coefficient, Power and sampling size in correlation and regression.	15 Hours	CLO3
Unit 4 Fundamentals of computer: Parts of computer, Hardware, BIOS, Operating systems, Binary system, Logic gates and Boolean algebra. Application software: Spreadsheet applications, Word-processing applications, Presentation applications, Internet browsers, Reference Management, and Image processing applications. Computer language: Basic DOS commands, AutoHotKey scripting language, HTML and basic structure of a webpage, Designing websites.	15 Hours	

World wide web: Origin and concepts, Latency and bandwidth, Searching the internet, Advanced web-search using Boolean logic, Cloud computing.	
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Transactional Modes: Lecture; Tutorial; Problem solving; Self-learning.

Suggested Readings:

1. Norman, G. and Streiner, D. (2008). Biostatistics: The Bare Essentials. (with SPSS), 4th Edition, People's Medical Publishing House, USA.
2. Sokal, R.R. and Rohlf, F.J. (1994). Biometry: The Principles and Practices of Statistics in Biological Research. 4th Edition, W.H. Freeman publishers, USA.
3. Banerjee P.K (2014). Introduction to Biostatistics. S.Chand, India
4. Daniel WW (2010). Biostatistics: A Foundation for Analysis in the Health Sciences. John Wiley and Sons Inc.
5. Bailet NTJ. Statistical Methods in Biology. Cambridge Univ. Press.
6. Glaser AN. High-Yield Biostatistics. Lippincott Williams & Wilkins.
7. Gupta, S. (2008). Research Methodology and Statistical Techniques. Deep and Deep Publications (P) Limited, New Delhi.
8. Kothari, C. R. (2014). Research Methodology (s). New Age International (p) Limited. NewDelhi.
9. Sahay, Vinaya and Pradumna Singh (2009). Encyclopedia of Research Methodology in life Sciences. Anmol Publications. New Delhi

Course Code: HGE.702
Course Title: Advanced Human Genomics and Epigenetics
Total Hours: 60

L	T	P	C
4	0	0	4

Learning Outcomes:

On successful completion of the course the student will be able to:

1. Know the basic and advanced concepts in human cytogenetics
2. Practice cell culture techniques to perform experiments
3. Conceptualize and gather knowledge on human genome sequencing
4. Know in depth concepts of epigenomics

Unit 1 Advanced Human Cytogenetics: GTG banding and nomenclature of human chromosomes; structure of X and Y chromosomes; X and Y pairing and pseudo autosomal region; Molecular mechanism of X inactivation; Molecular cytogenetics methods: FISH, CGH, SKY; Cytogenetics of cancer.	15 Hours	CLO1
Unit 2 Tissue culture techniques: Whole blood culture; bone marrow culture; amniocyte culture; chorionic villi culture; skin fibroblast culture.	15 Hours	CLO2
Unit 3 Pharmacogenomics and Overview of Human Genome Project: Concept of individual based treatment, Drug Metabolism; Genetic makeup and Drug Response; High throughput screening for drug discovery; Identification of drug targets; Pharmacogenetics and drug development, Personalized Medicine; goals of Human Genome Project, its implications on research and human society; Strategies for genome sequencing; Early, next and third generation DNA sequencing methods; Personalized medicine.	15 Hours	CLO3

Unit 4 Introduction and molecular mechanisms of Epigenetics: Mechanisms of DNA methylation; Histone modifications; Chromosomal position effect and gene variegation; Epigenetic control of gene activity; Analysis of gene-specific DNA methylation; Methods of assessing genome-wide DNA methylation; Model organism of epigenetic: Drosophila	15 Hours	CLO4
Transactional Modes: Lecture; Demonstration; Tutorial; Lecture cum demonstration; Problem solving; Self-learning.		

Suggested Readings:

1. Tollefsbol T.(2011). Handbook of Epigenetics, Elsevier Publications
2. Carey N. (2013). The Epigenetics Revolution: How Modern Biology Is Rewriting Our Understanding of Genetics, Disease, and Inheritance. Columbia Univ Pr.
3. Wallach J (2014). Epigenetics: The Death of the Genetic Theory of Disease Transmission. Kindle Publications, Columbia University Press.
4. Francis R.C. (2012). Epigenetics: How Environment Shapes Our Genes. W.W. Norton and Company, New York.
5. Jocelyn, E. K., Elliot, S. G. and Stephen, T. K. (2009), Lewin's Gene X. Jones & Barlett.
6. Korf, B.R. (2006). Human Genetics and Genomics. Wiley Blackwell.
7. Lodish, H., Berk, A., Chris, A. K., Krieger, M. (2008), Molecular Cell Biology. W.H.Freeman, USA.

Course Code: HGE.703
Course Title: Essential of Research in Human Genetics
Total Hours: 60

L	T	P	C
4	0	0	4

Learning Outcomes:

On successful completion of the course the student will be able to:

CLO1: To equip students with bioethical issues and biological safety protocols while carrying out research

CLO2: to equip students with concepts of statistical models, study design and data analysis

CLO3: To understand the theoretical principal of microscopy and cell culture techniques

CLO4: To enable students to understand the role of genetic sequence and epigenetic mechanism in regulating gene expression

Unit I: Biosafety and Bioethics: Good laboratory practices; Biological containment (BC) and physical containment (PC); biohazard management. Ethical in human and animal research; Regulatory compliance – Institutional Ethical committee, Institutional Animal Ethics committee and Institutional Biosafety committee; the element of informed consent- confidentiality, rights of subjects	CLO1
Unit II Statistical Genetics and Epidemiology: Methods of analyzing genetic data for disease risk prediction and drug discovery; Study Design; Cohort study; Cross-sectional studies; Methods of sampling and sample size calculation; Systematic review, meta-analysis and evidence synthesis.	CLO2

Unit III: Microscopy and Cell Culture: Requirements of a Cell culture laboratory; Types of cell culture- Primary cell culture and cell line culture, Peripheral and adherent culture; Parts of microscope, magnification and resolution	CLO3
Unit IV: Genetics and Epigenetics: Nucleic acid isolation, quality assessment and quantification; Gel Electrophoresis- Agarose and PAGE; mechanism of epigenetic and gene expression regulation; Variation in Genetic sequence and methylation analysis.	CLO4

Transactional Modes: Lecture; Demonstration; Tutorial; Lecture cum demonstration; Problem solving; Self-learning.

Suggested Readings:

1. Beauchamp, T.L. and Childress, J.F. (2019). Principles of Biomedical University Press
2. Dawn, P.W. and Byers, K.B. (2017). Biological Safety: Principles and Practices (Fifth Ed.). ASM Press
3. Lyle, Armstrong. (2013). Epigenetics Garland Science
4. Shannon, T.A. (2023). An Introduction to Bioethics (4th ed). Paulist Press, USA
5. Thomas, D. C. (2004). Statistical methods in genetic epidemiology. Oxford University Press.
6. Woodward, M. (2013). Epidemiology: study design and data analysis (3rd ed.). CRC Press.

Course Code: HGE.751
Course Title: Research and Publication Ethics
Total Hours: 30

L	T	P	C
2	0	0	2

Learning Outcomes:

On successful completion of the course the student will be able to:

1. Understand the philosophy of research
2. Perform good research practices
3. Write scientific literature for peer reviewed publication
4. Perform scientific literature writing in an efficient manner

Unit 1 5 Hours Philosophy and Ethics • Introduction to philosophy: Detention, nature and scope, concept, branches • Ethics: definition, moral philosophy, nature of moral judgments and reactions	CLO1
Unit 2 5 Hours Scientific conduct • Ethics with respect to science and research • Intellectual honesty and research integrity. • Scientific misconduct: Falsification, Fabrication and Plagiarism (FFP) • Redundant publications: duplicate and overlapping publication, salami slicing • Selective reporting and misrepresentation of data.	CLO2

Unit 3 5 Hours Publication ethics	CLO3
• Publication ethics: definition, introduction and importance • Best practice/ standards setting initiatives and guidelines: COPE, WAME, etc. • Conflicts of interest • Publication misconduct: definition, concept, problems that leads to unethical behavior and vice versa, types • Violation of publication ethics, authorship and contribution ship • Identification of publication misconduct, complaints and appeals • Predatory publishers and journals	
Unit 4 15 Hours Open access publishing • Open access publications and initiatives • SHERPA/RoMEO online resource to check publisher copyright & self-archiving policies • Software tool to identify predatory publications developed by SPPU • Journal finder / journal suggestion tools viz., JANE, Elsevier Journal Finder, Springer Journal Suggester, etc. Publication misconduct A. Group Discussion • Subject specific ethical issues, FFP, authorship, Conflicts of interest • Complaints and appeals: examples and fraud from India and abroad B. Software tools • Use of plagiarism software like Turnitin, Urkund, and other open source software tools Databases and Research metrics A. Databases • Indexing databases • Citation databases: Web of Science, Scopus, etc. B. Research Metrics • Impact Factor of journal as per Journal Citation Report, SNP, SJR, IPP, Cite Score • Metrics: h-index, g index, i10 index, altmetrics	CLO4

Transactional Modes: Lecture; Demonstration; Tutorial; Quiz; Lecture cum demonstration; Problem solving; Self-learning.

Course Code: HGE.752
Course Title: TEACHING ASSISTANTSHIP
Total Hours: 30

L	T	P	C
0	0	2	1

Learning Outcomes:

At the end of this skill development course, the scholars shall be able to

1. Familiarize themselves with the pedagogical practices of effective classroom delivery and knowledge evaluation system
2. Manage large and small classes using appropriate pedagogical techniques for different types of content

Activities and Evaluation:

- The scholars shall attend Master degree classes of his/her supervisor to observe the various transaction modes that the supervisor follows in the class room delivery or transaction process

one period per week.

- The scholars shall be assigned one period per week under the direct supervision of his/her supervisor to teach the master's degree students adopting appropriate teaching strategy(s).
- The scholars shall be involved in examination and evaluation system of the master's degree students such as preparation of questions, conduct of examination and preparation of results under the direction of the supervisor.
- At the end of the semester, the supervisor shall conduct an examination of teaching skills learned by the scholar as per the following evaluation criteria:
- The scholars shall be given a topic relevant to the master's degree course of the current semester as his/her specialization to prepare lessons and deliver in the classroom before the master degree students for one hour (45 minutes teaching + 15 minutes interaction).
- The scholars shall be evaluated for a total of 50 marks comprising content knowledge (10 marks), explanation and demonstration skills (10 marks), communication skills (10 marks), teaching techniques employed (10 marks), and classroom interactions (10).

Course Code: UNI.753
Course Title: Curriculum, Pedagogy and Evaluation
Total Hours: 30

L	T	P	C
2	0	0	2

Learning Outcomes:

After completion of the course, scholars shall be able to:

CLO1 Analyze the principles and bases of curriculum design and development

CLO2 Examine the processes involved in curriculum development

CLO3 Develop the skills of adopting innovative pedagogies and conducting students' assessment

CLO4 Develop curriculum of a specific course/programme

Course Content

Unit I Bases and Principles of Curriculum 4 hours 1. Curriculum: Concept and Principles of curriculum development, Foundations of Curriculum Development. 2. Types of Curriculum Designs- Subject centered, learner centered, experience centered and core curriculum. Designing local, national, regional and global specific curriculum. Choice Based Credit System and its implementation.	CLO1
Unit II Curriculum Development 4 hours 1. Process of Curriculum Development: Formulation of graduate attributes, course/learning outcomes, content selection, organization of content and learning experiences, transaction process. 2. Comparison among Interdisciplinary, multidisciplinary and trans-disciplinary approaches to curriculum.	CLO2
Unit III Curriculum and Pedagogy 3 hours 1. Conceptual understanding of Pedagogy. 2. Pedagogies: Peeragogy, Cybergogy and Heutagogy with special emphasis on Blended learning, Flipped learning, Dialogue, cooperative and collaborative learning 3. Three e- techniques: Moodle, Edmodo, Google classroom	CLO3

Unit IV Learners' Assessment	4 hours	CLO4
1. Assessment Preparation: Concept, purpose, and principles of preparing objective and subjective questions. 2. Conducting Assessment: Modes of conducting assessment – offline and online; use of ICT in conducting assessments. 3. Evaluation: Formative and Summative assessments, Outcome based assessment, and scoring criteria.		

Transaction Mode

Lecture, dialogue, peer group discussion, workshop

Evaluation criteria

There shall be an end term evaluation of the course for 50 marks for duration of 2 hours. The course coordinator shall conduct the evaluation.

Suggested Readings

- Allyn, B., Beane, J. A., Conrad, E. P., & Samuel J. A., (1986). Curriculum Planning and Development. Boston: Allyn & Bacon.
- Brady, L. (1995). Curriculum Development. Prentice Hall: Delhi. National Council of Educational Research and Training.
- Deng, Z. (2007). Knowing the subject matter of science curriculum, Journal of Curriculum Studies, 39(5), 503-535. <https://doi.org/10.1080/00220270701305362>
- Gronlund, N. E. & Linn, R. L. (2003). Measurement and Assessment in teaching.
- Singapore: Pearson Education
- McNeil, J. D. (1990). Curriculum: A Comprehensive Introduction, London: Scott,
- Foreman/Little
- Nehru, R. S. S. (2015). Principles of Curriculum. New Delhi: APH Publishing Corporation.
- Oliva, P. F. (2001). Developing the curriculum (Fifth Ed.). New York, NY: Longman
- Stein, J. and Graham, C. (2014). Essentials for Blended Learning: A Standards-Based Guide. New York, NY: Routledge.

Web Resources

- https://www.westernsydney.edu.au/__data/assets/pdf_file/0004/467095/Fundamentals_of_Blended_Learning.pdf
- <https://www.uhd.edu/academics/university-college/centers-offices/teaching-learningexcellence/Pages/Principles-of-a-Flipped-Classroom.aspx>
- <http://leerwegdialoog.nl/wp-content/uploads/2018/06/180621-Article-The-BasicPrinciples-of-Dialogue-by-Renate-van-der-Veen-and-Olga-Plokhooij.pdf>