Syllabus of M.Sc. (HUMAN GENETICS)
Examinations for
2018-2019 and 2019-2020 Academic Sessions

OUTLINES OF TESTS

1. The examination will consist of four semesters i.e. Semester I to IV. Total Marks for M.Sc. course will be 2000, each semester will be of maximum 500 marks divisible in the ratio of 60:40 for theory and practical papers.
2. External Evaluation and Internal Assessment will be in the ratio of 72:28 for theory papers.
3. The Internal Assessment of 21 marks for each theory paper will be based on house test (8.5 marks), assignment and seminar (8.5 marks) and attendance (20%).

Semester-III

Course code.	Course type	Course title	L	T	P	Total credits
Core courses (19 Credits)						
HGL-301	C	Research Design and Methodology	4	0	0	4
HGL-302	C	Human Cytogenetics	4	0	0	4
HGL-303	C	Bioinformatics and Pharmacogenomics	4	0	0	4
HGP-304	C	Practical-I Human Cytogenetics	0	0	3	3
HGP-305	C	Practical-II Bioinformatics	0	0	3	3
HGS-306	C	Seminar	0	0	1	1
Elective course (4 Credits)						
HGL-307	E	Human Biochemical Genetics and Immunogenetics	4	0	0	4
HGL-308	E	To be taken from Massive Open Online Courses (MOOC)	4	0	0	4
		Total Credits	16	0	7	**23**

Semester-IV

Course code	Course type	Course title	L	T	P	Total credits
Core courses (18 Credits)						
HGL-401	C	Analytical Techniques in Human Genetics	4	0	0	4
HGL-402	C	Human Molecular Genetics	4	0	0	4
HGL-403	C	Medical Genomics	4	0	0	4
HGD-404	C	Dissertation	0	0	6	6
Elective courses (4 Credits)						
HGL-405	E	Human Population and Quantitative Genetics	4	0	0	4
HGL-406	E	To be taken from Massive Open Online Courses (MOOC)	4	0	0	4
Open elective courses (Optional Subject)						
–	I	To be taken from other departments	0	0	0	3
		Total Credits	16	0	6	**22**

Semester	Credits			
	Core	Elective	Open Elective (Optional)	Total Credits
I	19	4	0	23
II	18	4	0	22
III	19	4	0	23
IV	18	4	3	22
Total Credits	**74**	**16**	0	**90**

SEMESTER-III

EXTERNAL EVALUATION

Core Courses

Course Code	*Title of Paper*	*Max.Marks*
HGL-301	Research Design and Methodology	54
HGL-302	Human Cytogenetics	54
HGL-303	Bioinformatics and Pharmacogenomics	54
HGP-304	Practical-Human Cytogenetics	100
HGP-305	Practical-Bioinformatics	100

Elective Courses

HGL-307	Human Biochemical Genetics and Immunogenetics	54
HGL-308	To be taken from Massive Open Online Courses	54

INTERNAL ASSESSMENT

4 Theory Papers	84

TOTAL MARKS FOR SEMESTER-III

Theory Papers	300
Practical Papers	200
Total	500

SEMESTER-IV

EXTERNAL EVALUATION

Core Courses

Course Code	*Title of Paper*	*Max.Marks*
HGL-401	Analytical Techniques in Human Genetics	54
HGL-402	Human Molecular Genetics	54
HGL-403	Medical Genomics	54
HGD-404	Dissertation	100

Elective Courses

HGL-405	Human Population and Quantitative Genetics	54
HGL-406	To be taken from Massive Open Online Courses	54

INTERNAL ASSESSMENT

4 Theory Papers	84

TOTAL MARKS FOR SEMESTER-III

Theory Papers	300
Practical Papers	200
Total	**500**

TOTAL MARKS FOR M.Sc. (HUMAN GENETICS) PART-II

SEMESTERS III AND IV SHALL BE AS UNDER

Theory Papers	600
Dissertation	400
GRAND TOTAL	**1000**

The students are required to opt for one elective course in each of the 4 Semesters and one open elective course either in Semester-II or Semester-IV from other departments of the Punjabi University, Patiala.

The department offers an open elective course (Optional Subject) "Concepts and applications of Human Genetics" to the students of other departments of Punjabi University, Patiala.

Final decision regarding the selection of elective course from MOOCs will be taken by the Administrative committee of the Department (ACD)

SEMESTER-III

THEORY PAPERS

HGL-301: RESEARCH DESIGN AND METHODOLOGY

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Section A and B of the question paper and the entire Section C.

SECTION A

Research design: Types of data, Methods of data collection, Sampling design: Descriptive, prospective, Retrospective, Cross-sectional, Longitudinal, Cohort, Case-control studies and Meta analysis. Clinical trials and their utility in Research
Null and Alternative hypothesis: Questionnaire and Schedule. Sampling methods: Probability and non probability methods.Preparation of scientific report.References and their citation.

SECTION – B

Statistical treatment of data: Mean, Mode, Median, Standard Deviation, Standard Error, Coefficient of Variation. Confidence interval and Confidence limits. Tests of significance: Chi square test, students t-test, odds ratio. Correlation and regression.Binomial distribution.Skewness, Kurtosis and Moments.
Collection, Transportation and storage of various biological materials.Hazardous and radiolabelled chemicals and their handling.Sterilization and incineration.Decontamination and disposal.Safety checklist for biomedical labs.

BOOKS SUGGESTED

1. Goode, W.J. and Hatt, P.K. 1989. Methods in Social Research, McGrawhill, Singapore.
2. Gupta, S.P. 1997. Statistical Methods, Sultan Chand & Sons, New Delhi.
3. Weiner, J.S. and Louire, J.A. 1969 Human Biology: A Guide to Field Methods, Blackwell Scientific Publication, Oxford
4. Young, P.V. 1982. Scientific Social Surveys and Research, Prentice-Hall, New Delhi.

HGL-302: HUMAN CYTOGENETICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Section A and B of the question paper and the entire Section C.

SECTION-A

Scope and development of Human Cytogenetics. Types of chromosomal changes: Numerical and Structural chromosomal changes, Uniparentaldiploidy, disomy and genomic imprinting. Constitutive and facultative heterochromatin.Dosage compensation and X-chromosome.
International system for chromosome nomenclature [ISCN] for acquired Cytogenetic abnormalities. Types of chromosome banding and their significance. Sister Chromatid Exchanges (SCE_S), Micronucleus assays. Causes of chromosomal damage.Implications of chromosomal aberrations in gamete formation.Cytogenetics of pregnancy wastage.

SECTION-B

Cancer cytogenetics: constitutional chromosomal instability and Cancer risk. Chromosomal abnormalities in Leukaemia, Lymphomas and solid tumours.Karyotypic evolution in malignancy.Oncogene amplification, chromosome markers and Cancer. Role of specific chromosomal changes in the diagnosis and monitoring of acquiredmalignancy.
Molecular cytogenetics: Principles of Fluorescence in situ hybridization, [FISH], FISH probes. Types of FISH: Q-FISH, T-FISH, GISH, Multiplex FISH (M-FISH) and Spectral karyotyping (SKY). Comparative genomic hybridization [CGH] Applications of FISH and CGH in molecular diagnostics and basic research.

BOOKS SUGGESTED

1. Cumming, M. 2010. Human Heredity: Principles and Issues. 9th ed. Brooks Cole.
2. Czapulkowski, B. 2001. Analyzing Chromosomes: The Basics Bios Scientific Publishers Ltd.
3. Gardner, A., Howell, R. T. and Davies, T. 2008. Human Genetics. Viva Books Pvt. Ltd., New Delhi.
4. Heim, S. and Mitelman, F. (ed.). 2009. Cancer Cytogenetics: Chromosomal and Molecular Genetic Abberations of Tumor Cells, John Wiley & Sons, New Jersey.
5. Korf, B.R. 2006. Human Genetics and Genomics. Blackwell Publishing Co., Oxford.
6. Lewin 2007. Genes IX. Pearson Publishers.
7. Rooney, D. E. (ed.). 2001. Human Cytogenetics. Constitutional Analysis. A Practical Approach. Oxford University Press, Oxford.
8. Schwarzacher, H.G and Wolf, U. (eds.) 1974. Methods in Human Cytogenetic, Springer-Verlag, Berlin.
9. Summer, A.T.2003. Chromosomes: Organization and Function. Blackwell Publishing Co., Oxford.
10. Turpin, R. and Lejeune, J. 1969. Human Afflictions and Chromosomal Aberrations. Pergamon Press, Oxford.
11. Wegner, R. D. 1999. Diagnostic Cytogenetics, Springer-Verlag, Berlin.
12. Yunis, J.J. 1977. New Chromosomal Syndromes, Academic Press, New York.

HGL-303: BIOINFORMATICS AND PHARMACOGENOMICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

Bioinformatics: Introduction and scope of bioinformatics in genetic research. Nucleic acid sequence databases (GenBank, EMBL), Protein Sequence Databases (SWISS-PROT, TrEMBL, PIR), Genome Databases at NCBI, EBI, TIGR, SANGER. Repositories for high throughput genomic sequences: EST, STS and GSS. Search Engines (Entrez and SRS).
Database Similarity Search Tools: BLAST and FASTA. Comparison of FASTA and BLAST. Protein modelling softwares: RASMOL, MOLMOL, INSIGHT, MODDELER, EMBOSS, CHIMERA, PYMOL and their applications.

SECTION – B

Structural Bioinformatics and proteomics: Prediction of protein structure and its function, protein expression analysis, Protein-protein interactions, Principles of docking and ligand design.
Pharmacogenomics: Introduction, application in drug development. Genotoxicity testing, ethical issues. Bioinformatics in pharmaceutical industry: drug discovery, pharmacogenomics of chronic disease (obesity, hypertension, diabetes and cardiovascular diseases).

BOOKS SUGGESTED

1. Attwood, T. K. and Parry-Smith, D. J. 2001. Introduction to Bioinformatics. Pearson Education Asia, Delhi.
2. Chaverive, L.M. and Notridome, C. 2003. Bioinformatics: A Beginners Guide, Wiccy-Dream Tech. India Pvt. Ltd.
3. Dear, P.H. 2007. Bioinformatics, Cold Spring Harbor Laboratory Press.
4. Michael, R.B. 2007. Bioinformatics for Geneticists. A Bioinformatics Primer for the Analysis of Genetic Data. Wiley.
5. Polanski, A. and Kinnel, M. 2007. Bioinformatics. 1st Edition, Springer.
6. Rashidi, H. and Buchler, L.K. 2005. Bioinformatics Basis: Application in Biological Science and Medicine. 2nd Edition. CRC Preven Taylor & Francis Group.
7. Westhead, D.R., Parish, J.H. and Twyman, R.M. 2002. Bioinformatics. Taylor & Francis, London.
8. Xiong, J. 2006. Essential Bioinformatics. Cambridge University Press, Cambridge.

PRACTICAL PAPERS

PRACTICAL HGP-304: HUMAN CYTOGENETICS

Time Allowed: 3 hours | Total Credits 3
Lectures to be Delivered: 90 | Max. Marks: 100
Pass Marks: 35%

Preparation of at least ten karyotypes from metaphase spreads of normal and abnormal chromosome complements.

Blood lymphocyte culture: Preparation of culture medium and other required solutions, Initiation of culture, harvesting lymphocyte culture, slide preparation, staining , G-banding and microscopic analysis of chromosome preparation.

Techniques for Amniotic Fluid cell culture, chorionic villus culture and fibroblast culture (solid tissue specimen)

Microscopic and image analysis systems:Introduction, setting up the microscope using Kohlar's Illumination. Micronucleus assays from exfoliated buccal mucosal cells. Evaluation of X-chromatin frequency from buccal mucosal cell preparations.Cell viability test using Trypan Blue method.

BOOKS SUGGESTED

1. Obe, G. and Basler, A. 1987. Cytogenetics Basics and Applied Aspects. Springer-Verlag, Berlin.
2. Rooney, D. E. (ed.). 2001. Human Cytogenetics. Constitutional Analysis. A Practical Approach. Oxford University Press, Oxford.
3. Schwarzacher, H.G and Wolf, U. (eds.). 1974. Methods in Human Cytogenetics, Springer-Verlag, Berlin.
4. Turpin, R. and Lejeune, J. 1969. Human Afflictions and Chromosomal Aberrations. Pergamon Press, Oxford.
5. Yunis, J.J. 1977. New Chromosomal Syndromes. Academic Press, New York.

PRACTICAL HGP-305: BIOINFORMATICS

Time Allowed: 3 hours | Total Credits 3
Lectures to be Delivered: 90 | Max. Marks: 100
Pass Marks: 35%

Getting familiar with user interface of Nucleic sequences (EMBL, GenBank, DDBJ), Protein sequences (UniProt, SwissProt), 3D structure of macromolecules (PDB), Genome browsers (EnsEMBL, UCSC, ECR), Comparitive Genomics (Integr8), Protein domains (CATH,Pfam, Prosite, InterPro), Gene Ontology (GO).

Retrieving information (for any gene) and its phenotype using OMIM, DNA, RNA and protein information of the gene, to find the SNP/SNPs that have been discovered in the gene, what effect the SNP will have on the DNA/RNA/protein structure of the gene. Changing the format of sequence to FASTA and saving the file.

Retrieving information (for any protein) from the NCBI with Entrez and from various databases with SRS (selecting a database, imposing constraints on field contents, retrieving sequences, combining queries, selecting multiple fields, linking databases, selecting custom fields across multiple databases).

Primer designing using Primer3 and BLAST.

BOOKS SUGGESTED

1. Agostine, M. 2012. Practical Bioinformatics. Garland Science. Taylor and Francis Group.
2. Ye, S. Q. 2008. Bioinformatics: A Practical Approach. Chapman and Hall/CRC.

THEORY PAPER

HGL-307: HUMAN BIOCHEMICAL GENETICS AND IMMUNOGENETICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

Genetic Polymorphism: transient and balanced. Enzyme and protein diversity in human populations: common and rare alleles. Erythrocyte enzyme and plasma protein polymorphisms.Gene nomenclature and genetics of selected triats.

Normal haemoglobins.Haemoglobinopathies: Genetics abnormal haemoglobins, thalassaemias. Sickle cell and foetal haemoglobin. Inborn errors of metabolism: Phenylketonuria, alkaptonuria, galactosemia, glucose-6-phosphate dehydrogenase deficiency.

SECTION – B

Cell of the immune system-Macrophages, B and T lymphcoytes, Dendritic cells, Natural Killer and Lymphokine activated killer cells, Eosinophils, Neutrophils and Mast cells.

Haemopoiesis and differentiation, lymphocyte, trafficking, Antigen-biology, structure and functions of different classes of immunoglobulin.Biology of Super antigens.

Humeral and cell mediated immunity. Cell mediated Cytotoxicity: Mechanism of T cell and NK Cell mediated lysis, Antibody dependent cell mediated Cytotoxicity and macrophage mediated Cytotoxicity. Cytokines and their role in immune regulation.Biology of Complement system, Complement fixation test and assessment of immune complexes in tissues.Hypersensitivity, Immune suppression and immune tolerance.

BOOKS SUGGESTED

1. Abbas, A. K., Lichtman, A. H. and Pober, J. S. 2000. Cellular and molecular immunology, 4th ed. W.B.Saunders, Orlando.
2. Barrett J. T. 1988. Textbook of Immunology: An Introduction to Immunochemistry and Immunobiology. Mosby Publication, New York.
3. Barua, S. 2002. Human Genetics. An Anthropological Perspective Classique Books, Kolkata.
4. Benjamini, E., Coico, R. and Sunshine, G. 2000. Immunology: A Short Course, 4th ed. Wiley-Liss Publisher, New York.
5. Bhasin, M.K. and Chahal, S.M.S. 1996. A Laboratory Manual for Human Blood analysis, Kamla-Raj Enterprises, Delhi.
6. Cavalli-Sforza, L.L. and Bodmer, W.F.1971. The Genetics of Human Populations. Freeman, San Francisco.
7. Giblett, E.R. 1969. Genetic Markers in Human Blood. Davis, Philadelphia.
8. Harris, H. 1975. The Principles of Human Biochemical Genetics. Elsevier/North-Holland, Amsterdam.
9. Jorge, R. and Brassat, O. D. 2006. Immunogenetics and Autoimmune Disease, Springer Science and Business Media, New York.
10. Mourant, A.E., Kopec, A.C. and Domaniewska-Sobczak, K. 1976. The Distribution of the Human Blood Groups and Other Polymorphisms. Oxford University Press, London.
11. Mourant, A.E., Kopec. A.C. and Domaniewska-Sobczak, K. 1978. Blood Groups and Diseases. Oxford University Press, Oxford.

SEMESTER-IV

THEORY PAPERS

HGL -401: ANALYTICAL TECHNIQUES IN HUMAN GENETICS

Time Allowed: 3 hours | Total Credits: 4
Lectures to be Delivered: 60 | Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

Serological and Biochemical Techniques: Antigen antibody reaction: typing of human blood for A1A2BO, MNSs, Rhesus (CcDEe), Kell, Duffy, Kidd and P blood groups, detection of ABH substance in saliva, Typing of human haemolysates for blood protein haemoglobin and red cell enzymes ESD, PGM1, ACP, GLO1, ADA and AK1 and serum proteins by horizontal gel electrophoresis. Single cell gel electrophoresis (COMET Assay)

Clinical Genetic Techniques: Peripheral blood lymphocyte culture (PBLC), Micronuclei assay, Enzyme-linked immune-sorbent assay (ELISA): Indirect ELISA, Sandwitch ELISA, Competetive ELISA, Multiple and portable ELISA, Flow cytometry, Bioelectrical impedance analysis (BIA), Biochemistry analysis.

SECTION-B

Techniques for Protein Analysis: Polyacrylamide gel electrophoresis (PAGE), Two dimensional gel electrophoresis (2DGE): first dimension-Isoelectric focusing with immobilized pH gradient (IEF-IPG), second dimension-SDS-PAGE, Chromatographic methods for protein identification, Dot blot, Western blotting, protein tagging.

Molecular Genetic Techniques: DNA/RNA extraction, Polymerase chain reaction (PCR), quantitative polymerase chain reaction (qPCR), reverses transcription polymerase chain reaction (RT-PCR), Single strand conformation polymorphism (SSCP), Gene mapping, gene cloning, DNA sequencing, DNA microarray, Exome sequencing.

BOOKS SUGGESTED

1. Bhasin, M.K. and Chahal, S.M.S. 1996. A Laboratory Manual for Human Blood analysis, Kamla-Raj Enterprises, Delhi.
2. Brown, T. A. 2000. Essential Molecular Biology. A Practical Approach,vols. I and II. Oxford University Press.
3. Vandenbergh, D. J. 2010. Techniques of Molecular Genetics. Cambridge University Press.
4. Harris, H. 1975. The Principles of Human Biochemical Genetics. Elsevier/North-Holland, Amsterdam.
5. Kamp RozaMaria, Calvete Juan J andCholi-Papadopoulou Theodora (eds). 2004. Methods in Proteome and Protein Analysis. Springer Verlag.
6. Glynn and M. andDrake, W. 2012. Hutchison's Clinical Methods: An Integrated Approach to Clinical Practice.Elseviers.
7. Robyt, J. F. and White, B. J.1990. Biochemical Techniques: Theory and Practice. Waveland PressInc., Illinois.

HGL-402:HUMAN MOLECULAR GENETICS

Time Allowed: 3 hours | Total Credits: 4
Lectures to be Delivered: 60 | Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

DNA organisation in Eukaryotic Chromosomes, DNA markers: RFLPs, VNTRs, CNVs, STRs, SNPs, SINEs, LINEs, mitochondrial markers, DNA genotyping and sequencing, concept of linkage disequilibrium (LD), Haplotypes and tag SNPs

Molecular basis of mutations: gene mutations, sources of mutations, types of mutations, effects of mutations, mutation detection, mutation rate, site-directed mutagenesis, transposable elements.

SECTION-B

Recombinant DNA technology and its applications: gene structure and expression, cell based DNA cloning, cell free DNA cloning, Nucleic acid hybridization. Blotting techniques: Northern, Southern and Western blotting.DNA profiling, DNA fingerprinting and applications.

Principles and strategies in identifying disease genes: gene tracking, single gene disorders or Mendelian disorders, complex disorders and methods to study them: family and population based association studies, genome wide association studies. Molecular genetics of complex diseases and mitochondrial diseases.

BOOKS SUGGESTED

1. Nelson, D. L. and Cox, M. M. 2008. Lehninger Principles of Biochemistry,5th ed.Benjamin CummingsPublisher.W. H. Freeman.
2. Lewin, B. 2008. Genes XII. Pearson Prentice Hall, New Jersey.
3. Alberts, B., Johnson, A., Lewis, J., Raff, M., Roberts, K. and Walter, P. 2007. Molecular Biology of the Cell.Garland Science.
4. Sudbery, P. and Sudbery, I. 2009. Human Molecular Genetics, Pearson Education Limited.
5. Strachan, T. and Read, A.P. 1999. Human Molecular Genetics. John Wiley & Sons, Singapore.

HGL-403: MEDICAL GENOMICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

Definition and scope of medical genomics, Impact of genomics in medicine. Epigenome: Histone modifications, DNA methylation, Gene Expression, Small RNA Expression. Encyclopedia of DNA elements (ENCODE) and Encyclopedia of gene and gene variants (GENCODE) projects.
Genomic testing of complex disorders: Role of PCR in disease diagnosis, DNA sequencing, Gene therapy, Prenatal and pre-implantation Genetic diagnosis, Gene expression and Personalized medicine

SECTION-B

Public health genomics: Implications of Human Genome project for medical genomics. Exome and Genome-wide association studies (GWAS) for common diseases. Genomics as a probe for prognostics, diagnostics and therapeutics of multifactorial disorders.
Ethics in genome research: Ethical, legal and social issues in medical genomics. Genetic discrimination and its impact on physician and researcher.

BOOKS SUGGESTED

1. Brown, S.M. 2003. Essentials of Medical Genomics. Wiley-Liss.
2. Jorde, L.B, Carey, J.C. and Bamshad, M.J. 2009. Medical Genetics. Elsevier Health Sciences.
3. Khoury, M. J. (ed.). 2010. **Human Genome Epidemiology:** *Building The Evidence for Using Genetic Information to Improve Health and Prevent Disease. Oxford University Press, Oxford.*
4. Primrose, S.B and Tayman, R. 2004. Genomics: Application in Human Biology, Blackwell Publications.
5. Quackenbush, J. 2011. The Human Genome. Charlesbridge Publishing.
6. Young, I.D. 2005. Medical Genetics.Oxford University Press, Oxford.

HGD-404:DISSERTATION

Time allowed: 3 hours
Total dissertation hours: 180

Total Credits 6
Max. Marks: 200
Pass Marks: 35%

The candidate will be required to submit a Dissertation based on field work on a research topic assigned by the Department

HGL-405: HUMAN POPULATION AND QUANTITATIVE GENETICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 4
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

SECTION-A

Aim and scope of population genetics.Human population as a unit of study.Mendelian population.Gene pool.Genes in families and populations.Hardy-Weinberg law. Factors affecting genetic equilibrium: Mutation, Selection, Hybridization and Genetic Drift.
Random and non randommatings.Consanguinity and inbreeding, Inbreeding coefficient. Consequences of inbreeding for mendelian populations, average inbreeding in human populations. Genetic load. Concept and significance of genetic admixture and its estimation from allele frequency data.

SECTION-B

Methods of allele frequency calculations: gene counting, square root and maximum likelihood methods. Measures of genic variation, genetic differentiation and genetic distance, graphic display methods
Quantitative trait loci (QTL).Polygenic variation. Genetic and environmental components of variance, heritability estimation: parent-offspring, Sib and twin data. Linkage and crossing over, Genetic maps and Lod scores.Gene mapping.

BOOKS SUGGESTED

1. Cavalli-Sforza, L.L. and Bodmer, W.F. 1971. The Genetics of Human Populations. Freeman, San Francisco.
2. Klug, W.S. and Cumming, M.R. 1977. Concepts of Genetics. Prentice Hall New, Jersey.
3. Lavitan, M. and Montagu, A. 1977. Textbook of Human Genetics, Oxford University Press, Oxford.
4. Hartl, D. L. and Clark, A. G. 1997. Principles of Population Genetics. SinauerAssociates, Sunderland, Massachusetts.
5. Smith, J. M. 1998. Evolutionary Genetics. Oxford University Press, Oxford.
6. Hamilton, M.B. 2009. Population Genetics. Wiley-Blackwell.
7. Balding, D.J., Bishop, M. and Cannings, C.C. (eds.). 2007. Handbook of Statistical Genetics, vols. 1&2, 3rd ed. John Wiley and Sons.

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Open Elective Course (Optional Subject)

CONCEPTS AND APPLICATIONS OF HUMAN GENETICS

Time Allowed: 3 hours
Lectures to be Delivered: 60

Total Credits: 3
Max. Marks: 75
Theory Marks: 54
Internal Assessment: 21
Pass Marks: 35%

INSTRUCTIONS FOR THE PAPER-SETTER

The question paper will consist of three sections: A, B and C. Sections A and B will have four questions from the respective sections of the syllabus and will carry 8 marks each. Section C (Compulsory) will consist of 11 short-answer type questions (carrying 2 marks each) which will cover the entire syllabus uniformly.

INSTRUCTIONS FOR THE CANDIDATE

The Candidate is required to attempt two questions from each Sections A and B of the question paper and the entire Section C.

Section-A

Structure of cell, Cell cycle, DNA structure, Genetic code, Normal Chromosome Complement, Chromosomal aberrations. Brief history of Human Genetics, Genes, Alleles, Lethal and sub-lethal genes, Penetrance and Expressivity. Genetic disorders due to chromosomal and single gene defects. Understanding human genetic variations: role of genetic variations in complex diseases, genetic susceptibility. .

Section-B

Modes of inheritance: autosomal codominant, autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked, Polygenic/complex inheritance and extra chromosomal inheritance. Construction of pedigrees and their interpretation for assessing modes of inheritance. Genetic screening and genetic counseling. Gene based therapeutic approaches, personalized medicine and gene therapy.