## **CENTRAL UNIVERSITY OF PUNJAB**



Ph.D in Human Genetics

Batch 2023-24

# 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				Credit		
		Туре	L	Т	P	
HGE.701	Research Methodology and Biostatistics	Core course	4	0	0	4
HGE.702	Advanced Human Genomics and Epigenetics	Core course	4	0	0	4
HGE.751	Research and Publication Ethics	Core course	4	0	0	2
HGE.752	GE.752 Teaching Assistantship		0	0	2	1
UNIV.753	UNIV.753 Curriculum, Pedagogy and Evaluation		1	0	0	1
HGE.797	Seminar/Journal Club	Skill Based	0	0	4	2
TOTAL				14		

## **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.	CLO1
Reference management using various softwares: Endnote, reference manager, refwork, etc.	
Communication skills – defining communication, types of communication, techniques of communication, etc.	
Unit 2 15 Hours	
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	CLO2
Intellectual Property Rights), TRIMS (Trade Related Investment Measures) and GATS (General	CLO2
Agreement on Trades in Services), Nuts and Bolts of Patenting, Technology	
Development/Transfer Commercialization Related Aspects, Ethics and Values in IP.	
Unit 3 15 Hours	
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	CLO3
(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 4 15 Hours	
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,	CLO4
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

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.	CLO1
Unit 2 Tissue culture techniques: Whole blood culture; bone marrow culture; amniocyte culture; chorionic villi culture; skin fibroblast culture.	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 andhuman society; Strategies for genome sequencing; Early, next and third generation DNA sequencing methods; Personalized medicine.	CLO3

Unit 4 15 Hours	
Introduction and molecular mechanisms of Epigenetics: Mechanisms of DNA methylation;	
Histone modifications; Chromosomal position effect and gene variegation; Epigenetic control of	CI O4
gene activity;	CLO4
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: HGE.751

Course Title: Research and Publication Ethics

Total Hours: 30

		1	
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 Philosphy and Ethics Introduction to philosophy: Detention, nature and scope, concept, branches Ethics: definition, moral philosophy, nature of moral judgments and reactions	5 Hours	CLO1
<ul> <li>Unit 2</li> <li>Scientific conduct</li> <li>Ethics with respect to science and research</li> <li>Intellectual honesty and research integrity.</li> <li>Scientific misconduct: Falsification, Fabrication and Plagiarism (FFP)</li> <li>Redundant publications: duplicate and overlapping publication, salami slicing</li> <li>Selective reporting and misrepresentation of data.</li> </ul>	5 Hours	CLO2
Unit 3 Publication ethics	5 Hours	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

CLO4

#### **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

**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 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 degree course of the current semester as his/her specialization to prepare lessons and deliver in the class room 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**

CLO1
CLO2
CLO3
CLO4

#### **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**

- $\bullet \ https://www.uhd.edu/academics/university-college/centers-offices/teaching-learningexcellence/Pages/Principles-of-a-Flipped-Classroom.aspx$
- $\bullet \ http://leerweg dialoog.nl/wp-content/uploads/2018/06/180621-Article-The-Basic Principles-of-Dialogue-by-Renate-van-der-Veen-and-Olga-Plokhooij.pdf \\$

Course Code: HGE.797

Course Title: Seminar/Journal Club

**Total Hours: 30** 

L	T	P	C
0	0	4	2

#### **Learning Outcomes:**

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

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

Seminar/Journal Club topics will be decided jointly by PhD supervisor and the student and will be presented in open house, followed by open discussion.

#### **Evaluation criteria:**

- A. The performance of the students will be continuously evaluated based on the choice of the topic, preparation of the topic, referring new research in the area and also discussing the future perspective = 50 marks
- **B.** Final presentation and report writing = 50 marks