

CENTRAL UNIVERSITY OF PUNJAB, BATHINDA



Ph.D in Human Genetics

Session - 2020

**Department of Human Genetics and
Molecular Medicine**

Programme Outcome

This programme will enrich students with basic 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

IQAC

Course Structure of the Programme

Course Code	Course Title	Course Type	Hours			Credit
			L	T	P	
LHG.701	Research Methodology and Biostatistics	Core course	4	0	0	4
LHG.702	Advanced Human Genomics and Epigenomics	Core course	4	0	0	4
LHG.797	Credit Seminar	Skill Based course	0	0	0	2
LHG.751	Research and Publication Ethics	Core course	2	0	0	2
Total			12	0	0	12

IQAC

Course Code: LHG.701

Course Title: Research Methodology and Biostatistics

Total Hours: 60

L	T	P	Cr
4	0	0	4

Learning Outcomes:

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

- Analyze and evaluate wide variety of statistical data
- Represent statistical data and summary statistics in graphical and tabular forms
- Apply suitable statistical tools to analyze data
- Write and communicate scientific reports, projects and publications

UNIT I

Hours: 15

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, rework, etc. Communication skills – defining communication, types of communication, techniques of communication, etc.

UNIT II

Hours: 15

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.

UNIT III

Hours: 15

Biostatistics: Difference between parametric and non-parametric statistics, Univariate and multivariate 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.

UNIT IV

Hours: 15

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. World wide web: Origin and concepts, Latency and bandwidth, Searching the internet, Advanced web-search using Boolean logic, Cloud computing.

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

L	T	P	Cr
4	0	0	4

Course Code: LHG.702

Course Title: Advanced Human Genomics and Epigenetics

Total Hours: 60

Learning Outcomes:

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

- Know the basic and advanced concepts in human genomics and epigenomics
- Practice cell culture techniques to perform experiments
- Synthesise idea about the modern aspects of epigenomics
- Know in depth concepts of pharmacogenomics

UNIT I

Hours: 15

Advanced Human Cytogenetics: GTG banding and nomenclature of human chromosomes; structure of X and Y chromosomes; X and Y pairing and pseudoautosomal region; Molecular mechanism of X inactivation; Molecular cytogenetics methods: FISH, CGH, SKY; Cytogenetics of cancer.

UNIT II

Hours: 15

Tissue culture techniques: Whole blood culture; bone marrow culture; amniocyte culture; chorionic villi culture; skin fibroblast culture.

UNIT III

Hours: 15

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.

UNIT IV

Hours: 15

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

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: CPE-RPF
Course Title: Credit Seminar

L	T	P	Cr
0	0	0	2

Total Hours: 30

Learning Outcomes:

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

- Improve communication aptitude
- Learn presenting paper or data in scientific forum

Credit Seminar topics will be decided jointly by PhD supervisor and the student and will be presented in open house. Seminar presentation will be followed by open discussion.

Evaluation criteria: The detailed assessment criteria are as per University policy. The students will be assessed based on presentation and report submitted on the topics assigned by seminar coordinator.

IQAC

Course Code: LHG.751

Course Title: Research and Publication Ethics

L	T	P	Cr
2	0	0	2

Total Hours: 30

Learning Outcomes:

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

- Improve communication aptitude
- Learn presenting paper or data in scientific forum

THEORY

UNIT I: Philosophy and Ethics

Hours: 3

- Introduction to philosophy: Detention, nature and scope, concept, branches
- Ethics: definition, moral philosophy, nature of moral judgments and reactions

UNIT II: Scientific conduct

Hours: 5

- 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.

UNIT III: Publication ethics

Hours: 7

- 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

PRACTICE

UNIT IV: Open access publishing

Hours: 4

- 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.

UNIT V: Publication misconduct

Hours: 4

A. Group Discussion

Hours: 2

- Subject specific ethical issues, FFP, authorship
- Conflicts of interest
- Complaints and appeals: examples and fraud from India and abroad

B. Software tools

Hours: 2

- Use of plagiarism software like Turnitin, Urkund, and other open source software tools

UNIT VI: Databases and Research metrics

Hours: 7

A. Databases

Hours: 4

- Indexing databases
- Citation databases: Web of Science, Scopus, etc.

B. Research Metrics

Hours: 3

- Impact Factor of journal as per Journal Citation Report, SNP, SJR, IPP, Cite Score
- Metrics: h-index, g index, i10 index, altmetrics

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