Centre for Human Genetics

Scheme of Programme Ph.D. Life Sciences with specialization Human Genetics

SEMESTER I

S.No	Paper Code	Course Title	L	T	P	Cr	% Weightage				E
							A	B	C	D	
1	HGS.601	Research Methodology	5	-	-	5	25	25	25	25	125
2	HGS.602	Computer Applications and Biostatistics	5	-	-	5	25	25	25	25	125
3	HGS.603	Advanced Human Cytogenetics and Advanced Human Molecular Genetics	3	-	-	3	25	25	25	25	75
4	HGS.604	Advanced Human Genomics and Epigenetics	3	-	-	3	25	25	25	25	75
5	HGS.698	Credit Seminar	-	-	16	8	-	-	-	-	200
			16	1	12	24					

A: <u>Continuous Assessment:</u> Based on Objective Type Tests
B: <u>Mid-Term Test-1:</u> Based on Objective Type & Subjective Type Test
C: <u>Mid-Term Test-2:</u> Based on Objective Type & Subjective Type Test
D: <u>End-Term Exam (Final):</u> Based on Objective Type Tests
E: Total Marks
L: Lectures   T: Tutorial   P: Practical   Cr: Credits

Semester I

	L	**T**	**P**	**Credits**	**Marks**

Course Title: Research Methodology and Bio-statistics

Course Code: HGS.601

L	**T**	**P**	**Credits**	**Marks**
4	0	0	4	100

Course Objectives: The objective of this subject is to ensure that a student learns basis of scientific research and statistical methods to arrive at and verify the conclusions drawn.

PART-A   **10 hours**

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: Scientific writing, Writing synopsis, Research paper, Poster preparation and Presentations and Dissertation.

PART-B   **15 hours**

General Statistics: 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, Kurtosisand skewness

PART-C   **16 hours**

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

PART-D   **15 hours**

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.

Reference books:

1. Gupta, S. (2008). Research methodology and statistical techniques. Deep & Deep Publications (P) Limited, New Delhi.
2. Kothari, C. R. (2014). Research methodology (s). New Age International (p) Limited. New
3. Delhi.
4. Sahay, Vinaya and Pradumna Singh (2009). Encyclopedia of Research Methodology in life
5. sciences. Anmol Publications. New delhi
6. Kauda J. (2012). Research Methodology: A Project Guide for University Students. Samfunds
litteratur Publications.

7. Dharmapalan B. (2012). Scientific Research Methodology. Narosa Publishing House
ISBN:
978-81-8487-180-7.
8. Norman, G. and Streiner, D. (2008). Biostatistics: The Bare Essentials.3/e (with SPSS).
Decker Inc. USA.
9. Rao, P. P., S. Sundar and Richard, J. (2009). Introduction to Biostatistics and Research
10. Methods. PHI learning.
11. Christensen, L. (2007). Experimental Methodology. Boston: Allyn & Bacon.

Course Title: Computer Applications and Biostatistics	**L**	**T**	**P**	**Credits**	**Marks**
Course Code: HGS.602	4	0	0	4	100

Course Objectives:

On the completion of this course, the students will be able to:
• Convey the essential characteristics of a set of data by representing in tabular and graphical forms and computing relevant measures of average and measures of variation
• Explain or predict values of a dependent variable based on the values of one or more independent variables
• Estimate the characteristics of populations based on their sample data
• Test specific hypotheses about populations based on their sample data
• Use appropriate procedures to analyse qualitative data
• Demonstrate competence in the use of statistical packages for analysis of data

PART-A   **14 hours**
Fundamentals of Computers: Block Diagram of Computer, Hardware Components, Introduction to computer network and World Wide Web, Sharing Data over Network, Internet Terminology, Searching over Internet, Google: advance Search Operations, Email, Checking Plagiarism using Internet

PART-B   **14 hours**
Introduction to Word Processing and Microsoft Office, Creating and Saving Documents, Text Formatting, Tables, Document Review Option, Mail Merge, Inserting Table of Contents, Reference Management. Introduction to Spreadsheet and Microsoft Excel, Text Formatting, Formulas, Charts, Table formatting, Sorting Records, Filtering the content.

PART-C   **14 hours**
Computer Configuration, Memory Hierarchy, Software Structure, Introduction to Operating System, Operating System types and functions. Introduction to Disk Operating System, DOS Internal and External Commands, Introduction to Windows operating System, Windows Task Manger.

PART-D   **14 hours**
Introduction to MS Paint, Figure Designing components in MS Paint
Introduction to Microsoft PowerPoint, Layout Selection, Designing and Formatting Slides, Slide Design and background formatting, Bullets and Numbering, Transition Style, Custom Animations, Hyperlink to Local files and Web Pages, Movies and Sound, Slide Timings.

Reference books:

1. Gookin, D. (2007). MS Word for Dummies. Wiley.

2. Harvey, G. (2007). MS Excel for Dummies. Wiley
3. Sinha, P.K., Computer Fundamentals, BPB Publications

Course Title: Advanced Human Cytogenetics and Advanced Human Molecular Genetics	L	T	P	Credits	Marks
Course Code: HGS.603	4	0	0	4	100

Course Objectives: Human cytogenetics was born in 1956 and since then, this field and our understanding of the link between chromosomal defects and disease have grown in spurts that have been fuelled by advances in cytogenetic technology. As a mature enterprise, cytogenetics now informs human genomics, disease and cancer genetics, chromosome evolution and the relationship of nuclear structure to function. Human Molecular Genetics is a vast field that provides understanding of the structure and function of the normal human genome. Human molecular genetics not only forms the cutting edge of biomedical research, but at the same time it has immediate application to the diagnosis of disease and has great potential for treating disease. Thus it is of major interest to all students of biological science and medicine, and to a wide range of biomedical researchers.

PART-A 14 hours

Historical aspects of Human Cytogenetics; Nomenclature of human chromosomes; Human Sex chromosomes; Structure of X and Y chromosome; X&Y Pairing and Pseudoautosomal region; Molecular mechanism of X inactivation; Tissue culture techniques: Whole Blood Culture, Bone Marrow Culture, Aminocyte Culture, Skin Fibroblast culture

PART-B 14 hours

High Resolutiom mapping; DNA fibre fish mapping; RFLPs and restriction mapping; Size fraction of large DNA molecule – Pulse field gel electrophoresis; Single nucleotide polymorphisms (SNPs) and mapping of complex genetic disorders Eg. Hypertension; Molecular cytogenetics methods-FISH, CGH, SKY, etc.;Cytogenetics of Cancer (Leukemias); Cytogenetics of Cancer (Solid Tumors).

PART-C 14 hours

Mendelian pedigree patterns; Complications to the basic pedigree patterns; Factors affecting gene frequencies; Non-mendelian characters; Fundamentals of DNA technology and the importance of DNA cloning; Principles of cell-based DNA cloning; Vector systems for cloning different sizes of DNA fragments; Cloning systems for preparing single-stranded DNA and for studying gene expression.

PART-D 14 hours

An overview of mutation, polymorphism, and DNA repair; Simple mutations; Genetic mechanisms which result in sequence exchanges between repeats; Pathogenic mutations; The pathogenic potential of repeated sequences; DNA repair; The history, organization, goals and value of the Human Genome Project; Genetic and physical mapping of the human genome; Model organism and other genome projects; Life in the post-genome (sequencing) era

Reference books:

1. Gillham, N. (2011). *Genes, Chromosomes and Disease*. Pearson

2. Griffiths, A.J.F., Wessler, S.R. and Carroll, S.B. (2012). *An Introduction to Genetic Analysis*. W.H. Freeman Publication, USA.
3. Hein, S. and Mitelman, F. (2009). *Cancer Cytogenetics*. Wiley-Blackwell.
4. Klug, W.S., Cummings, M.R., Spencer, C.A. and Palladino, M.A. (2012). Concepts of Genetics. Pearson.
5. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. Second Completely
6. Revised Edition, Springer-Verlag. (latest edition)
7. Golder N. Wilson, M.D., Ph.D.Clinical Genetics-A Short Course. A John Wiley and Sons,
8. Inc., Publication. (latest edition)

Course Title: Advanced Human Genomics and Advanced Epigenetics	L	T	P	Credits	Marks
Paper Code: HGS.604	4	0	0	4	100

Course Objectives:

PART-A   **14 hours**

Recombinant DNA Technology: Molecular cloning of DNA or RNA fragments in bacterial and eukaryotic systems; Expression of recombinant proteins using bacterial, animal and plant vectors; Isolation of specific nucleic acid sequences, Generation of genomic and cDNA libraries in plasmid, phage, cosmid, BAC and YAC vectors; In vitro mutagenesis and deletin techniques, gene knock out in bacterial and eukaryotic organisms; protein sequencing methods, detection of post translation modification of proteins; DNA sequencing methods, strategies for genome sequencing; Methods for analysis of gene expression at RNA and protein level, large scale expression such as micro array based techniques.

PART-B   **14 hours**

Pharmacogenomics and Overview of Human Genome Project: Concept of individual based treatment, Drug Metabolism; Genetic makeup & 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.

PART-C   **14 hours**

Introduction and molecular mechanisms of Epigenetic: 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

PART-D   **14 hours**

Epigenetic Epidemiology: Effects of diet and environmental agents on epigenetic processes; Impact of microbial infections on the human epigenome; Cancer epigenetic, Role of epigenetic in immune disorders; imprinting disorders in humans.

Reference books:

1. Trygve Tollefsbol (2011). *Handbook of Epigenetics*, Elsevier Publications
2. Nessa Carey (2013). *The Epigenetics Revolution: How Modern Biology Is Rewriting Our Understanding of Genetics, Disease, and Inheritance*. Columbia Univ Pr.
3. Joel Wallach (2014). *Epigenetics: The Death of the Genetic Theory of Disease Transmission. Kindle Publications,* Columbia University Press.
4. Richard C. Francis (2012). *Epigenetics: How Environment Shapes Our Genes*. W.W. Norton and Company, New York.
5. Jocelyn, E. K., Elliot, S. G., 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 Title: Review Writing and Presentation

Paper Code: HGS.698

L	T	P	Credits	Marks
0	0	16	8	200