

FACULTY OF LIFE SCIENCES

Syllabus

For

M.Sc. HUMAN GENETICS (Credit Based Evaluation & Grading System)

(SEMESTER: I - IV)

Examinations: 2019-20



Guru Nanak Dev University
Amritsar

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M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)

Credit Structure of M.Sc. Human Genetics

Semester	Credits			Total
	Core	Elective	Interdisciplinary/ Optional	
I	24	-	-	24
II	18	3	-	21
Total Credits	39	6	-	45
III	17.5	3	4	24.5
IV	10.5	3	8	21.5
Total Credits	28	6	12	46

M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)

SCHEME

Semester-I

Course No.	C/E/I	Course Title	L	T	P	Total Credits	Total Marks		
							Max.	Mid Sem	End Sem
Core Courses (24 Credits)									
HGL-401	C	Molecular Genetics	3	0	0	3	100	20	80
HGL-402	C	Principles of Human Genetics	3	0	0	3	100	20	80
HGL-404	C	Human Anatomy and Physiology-I	3	0	0	3	100	20	80
HGL-405	C	Computer Applications	3	0	0	3	100	20	80
HGL-407	C	Human Growth and Body Composition	3	0	0	3	100	20	80
MTL-261	C	Biostatistics	3	1	0	4	100	20	80
HGP-402	C	Principles of Human Genetics and Molecular Genetics Practical	0	0	2	2	100	-	-
HGP-404	C	Human Anatomy Practical	0	0	1.5	1.5	100	-	-
HGP-407	C	Human Growth and Body Composition Practical	0	0	1.5	1.5	100	-	-

NOTE: PSL-053 ID Course Human Rights & Constitutional Duties (**Compulsory Paper**) Students can opt. this paper in any odd semester. This ID Paper is one of the total ID Papers of this course.

M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)

Semester-II

Course No.	C/E/I	Course Title	L	T	P	Total Credits	Total Marks		
Core Courses (21 Credits)							Ma	Mid	End
							x.	Sem	Sem
HGL-406	C	DNA Technology	3	0	0	3	100	20	80
HGL-452	C	Human Cytogenetics	3	0	0	3	100	20	80
HGL-454	C	Human Anatomy and Physiology-II	3	0	0	3	100	20	80
HGL-457	C	Human Biochemical Genetics and Immunogenetics	3	0	0	3	100	20	80
HGL-458	C	Research Design and Methodology	2	1	0	3	100	20	80
HGP-452	C	Lab Exercises in Human Cytogenetics	0	0	1.5	1.5	100	-	-
HGP-454	C	Human Physiology and Immunology Practical	0	0	1.5	1.5	100	-	-
Elective Course (3 Credits)									
HGL-462	E	Lab Design and Biosafety Guidelines	3	0	0	3	100	20	80
HGL-463	E	Tissue Culture and Microscopy	3	0	0	3	100	20	80

Note : The students are required to opt one 'Elective' paper.

M.Sc. HUMAN GENETICS
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Semester-III

Course No.	C/E/I	Course Title	L	T	P	Total Credits	Total Marks		
							Max.	Mid Sem	End Sem
Core Courses (24.5 Credits)									
HGL-502	C	Functional Genomics	3	0	0	3	100	20	80
HGL-505	C	Human Embryology and Developmental Genetics	3	0	0	3	100	20	80
HGL-508	C	Essentials of Medical Genetics	2	1	0	3	100	20	80
HGL-510	C	Human Population Genetics and Variation	3	1	0	4	100	20	80
HGS-506	C	Seminar	0	0	1	1	100	20	80
HGP-508	C	Exercises in Medical Genetics	0	0	1.5	1.5	100	-	-
HGP-502	C	Functional Genomics Practical	0	0	2	2	100	-	-
Elective Course (3 Credits)									
HGL-511	E	Immunogenetics in Health and Disease	3	0	0	3	100	20	80
HGL-514	E	Genetics of Human Pathogens and Infectious Diseases	3	0	0	3	100	20	80
Interdisciplinary/Optional Course									
-	I	To be taken from outside the department	4	0	0	4	100	20	80

NOTE:

1. The students are required to opt one 'Elective' paper.
2. The students are also required to take one paper (Interdisciplinary/Optional) of 4 credits of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.
3. PSL-053 ID Course Human Rights & Constitutional Duties (**Compulsory Paper**) Students can opt. this paper in any odd semester. This ID Paper is one of the total ID Papers of this course.

M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)

Semester-IV

Course No.	Course Category	Course Title	L	T	P	Total Credits	Total Marks		
							Max.	Mid Sem	End Sem
Core Courses (21.5 Credits)									
HGL-551	CC	Perspectives of Human Genome and Bioethics	3	0	0	3	100	20	80
HGL-554	CC	Structural Biology and Pharmacogenomics	3	0	0	3	100	20	80
HGL-557	CC	Clinical Genetics	2	1	0	3	100	20	80
CORE COURSE PRACTICAL									
HGP-557	CC	Exercises in Clinical Genetics	0	0	1.5	1.5	100	-	-
*DISCIPLINE SPECIFIC ELECTIVE COURSE									
HGL-561	DSE	Regulation in Human Genetics Research	3	0	0	3	100	20	80
HGL-562	DSE	Concepts of Biological Demography	3	0	0	3	100	20	80
HGP-563	DSE	Research Training Report**	0	0	3	3	100	-	-
GENERIC ELECTIVE COURSE (8 Credits) (Any of Two)									
	GEC	To be taken from outside the department	4	0	0	4	100	20	80
	GEC	To be taken from outside the department	4	0	0	4	100	20	80
EDL-062	GEC	Assessment of Learning	4	0	0	4	100	20	80
Total Credits			19	1	5	22			

- *The students are required to op one **Discipline Specific Elective** paper.
- The students are also required to take two **Generic Elective Course (GEC)** (Interdisciplinary / Optional) of 4 Credits each of their choice from any other department of GNDU Campus.
- **Topic of **Research Training Report** and name of supervisor to be finalized during the end of Semester-II.
- **CC – Core Course ; EC-Elective Course ; DSE – Discipline Specific Elective Course; GEC – Generic Elective Course; AEC – Ability Enhancement Course; AECC – Ability Enhancement Compulsory Course; SEC – Skill Enhancement Course.**

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Continuous Evaluation Grading System)

HGL-401 MOLECULAR GENETICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Central dogma of life, Composition and structure of nucleic acids, Factors affecting DNA stability, Mechanisms of DNA replication in prokaryotes and eukaryotes, DNA markers: Tandem repeat polymorphisms, Single nucleotide polymorphisms (SNPs), Random amplified polymorphic DNA (RAPD) markers and their applications, DNA damage, DNA repair mechanisms.

Section-B

Different types of RNA polymerases and their role, Transcription, Promoters, enhancers and regulators of transcription, Post-transcriptional processing of primary transcript, RNA splicing, 5'-Capping 3'-Polyadenylation, Alternative splicing – different types and significance, Primary splicing defects, Secondary splicing defects, RNA editing, Transport and targeting of RNA, Post-transcriptional gene silencing.

Section-C

Gene expression regulation-operon and regulon, Lac operon, tryptophan operon, positive and negative regulation, Regulation by transcription attenuation in bacteria – Transcription antitermination by tRNA charging, Protein mediated termination or antitermination, Factors dependent antitermination.

Section-D

Analysis of human gene structure – construction of cDNA and genomic libraries and their applications, 5' rapid amplification of complementary ends (RACE), 3' RACE, Primer extension method to identify transcriptional start site, Genetic code, Prokaryotic and eukaryotic ribosomes, Mechanism of translation, Post-translation modifications, Inhibitors of protein synthesis,

Books Recommended:

1. Alberts, B., Bray, D., Lewis, J., Raff, M., Roberts, K. and Watson, J.D. (2007). Molecular Biology of the Cell. Garland Press, USA, 5th ed.
2. Brown, T.A. (2005). Genetics: A Molecular Approach. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
3. Brown, T.A. (2006). Genomes3. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
4. Brown, T.A. (2010). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 6th ed.
5. Hartl, D.L. and Jones, E.W. (2005). Genetics: Analysis of Genes and Genomes. Jones and Bartlett Publishers, Inc., USA, 6th ed.
6. Korf, B.R. (2007). Human Genetics and Genomics. Blackwell Scientific Publications, USA, 3rd ed.
7. Krebs, J.E. (2010). Genes-X. Jones and Barlett Publishers, Inc., USA.
8. Lewin, B. (2011). Genes-X. Jones and Barlett Publishers, Inc., USA. 10th ed.
9. Lodish, H., Baltimore, D., Berk, A., Zipursky, S.L., Matsudaira, P. and Daniell, J. (2004). Molecular Cell Biology. W.H. Freeman and Co., San Fransisco, 5th ed.
10. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

HGL-402 PRINCIPLES OF HUMAN GENETICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Overview of historical milestones in Human Genetics, Fields of Human Genetics, Molecular organization of nuclear and mitochondrial genome, Transposable elements in Human Genome, Genetic maps, Physical maps, Cytological maps, Chromosome maps

Section-B

Modes of inheritance in humans, Mendelian and Non-Mendelian inheritance, Multifactorial inheritance and quantitative traits, Threshold model and recurrence risks, Dosage compensation and X-inactivation Genetics of sex determination, Sex-linked, sex-limited and sex-influenced traits, Manifesting heterozygotes, Mosaics and chimeras, True- and pseudo-hermaphrodites, Sex ratio

Section-C

Various types of mutations, Role of radiations and chemicals in inducing mutations, Effects of mutations, Mutation rates in humans, Pleiotropy. Genetic basis of cancer — Proto-oncogenes, oncogenes and tumour-suppressor genes,

Section-D

Chromosomal basis of neoplasia, Cytogenetic basis of variation in chromosome number and structure, Marker chromosomes. Somatic cell hybridization in cytogenetic analysis, Applications of amniocentesis, chorionic villus sampling (CVS) and fluorescence *in situ* hybridization (FISH).

Books Recommended:

1. Strachan, T. (2015). Genetics and Genomics in Medicine. Garland Science, USA, 1st ed.
2. Weinberg, R.A. (2014). The Biology of Cancer. Garland Science, USA, 2nd ed.
3. Tobias, E.S. (2013). Essential Medical Genetics. Wiley Blackwell, UK, 6th ed.
4. Cummings, M.R. (2009). Human Genetics. Cengage Learning, USA.
5. Farzaneh, F. and Cooper, D.N (Eds.) (1995). Functional Analysis of the Human Genome. Bios Scientific Publishers Ltd., Oxford.
6. Gardner, E.J. (2008). Human Genetics. Viva Books Pvt. Ltd., India.
7. Hancock, J.H. (2008). Molecular Genetics. Viva Books Pvt. Ltd., India.
8. Harper, P.S. (2006). First Years of Human Chromosomes. Scion, USA.
9. Korf, B.R. and Irons, M.B. (2013). Human Genetics and Genomics. Wiley-Blackwell, Malaysia, 4th ed.
10. Sambamurty, A.V.S.S. (2006). Genetics. Narosa Publishing House, New Delhi.
11. Strachan, T. and Read, A. (2011). Human Molecular Genetics. Bios Scientific Publishers, Oxford, 4th ed.
12. Sumner, A.T. (2003). Chromosomes: Organization and Function. Blackwell Publishing Co., Oxford.
13. Tamarin, R.H. (2002). Principles of Genetics. Tata McGraw Hill, USA, 7th ed.
14. Therman, E. and Miller, O.J. (2001). Human Chromosomes. Springer-Verlag, New York, 4th ed.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

HGL-404 HUMAN ANATOMY AND PHYSIOLOGY-I

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Introduction to anatomical terms, Classification of bones, Histology of bones, Ossification, Growth, Role of bone in calcium homeostasis, Fracture and repair of bones, Structure and classification of joints.

Section-B

Classification of muscles, Structure of smooth, cardiac, skeletal muscle, neuromuscular junction, Physiology of muscle contraction. Brief description of constituent parts of digestive system, Physiology of digestion, absorption and taste.

Section-C

Blood composition and hemopoiesis, Functions of blood, Blood clotting, Blood pressure, Brief description of lymphatic system, Structure of heart, Cardiac cycle, Systemic and pulmonary circulation, Structure and types of blood vessels.

Section-D

Brief anatomical description of respiratory system, Physiology of respiration, Role of hemoglobin in respiration, Regulation of respiration, structure and physiology of olfactory receptor.

Books Recommended:

1. Drake, R., Vogl, W. and Mitchell, A. (2015). Gray's Anatomy for Students. Churchill Livingstone, USA.
2. Guyton, A.C. and Hall, J.E. (2016). Textbook of Medical Physiology. Elsevier Publications, New York.
3. Standring, S. (2015). Gray's Anatomy. Churchill Livingstone, USA. 40th ed.
4. Tortora, G.J. and Henderson, S.R. (2013). Principles of Anatomy and Physiology. Harper Collins College Publishers.

M.Sc. HUMAN GENETICS (SEMESTER-I)
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HGL-405 COMPUTER APPLICATIONS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Computer fundamentals, Internet basics and MS-Office 2003, Introduction to digital computers, Organization, Number system, I/O devices, Storage devices, Introduction to internet and its applications – www, email, ftp. MS-Windows basics, MS-Word – Meaning of Word-Processing, Creating, Saving, Printing documents, Formatting, Spell-Check, Adding page numbers, Header and Footer, Macros, Creating tables, Converting table to text and vice-versa, Mail Merge.

Section-B

MS-Excel – Spreadsheets, Using different types of formulae, Creating graphs and charts, Exporting charts to MS-Word, MS-PowerPoint – Creating presentations, Formatting, Adding effects and timings. Types of errors and level of significance, Tests of significance (F and t-test), Chi-square tests.

Section-C

Data analysis and database – Brief description and tabulation of data, Measure of central tendency and dispersion – Mean, Median, Mode, Range, Standard Deviation, Variance and Correlation coefficient using SPSS. Introduction to Data, Information, Database, DBMS (Advantages and disadvantages), Introduction to SQL (Data retrieval).

Section-D

Virtual library and some useful sites on Internet – Searching MEDLINE on the Pubmed system from National Centre for Biotechnology and Information. Assessing full text journals on the internet and printing articles using EndNote.

Books Recommended:

1. Sinha, P.K. (1992). Computer Fundamentals.
2. Peter Norton's Introduction to Computers, 6th ed.
3. Windows Based Computer Courses, Sumit Kumar, JBD Publishers.
4. Gupta, S.C. (2004). Fundamentals of Statistics. Himalaya Publishing House.

Website Links

Databases (Genes Bank), search tools and software at

<http://www.ncbi.nlm.nih.gov>.

Restriction enzyme site digestion webcutter2.0 at

<http://www.firsmarket.com/cutter/cut2.html>.

PCR and multiplex PCR guide and troubleshooting at

<http://www.med.yale.edu/genetics/ward/tavi/Trblesht.html>

Image analysis program at

<http://www.scioncorp.com>.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

HGL-407 HUMAN GROWTH AND BODY COMPOSITION

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Definitions of growth and development, Laws of growth, Methods of studying growth, Assessment of normal growth, Catch-up growth, Undernutrition, Underweight, Stunting, Wasting, Protein energy malnutrition (PEM), Marasmus, Kwashiorkor

Section-B

Identification of abnormal growth and growth failure, Growth references, Growth standards, Growth charts, Velocity of growth Growth patterns in various genetic disorders.

Section-C

Somatic growth during prenatal and post natal life, Assessment of prenatal age, Ultrasound measurements of prenatal growth, Regional growth disorders, Short and tall stature, Delayed puberty, Precocious puberty, Chromosomal aberrations and growth, Developmental disabilities.

Section-D

Conceptual models of body composition, Elementary composition of human body, Reference man and reference woman, Classification of human physique and somatotyping.

Books Recommended:

1. Cummings, M.R. (2009). Human Genetics. Cengage Learning India Pvt. Ltd. New Delhi.
2. Ghai, O.P., Paul, VK. Arvind Bagga. (2010). Essential Pediatrics. CBS Publishers and Distributors, New Delhi
3. Harrison, G.A. Weiner, J.S. Tanner, J.M. and Barnicot, N.A. (1990). Human Biology. Oxford University Press.
4. Ulizaszek, S.J., Johnston, F.E. and Preece. M.A. (1998) The Cambridge Encyclopedia of Human Growth and Development. Cambridge University Press.
5. Forbes, C.B. (1987). Human Body Composition. Springer-Verlag, New York, Berlin.
6. Gibney, M.J., Macdonald, I.A. and Rocho, H.M. (2004). Nutrition and Metabolism. Blackwell Publishing Co., Oxford.
7. Harrison, G.A., Weiner, J.S., Tanner, J.M. and Barnicot, N.A. (1990). Human Biology. Oxford University Press.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

MTL-261-BIOSTATISTICS

Credit hrs.		
L	T	P
3	1	0

Time: 3 Hours

Max. Marks: 100
Mid Semester Marks : 20
End Semester Marks : 80

Mid Semester Examination: 20% weightage
End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Note:- The scope of this paper is restricted only to the applications of various statistical techniques. The mathematical derivations of various results are excluded.

Section-A

Statistical Methods — Collection of data, Frequency distribution and, Measures of Central Tendency, Dispersion.

Correlation and Regression — Relationship between variables, Covariance, Karl-Pearson's Correlation Coefficient, Spearman's rank Correlation Coefficient, Least square technique for regression lines (without proof), Regression Coefficients, Relationship between Correlation analysis and Regression Analysis.

Section-B

Probability — Mathematical definition of probability of an event, Use of permutations and combinations in calculations of Probability, Conditional probability, Additive and Multiplication law of Probability, Random Variables and its pmf, pdf, cdf, Mathematical expectation and variances, Theoretical Distributions: Binomial, Poisson and normal, Properties of these distributions (applications only).

Section-C

Hypothesis Testing — Sample, Population, Statistics and Parameters, Null Hypothesis, Level of significance, Definitions of Chi-square, 't' and 'F' variates and their pdfs only, Applications of these distributions in testing of hypothesis.

Section-D

Large sample test- Testing of significance of proportion in single population, Testing of equality of proportions in two populations, Testing of significance of mean in single population, Testing of equality of means in two populations.

Analysis of Variance — Meaning of analysis variance with linear models, Analysis of variance for one-way classified data, Analysis of variance for two-way classified data with one observation for cell.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

Text Books

1. Fowler, J., Cohen, L. and Jarvis, P. (1998). Practical Statistics for Field Biology. John Wiley and Sons, 2nd ed. [Chapters: 4,5,6,7,(7.1–7.6), 9 (9.1–9.4), 12 (12.1–12.7), 13 (13.1–13.4, 13.6,13.7), 14 (14.1–14.5, 14.7), 15 (15.3–15.8, 15.10–15.11), 16 (16.9–16.13), 17 (17–.1– 17.3, 17.5,17.6,17.8)].
2. Raghavarao, D. (1983). Statistical Techniques in Agricultural and Biological Research Oxford and IBH Publishing Co. [Chapters: 2,3,4,5,7,8,9 and 10].

Reference Books:

1. Bland, M. (2006). An Introduction to Medical Statistics. Oxford University Press, 3rd ed.
2. Finney, D.J. (1980). Statistics for Biologists. Chapman and Hall Ltd.
3. Hoel, P.G. (1971). Elementary Statistics. John Wiley and Sons, 3rd ed.
4. Ross, S.M. (2005). Introductory Statistics. Academic Press, 2nd ed.
5. Wayne, W, Daniel (1999). Biostatistics: A Foundation for Analysis in Health Sciences. John Wiley and Sons, 7th ed.
6. Woodworth, G. (2004). Biostatistics: A Bayesian Introduction. John Wiley and Sons.

M.Sc. HUMAN GENETICS (SEMESTER-I)
(Credit Based Evaluation & Grading System)

HGP-402 PRINCIPLES OF HUMAN GENETICS AND MOLECULAR GENETICS
PRACTICAL

MAX. MARKS : 100

Credits: 0-0-2

Numericals on Mendelian and non-Mendelian inheritance in humans, To study pedigree patterns, Numericals on pedigree analysis — Autosomal patterns, X-linked patterns, Y-linked patterns, Mitochondrial inheritance patterns, To study morphogenetic traits with regard to their gene frequencies, variation patterns, Prepare genetic individuality charts for morphogenetic traits, To test PTC tasting ability in a random sample and calculate gene frequencies for the taster and nontaster alleles, Frequency of Barr body in buccal smear, Drumsticks in blood smear as means of sex determination, To study fingerball and palmar dermatoglyphics and calculate indices, To test for colour blindness using Ishihara charts. Sterilization of glassware and plasticware, Preparation of stock solutions, Preparation of working solutions from stock solutions, Extraction of DNA from human buccal cells by inorganic method, Quality checking of DNA by agarose gel electrophoresis, Quantification of DNA, Amplification of genomic DNA by PCR.

M.Sc. HUMAN GENETICS (SEMESTER-I)
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HGP-404 HUMAN ANATOMY PRACTICAL

MAX. MARKS : 100

Credits: 0-0-1.5

Anatomical positions — Superior, Inferior, Anterior, Medial, Posterior, Lateral, Proximal, Distal, External, Internal

Parietal, Visceral, Cavities and planes of human body.

Classification of bones

Skull – different views

Vertebrae, Typical, atypical-C1, C2, C7, T1, T10, T11, T12,L5

Sternum, Pectoral girdle, Bones of upper and lower limbs, Pelvic girdle

Sexual dimorphism in skeleton.

Overview of Types of joints and Movements at joints

M.Sc. HUMAN GENETICS (SEMESTER-I)
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HGP-407 HUMAN GROWTH AND BODY COMPOSITION
(PRACTICAL)

MAX. MARKS : 100

Credits: 0-0-1.5

Definition and location of various landmarks on body, Technique of various anthropometric measurements, Growth curves, Direction of growth curve, Assessment of growth status and growth rate, Heath and Carter method of somatotyping.

**M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)**

HGL-406 DNA TECHNOLOGY

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Principles and applications of Agarose gel, Polyacrylamide gel, Two-dimensional gel (2DE), Denaturing gradient gel and Pulse field gel electrophoresis. Polymerase chain reaction (PCR), Types and applications of PCR, Enzymes used in DNA technology: Restriction endonucleases, Polymerases, Ligases, Kinases and phosphatases.

Section-B

Principles and applications of Single-strand conformational polymorphism (SSCP), Simple sequence length polymorphism (SSLP), Restriction fragment length polymorphism (RFLP), Denaturing high-pressure liquid chromatography (dHPLC), Sanger DNA sequencing, Multiplex ligation dependent probe amplification (MLPA).

Section-C

Nucleic acid probes, Methods of labelling probes, Principles and applications of Southern blotting, Northern blotting, Western blotting, Dot blotting, DNA microarray, Serial analysis of gene expression (SAGE) and DNA foot printing.

Section-D

An overview of recombinant DNA (rDNA) technology, Cloning vectors: Plasmids, Cosmids, Phagemids, Artificial chromosomes, Shuttle vectors, Linkers and adaptors, Construction of cDNA and genomic DNA libraries, Identification of specific cloned sequences, Applications of recombinant DNA technology, Safety issue in rDNA experiments.

Books Recommended

1. Brown, T.A. (2016). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 7th ed.
2. Dieffenbach, C.W. and Dveksler, G.S. (2003). PCR Primer: A Laboratory Manual. Cold Spring Harbor Press, 2nd ed.
3. Freifelder, D.C. (2008). Molecular Biology. Narosa Publishing House, New Delhi, 2nd ed.
4. Green, M.R. and Sambrook, J (2012). Molecular Cloning: A Laboratory Manual. Cold Spring Harbor, New York, Vol. 1, 2&3. 4th ed.
5. Hartl, D.L. and Jones, E.W. (2011). Genetics: Analysis of Genes and Genomes. Jones and Barlett Publishers, USA, 7th ed.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

6. Hartl, D.L. and Ruvolo M (2012). Genetics: Analysis of Genes and Genomes. Jones and Barlett Publishers, USA, 8th ed.
7. Krebs J.E., Goldstein, E.S. and Kilpatrick, S.T. (2012). Genes-XI. Jones and Barlett Publishers, Inc., USA.
8. Lewin, B. (2011). Genes-X. Jones and Barlett Publishers, Inc., USA.
9. Pierce, B.A. (2012). Genetics: A Conceptual Approach. W.H. Freeman and Co., New York, 4th ed.
10. Pierce, B.A. (2017). Genetics: A Conceptual Approach. W.H. Freeman and Co., New York, 7th ed.
11. Sambrook, J., David, W. and Russell. (2001). Molecular Cloning: A Laboratory Manual. Cold Spring Harbor, New York, Vol. 1, 2&3. 3rd ed.
12. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGL-452 HUMAN CYTOGENETICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Human chromosome organization and structure, Chromatin, Nucleosome, Solenoid model, Centromeres, Neocentromeres, Kinetochores, Telomeres, Human artificial chromosomes.

Section-B

Human mitosis and meiosis, Mitotic cell-cycle, Cell-cycle progression and check points, Chromosome dynamics in M-phase and its regulation, Role of condensins, Cohesins.

Section-C

Animal cell culture technique — Primary culture, Secondary culture, Serum-free cultures, Cell lines. Chromosome banding techniques — G,C,R,Q,T,NOR, High-resolution banding, Molecular correlates of chromosome bands, Fragile sites, Chromosome nomenclature and cytogenetic notation for G-banded chromosomes,

Section- D

Genetic toxicology, Chromosomal instability syndromes – Ataxia telangectasia, Fanconi anemia, Bloom's Syndrome, Nijmegen breakage syndrome
 Microscopy— Introduction to light, fluorescent and confocal microscopy. Fluorescent in situ hybridization, Comparative genomic hybridization

Books Recommended:

1. Cummings, M.R. (2009). Human Genetics. Cengage Learning, USA.
2. Czepulkowski, B. (2004). Analysing Chromosomes. Bios Scientific Publishers Ltd., Oxford,
3. Farzaneh, F. and Cooper, D.N (Eds.) (1995). Functional Analysis of the Human Genome. Bios Scientific Publishers Ltd., Oxford.
4. Freshney, R.I. (2010). Animal Cell Culture: A Practical Approach. IRL Press, Oxford, 2nded.
5. Gardner, E.J. (2012). Human Genetics. Viva Books Pvt. Ltd., India.
6. Gersen, S.L. and Keagle, M.B. (2005). The Principles of Clinical Cytogenetics. Humana Press, USA, 2nd ed.
7. Hancock, J.H. (2008). Molecular Genetics. Viva Books Pvt. Ltd., India.
8. Weinberg, R.A. (2014). The Biology of Cancer. Garland Science, USA, 2nded.

**M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)**

HGL-454 HUMAN ANATOMY AND PHYSIOLOGY-II

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Brief anatomical description of excretory system, Physiology of excretion and urine formation: Glomerular filtration, tubular reabsorption and tubular secretion, Regulation of body fluid by kidneys, Formation of dilute and concentrated urine.

Section-B

Brief anatomical description of male and female reproductive systems, Spermatogenesis and Oogenesis, Female reproductive cycle, Hormonal control of reproduction. Structure and functions of endocrine glands, Mechanisms of hormone action, Control of hormones secretion.

Section-C

Gross anatomy of brain and spinal cord, Ascending and descending tracts in spinal cord, Action potential, Transmission at synapse, Neurotransmitters, Sensory, Motor and integrative functions of brain, Limbic system, Types of reflex actions

Section-D

Comparison of somatic and autonomic nervous system, Brief description of anatomy of autonomic nervous system, Neurotransmitters and receptors of autonomic nervous system, Physiology of autonomic nervous system, Anatomy and physiology of Eye, Ear, Skin and Touch receptors.

Books Recommended:

1. Drake, R., Vogl, W. and Mitchell, A. (2015). Gray's Anatomy for Students. Churchill Livingstone, USA.
2. Guyton, A.C. and Hall, J.E. (2016). Textbook of Medical Physiology. Elsevier Publications, New York.
3. Standring, S. (2015). Gray's Anatomy. Churchill Livingstone, USA. 40th ed.
4. Tortora, G.J. and Henderson, S.R. (2013). Principles of Anatomy and Physiology. Harper Collins College Publishers.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGL-457 HUMAN BIOCHEMICAL GENETICS AND IMMUNOGENETICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Molecular structure, biosynthesis and genetics of the ABH antigens, Haemoglobin variants: Hemoglobin Synthesis, Structure, and Function, Unstable Variants, High Oxygen Affinity Variants, Low Oxygen Affinity Variants, Methemoglobin ("M-Type") Variants, Globin Chain Elongation Mutants, Variants that Affect Multiple Hemoglobin Functions

Section-B

Innate errors of metabolism: Pathophysiology, diagnosis and management of Alkaptonuria, Phenylketonuria, Cystinuria, Albinism, Citrullinemia, Tay-Sachs disease, Gaucher disease, Niemann-Pick disease, Leish-Nyan syndrome, Adenosine deaminase deficiency.

Section-C

Introduction to immune system — Principles of innate and adaptive immunity, Cells of immune system, Humoral and cell-mediated immune responses, Complement system, Inflammation, Overview of antigen and antibody structure, Multigene organization of Ig genes, V-J rearrangements in light chain DNA, V-D-J rearrangement in heavy chain DNA, Enzymatic joining of gene segments and role of RAG-1 and RAG-2 genes.

Section-D

Molecular organization of MHC, Concept of MHC haplotypes, Organization of class-I and class-II molecules; class-I MHC-peptide interaction, Class-II MHC peptide interaction, Polymorphism of class-I and class-II molecules; Class-III molecules, MHC in infectious diseases.

Books Recommended:

1. Harris, H. (1980). The Principles of Human Biochemical Genetics. North-Holland/Elsevier, Boston.
2. Lodish, H., Berk, A., Kaiser, A.C., Krieger, M., Scott, M.P., Bretscher, A., Ploegh, H. and Matsudaira, P. (2016). Molecular Cell Biology. W.H. Freeman and Co., New York, 6th ed.
3. Murphy, K. and Weaver, C. (2017). Janeway's Immunobiology: The Immune System in Health and Disease. Garland Science, Taylor & Francis, New York, USA.
4. Nelson, D.L. and Cox, M.M. (2017). Lehninger's Principles of Biochemistry. W.H. Freeman and Co., New York, 5th ed.
5. Owen, J.A., Punt, J. and Stanford, S.A. Kuby Immunology (2013). W.H. Freeman and Company, New York.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGL-458 RESEARCH DESIGN AND METHODOLOGY

Time: 3 Hours

Credits: 2-1-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Research problem, Objectives and hypothesis, Uses of sources of information such as journals, books, Index Medicus, Excerpta Medica, Biological Abstracts, Science Citation Index.

Section-B

Preparing a manuscript for publication, Editing and galley proof correction of manuscript, Publication ethics, Conflicts of interest, Plagiarism, Protocol content for research project, Preparation and delivering of scientific talk, Guidelines for putting up poster displays.

Section-C

Sampling concepts, Questionnaire, Schedule, Advantage, Limitations and precautions of sampling, Common methods of random sampling, Area sampling, Non-random methods of sampling, Prognostic and concomitant variables, Cohort and longitudinal study, Case-control and nested case-control design, Cross-sectional studies with merits and demerits, Sample size determination, general consideration and power analysis.

Section-D

Statistical significance and 'P'-values, Null and alternative hypothesis, Type-I and Type-II errors, General procedure to obtain 'P'-value, Test of methods through sensitivity, specificity and likelihood ratio, Positive and negative predictivity, Odds ratio for independent and matched samples.

Books Recommended:

1. Dear, P.H. (Ed.) (1997). Genome Mapping. IRL Press / Oxford University Press, U.K.
2. Ford, E.D. (2000). Scientific Methods in Ecological Research. Cambridge University Press, UK.
3. Gurumani, N. (2007). Research Methodology for Biological Sciences. M.J.P. Publishers, India.
4. Hawkins, C. and Sorgi, M. (1985). Research – How to Plan, Speak and Write about it. Springer-Verlag, Berlin.
5. Indrayan, A. (2008). Medical Biostatistics. Chapman and Hall, New York.
6. Kothari, C.R. (2009). Research Methodology: Methods and Techniques. New Age International Publication, New Delhi.
7. Kothari, C.R. and Garg, G. (2014). Research Methodology: Methods and Techniques. New Age International Publication, New Delhi. 3rd. ed.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGP-452 LAB EXERCISES IN HUMAN CYTOGENETICS

Max. Marks : 100

Credits: 0-0-1.5

Demonstration of short-term blood lymphocyte culture — Washing and sterilization of glassware and plasticware, Preparation of solutions and culture medium, Harvesting the culture, Staining, Banding and Scoring of prepared slides, Demonstration of photomicrography, Developing and printing of photographs, Karyotyping of solid stained and G-banded chromosome preparations, Identification of structural and numerical chromosomal aberrations from photographs, Numericals on chromosome nomenclature.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGP-454 HUMAN PHYSIOLOGY AND IMMUNOLOGY
(PRACTICAL)

Max. Marks : 100
Credits: 0-0-1.5

Estimation of Bleeding time, clotting time, Haemoglobin concentration,

Spirometry,

Osmotic fragility of RBC,

Heart rate, pulse rate and Blood pressure estimation

Immunological Techniques including Agglutination, Double Immunodiffusion,

Immunoelectrophoresis, Enzyme linked Immunosorbant assay

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGL-462 LAB DESIGN AND BIOSAFETY GUIDELINES
(Elective Paper)

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Risk classification of laboratory, Design and safety guidelines for biosafety level, I, II, III, IV laboratories, Laboratory animal facility.

Section-B

Safe laboratory techniques for transport, storage, handling of human blood, sputum, urine, tissue samples, Handling and storage of hazardous chemicals, Chemical incompatibility, Health hazards of chemicals, Chemical disinfectants.

Section-C

Equipment-related hazards, Safety rules for support staff and training programmes, Emergency procedures for fire and chemical exposure, Safety checklist.

Section-D

Use and care of pipettes, Pipetting aids, Centrifuges, Ovens, Incubators, Sterilizers, Biological safety cabinets, Thermal Cyclers, Microscopes, Electrophoresis apparatus, Incinerators,

Books Recommended

1. WHO (World Health Organization) (1993). Laboratory Biosafety Manual. AITBS Publishers, Delhi, 2nd ed.
2. WHO (World Health Organization) (2003). Laboratory Biosafety Manual. WHO, Geneva.
3. Wilson, K. and Walker, J. (2010). Principles and Techniques of Biochemistry and Molecular Biology. Cambridge University Press, London, 7th ed.

M.Sc. HUMAN GENETICS (SEMESTER-II)
(Credit Based Evaluation & Grading System)

HGL-463 TISSUE CULTURE AND MICROSCOPY
(Elective Paper)

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Basic laboratory procedures for tissue culture, Overview of cell culture and harvest, peripheral blood samples, bone marrow aspirates, amniotic fluid specimens, solid tissue samples, culture maintenance and growth interval.

Section-B

Banding, Cell viability assays, Mitotic index. High resolution studies, Cell synchronization techniques, Culture failure, Preservation of cells, Chromosomal analysis,

Section-C

Fundamentals of microscopy, Bright field microscopy, Video microscopy and image processing.

Section-D

Fluorescence microscopy and related techniques, Confocal microscopy, Electron microscopy, Theoretical and practical aspects of SEM and TEM, Specimen preparation for electron microscopy.

Books Recommended:

1. Gersen, S.L. and Keagle, M.B. (2005). The Principles of Clinical Cytogenetics. Humana Press, 2nded.
2. Karp, G. (2014). Cell Biology. Wiley, 7thed.
3. Rooney, D.E. and Czepulkowski, B.H. (1992). Human Cytogenetics: A Practical Approach. IRL Press, Vol. 1&2, 2nded.
4. Stoddart, M.J. (2011). Mammalian Cell Viability: Methods and Protocols. Humana Press, New York.

M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Continuous Evaluation Grading System)

HGL-502 FUNCTIONAL GENOMICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Principles and strategies in identifying disease gene, Mapping and cloning of human disease genes, Split genes and overlapping genes, Multigene families and repetitive DNA, Direct and indirect DNA testing and its applications.

Section-B

Genetic approaches in treating human diseases, Principles and applications of gene therapy, Gene transfer Methods, Gene therapy for cystic fibrosis, Duchenne muscular dystrophy, bleeding disorders and severe combined immunodeficiency syndrome.

Section-C

Structure and function of microRNAs, snRNA, RNA interference, Role of microRNAs in human disease diagnostics, Epigenetics, Next generation sequencing and its applications, DNA profiling and its applications.

Section-D

Evolution of mitochondrial genome, eukaryotic nuclear genome, human sex chromosome, human DNA sequence families and DNA organization, gene structure and modern humans.

Books Recommended:

1. Brown, T.A. (2005). Genetics: A Molecular Approach. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
2. Brown, T.A. (2006). Genomes 3. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
3. Brown, T.A. (2010). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 6th ed.
4. Brown, T.A. (2016). Gene Cloning and DNA Analysis: An Introduction. Blackwell Publishing Co., Oxford, 7th ed.
5. Strachan, T. and Read, A. (1999). Human Molecular Genetics. Garland Publishers, London, 2nd ed.
6. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
7. Sudbery, P. and Sudbery I (2009). Human Molecular Genetics. Pearson Education, UK, 3rd ed.
8. Primrose, S.B. and Twyman, R.M. (2003). Principles of Genome Analysis and Genomics. Blackwell Publications, London.
9. Watson, J.D., Myers, R.M., Caudy, A.A. and Witkowski, J.A. (2007). Recombinant DNA Genes and Genomes – A Short Course. Cold Spring Harbor Laboratory Press, 3rd ed.

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

HGL-505 HUMAN EMBRYOLOGY AND DEVELOPMENTAL GENETICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Fertilization and molecular events during fertilization, and prenatal development of human embryo up to three germinal layers. differential gene activity and cell differentiation, Gastrulation, cleavage patterns, fate map during gastrulation,

Section-B

Implantation, Formation and types of placenta, Notochord formation, Neurulation, development of nervous system. Genetic and molecular control of development of head and neck region.

Section-C

Formation of somites, Structure of somites, Formation of blood vessels, and development of cardiovascular system, Genetic and molecular control of development of limbs.

Section-D

Development of gastrointestinal system, development of urogenital system and Genetics of sex determination in humans.

Books Recommended

1. Gilbert, S.F. (2016). Developmental Biology. Sinauer Association, Inc., Sunderland, MA. 11th ed.
2. Mueller, R.F. and Young, I.D. (2010). Emery's Elements of Medical Genetics. Churchill Livingstone, New York, 11th ed.
3. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2007). Thompson and Thompson Genetics in Medicines. W.B. Saunders and Co., New York, 7thed.
4. Sadler, T.W. (2014). Langman's Medical Embryology. Williams and Wilkins, Philadelphia, USA, 13th ed.
5. Snustad, P. and Simmons, M.J. (2010) Principle of Genetics. John Wiley and Sons, Inc., N.J., USA.
6. Singh, I. and Paul, G.P. (2014). Human Embryology. Jaypee Brothers Medical Publishers (P) Ltd., New Delhi.

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

HGL- 508 ESSENTIALS OF MEDICAL GENETICS

Time: 3 Hours

Credits: 2-1-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section A

Historical development of Medical Genetics. Impact of genetics in medicine, Medical relevance of meiosis and mitosis, General concepts on enzyme deficiencies and diseases

Section B

Spectrum of genetic diseases (single gene, chromosomal, multifactorial , mitochondrial, somatic cell genetic diseases) and patterns of their inheritance.

Section C

Atypical inheritance. Dominance and recessiveness, Clinical consequences of mutations.

Section D

Pharmacogenetic diseases, Disorders of sexual development, Chromosomal instability syndromes, Chromosomal microdeletion syndromes, Prion diseases.

Books Recommended:

1. Gardner, A. and Davies, T. (2017). Human Genetics. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited, 2nd ed.
2. Gibson, G. (2015). A primer of Human Genetics. Sinauer.
3. Korf, B.R. and Irons, H.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
4. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2015). Thompson & Thompson's Genetics in Medicine. Saunders, 8th ed.
5. Rimoin, D.L., Pyeritz and Korf, P.R. (2013). Emery and Rimoin's Principles and Practice of Medical Genetics, Academic Press, New York, 6thed.
6. Skirton, H. and Patch, C. (2017). Genetics for the Health Sciences. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited.
7. Speicher, M. Antonarakis, S.E. and Motulsky, A.G. (2010) Vogel and Motulsky's Human Genetics: Problems and Approaches. Springer-Verlag, 4th ed.
8. Strachan, T. Goodship, J. and Chinnery P. (2015). Genetics and Genomics in Medicine. Garland Publishers, New York.
9. Turnpenny, P.D. and Ellard, S. (2015). Emery's Elements of Medical Genetics. Elsevier, 15th ed.

M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)

HGL-510 HUMAN POPULATION GENETICS AND VARIATION

Time: 3 Hours

Credits: 3-1-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Historical emergence and application of Human population Genetics. Kinetics of changes of gene and genotype frequencies. Dynamics of Hardy-Weinberg law and its relation with for autosomal locus for two and multiple alleles, Testing of Hardy-Weinberg equilibrium in the population.

Section-B

Impact of recurrent and non-recurrent mutation in the Hardy-Weinberg equilibrium population. Mutation pressure and estimates of mutation rates. Selection coefficient and fitness. Selection against recessive/ dominant/partial dominant/overdominant genes. Heterozygote advantage. Equilibrium between and selection.

Section-C

Consanguinity and inbreeding. Inbreeding coefficient of a population and individual through path analysis. Biological consequences of inbreeding with genetic load and its measurements. Effect of migration and genetic drift on gene frequencies.

Section-D

Classification of races through UNESCO guidelines. Admixture of the races. Genetic imprint of admixture and its biological impact. Population stratification. Ethnic elements in Indian population. Objectives of racial classification.

Recommended Books :

1. Balding, D.J., Bishop, M. and Cannings, C.C. (Eds.) (2007). Handbook of Statistical Genetics. John Wiley and Sons, England, Vol. 1&2, 3rd ed.
2. Falconer, F.S. and MacKay, T.F.C. (1996). Introduction to Quantitative Genetics. ELBS/Longman, England, 4th ed.
3. Hamilton, M.B. (2009). Population Genetics. Wiley-Blackwell, USA.
4. Hartl, D.L. and Clark, A.G. (2007). Principles of Population Genetics. Sinaur Associates, Inc., Massachusetts, 4th ed.
5. Hedrick, P.W. (2011). Genetics of Population. Jones and Bartlett Publishers, Massachusetts, 4th ed.
6. Neale, B., Ferreira, M.A.R., Medland, S.E. and Posthuma, D. (Eds.) (2008). Statistical Genetics: Gene Mapping through Linkage and Association. Taylor and Francis Group, USA

M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)

HGS-506 SEMINAR

Max. Marks : 100

Credits: 0-0-1

The paper is based on the seminars delivered by the students on current topics related to various disciplines of Human Genetics.

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

HGP-508 EXERCISES IN MEDICAL GENETICS

Max. Marks : 100

Credits: 0-0-1.5

Numericals, Pedigree analysis and case studies on medical genetics, Common chromosomal conditions, Karyotyping from abnormal chromosomal preparations, Genetic databases (OMIM, London Dysmorphology, Possum) and Online Medical Genetic Support Groups for patients (Genetic Alliance, Family village).

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

**HGP-502 FUNCTIONAL GENOMICS
(PRACTICAL)**

Max. Marks : 100

Credits: 0-0-2

Preparation of different stock solutions for DNA extraction and electrophoresis, Preparation of working solutions from stock solutions, Extraction of DNA from human blood by organic method, DNA quantification, Amplification of genomic DNA by PCR, PCR-RFLP, PCR-SSCP analysis, Demonstration of RT-PCR, Biological database searches (NCBI, ENSEMBL, UCSC, GDB, PDB, HGMD).

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

**HGL-511 IMMUNOGENETICS IN HEALTH AND DISEASE
(Elective Paper)**

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Evaluation of antigen-antibody interactions: Immunoprecipitation Immuno-electrophoresis agglutination assays, western blotting, ELISA, RIA, Immunoelectron microscopy, Flow cytometry. Evaluation of lymphocyte function. Immunolocalization of antigens.

Section-B

Experimental systems in immunology: Cell lines, Knock out mice, Knock in mice, SCID mice, SCID Human mice, Inbred strains, Nude mice. Monoclonal, Humanized and Genetically engineered antibodies.

Section-C

Autograft, isograft, allograft, xenograft, Immunological basis of graft rejection, Clinical characteristics of allograft rejection, HLA typing, Circumvention of alloimmune response, Maternal-fetal incompatibility, Role of MHC in resistance and susceptibility to diseases.

Section-D

Tumor antigens, Limitations of effectiveness of immune response against tumors, Immunodiagnosis of tumor antigens, Types and properties of immunotolerance, T-cell tolerance to self-antigens, B-cell tolerance to self-antigens, Central tolerance, Peripheral tolerance, acquired allogeneic tolerance.

Books Recommended:

1. Owen, J.A., Punt, J. and Stanford, S.A. Kuby Immunology (2013). W.H. Freeman and Company, New York.
2. Roitt, I.M., Brostoff, J. and Male, D.K. (2012). Immunology. Mosby Inc, UK.
3. Murphy, K. and Weaver, C. (2017). Janeway's Immunobiology: The Immune System in Health and Disease. Garland Science, Taylor & Francis, New York, USA.
4. Parham, P. (2009). The Immune System. Taylor and Francis Group, New York.

**M.Sc. HUMAN GENETICS (SEMESTER-III)
(Credit Based Evaluation & Grading System)**

**HGL-514 GENETICS OF HUMAN PATHOGENS AND INFECTIOUS DISEASES
(Elective Paper)**

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Brief history of infectious diseases, Infectious disease epidemiology, Epidemiological triad, Rothman causal pies, Chain of infection, Natural history of infectious diseases, Vector borne diseases, Emerging and Re-emerging infectious diseases, Herd immunity, Quarantine.

Section-B

Role of horizontal gene transfer in pathogen evolution, Transformation, Conjugation and Transduction, Virulence factors for evasion of host defences, Toxins (cholera, diphtheria, tetanus, botulinum, anthrax), Superantigens, Resistance to antimicrobial peptides (AMPs), AMP efflux pumps; LanFEG and LanI proteins, Iron acquisition mechanisms, Pili and fimbriae as adherence factors, Biofilms.

Section-C

Virulence regulation; phase and antigenic variation with examples in *Salmonella* and *Neisseria*, Two component regulatory system, Quorum sensing; Lux system of *V. cholera* and Agr system of *S. Aureus*, Pathogenicity islands, Antimicrobial compounds, Mechanisms of antibiotic resistance, MRSA strains.

Section-D

Pathogenesis of opportunistic infections; *S. aureus*, *E.coli*, *P. aeruginosa*, Pathogenesis of intracellular pathogens; Tuberculosis, HIV-AIDS, Leishmaniasis, Brief introduction to emerging viral diseases, Zika, Dengue, Chikungunya, Ebola and Marburg.

Books Recommended:

1. Alberts B, Johnson A, Lewis J (2002). Molecular Biology of the Cell, New York, Garland Science, 4th ed.
2. Brenda A. Wilson, Abigail A. Salyers, Dixie D. Whitt, Malcolm E. Winkler (2011). Bacterial Pathogenesis: A molecular approach, 3rd ed. ASM Press, Washington, DC.
3. Ray M. Merrill (2010). Introduction to Epidemiology. 5th ed, John & Bartlett Publishers.
4. Richard A. Kaslow, Janet McNicholl, Adrian V. S. Hill (2008). Genetic susceptibility to Infectious Diseases, Oxford University Press, USA.
5. Joanne Willey, Linda Sherwood, Chris Woolverton (2007). Prescott's Microbiology, McGraw-Hill Higher Education, 7th ed.
6. Tjeerd G. Kimman (2001). Genetics of Infectious Disease Susceptibility, Springer Science & Business Media.

M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Continuous Evaluation Grading System)

HGL- 551 PERSPECTIVES OF HUMAN GENOME AND BIOETHICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section A

The Human Genome Project, origins, goals and organization. Ethical, legal and social implications of the HGP, Applications of the HGP.

Section B

Overview of The Human Genome Diversity Project, Environmental Human Genome Project, Cancer Genome Anatomy Project, The SNP project, The Phenome project, The Metabolome Project.

Section C

Bioethics –Definition, historical aspects, types and scope, Rights’-based ethics theories, Duty-based ethics theories and utilitarian ethics, Basic aspects of Principles of autonomy, non-maleficence, beneficence and justice; Informed consent and confidentiality, Environmental ethics.

Section D

Ethical dilemmas: Contextual vignettes in medical genetic advances – Genetic screening, Gene Editing, Genome ownership, Genetic discrimination, Genetic insurance, Genetic privacy.

Books Recommended:

1. Beauchamp T.L. and Childress, J.F. (1994). Principles of Biomedical Ethics. Oxford Univ. Press, New York.
2. Gibson, G. and Muse, S.U. (2009). A Primer of Genome Science. Sinauer.3rd ed.
3. Jecker, N.S., Jonsen, A.R. and Pearlman, R.A. (2010). Bioethics: An Introduction to History, Methods and practice. Jones and Bartlett, New Delhi 2nd ed.
4. Kumar, D. (2015). Genomics and Society: Ethical, Legal, cultural and socioeconomic implications. Academic Press, 1st ed.
5. Macer, D.R.J. (1998). Bioethics is Love of Life: Alternative Textbook. Eubios Ethics Institute Publications, Japan.
6. Yashon, R.K. and Cummings, M.R. (2012). Human Genetics and Society. Oxford University Press.

Online Links

<http://www.nhgri.nih.gov/>

<http://www.hgmp.mrc.ac.uk./GenomeWeb/>

<http://www.ncbi.nlm.nih.gov/genemap99/>

<http://www.ncbi.nlm.nih.gov/CGAP>

<http://www.ncbi.nlm.nih.gov/HUGO>

<http://www.ebi.ac.uk/~sterk/genome-MOT/>

<http://www.nuffieldbioethics.org>

**M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)**

HGL- 554 STRUCTURAL BIOLOGY AND PHARMACOGENOMICS

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Hierarchical proteins structure, amino acid properties; optical, acid base and absorption, Secondary structures, Peptide bond, Ramachandran plots, Structure of polysaccharides (Cellulose, Starch, Glycogen, peptidoglycan), Lipids (Phospholipids, Sphingolipids) Common motifs, Helix turn Helix, Greek Key, Tertiary structures, domains, Domains in DNA-binding proteins.

Section-B

Structure of Membrane proteins, Tertiary structures of DNA, ribose ring conformations and puckering, Hoogsteen base pairing, Triple helices, DNA topology, Supercoiling, Prediction of secondary structure from primary structure, Chou-Fasman method.

Section-C

Protein folding theories, Levinthal paradox, Interplay of non-covalent interactions in higher order structures, Methods for protein purification (affinity and size exclusion chromatography), MADI-TOF, protein-protein interactions; phage display and Yeast two hybrid display.

Section-D

Historical drug discovery (Paul Ehrlich magic bullet, chemoreceptor theory), Modern drug designing, Pharmacokinetics and pharmacodynamics, ADME properties, Dose response curves, LD, ED concepts, Clinical trials, Pharmacogenomics, CYP2D6 Pharmacogenetics, Application of pharmacogenomics in cancer treatment, Cardiovascular disorders and Diabetes.

Books recommended:

1. Williamson, M. (2012). How Proteins Work. Garland Science, Taylor & Francis Group USA.
2. Lovric, J. (2011). Introducing Proteomics. Wiley- Blackwell, USA.
3. Creighton, T.E. (2010). The Physical and chemical Basis of Molecular Biology. Helvetian Press Edition.

**M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)**

HGL- 557 CLINICAL GENETICS

Time: 3 Hours

Credits: 2-1-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section A

Indications for and types of invasive and non-invasive prenatal diagnostic techniques. Amniocentesis, Chorionic villus sampling, Ultrasonography, Fetoscopy, Maternal serum screening

Section B

Pre-conceptional and Preimplantation genetic diagnosis, Teratogen exposure in early pregnancy. Importance of Gene and stem cell therapy for genetic conditions.

Section C

Genetic screening and genetic testing, Newborn screening, population carrier screening, Presymptomatic and predispositional testing.

Section D

Genetic counselling: definition, process and diagnostic problems. Role of genetic counsellor and allied health professionals in the Genetic Care Pathway.

Books Recommended:

1. Gardner, A. and Davies, T. (2017). Human Genetics. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited, 2nd ed.
2. Gibson, G. (2015). A Primer of Human Genetics. Sinauer.
3. Korf, B.R. and Irons, H.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
4. MacFarlane, I. M., Peach, V.M. and Leroy, B.S. (2014). Genetic Counseling Research: A Practical Guide. Oxford University Press, 1st ed.
5. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2015). Thompson & Thompson's Genetics in Medicine. Saunders, 8th ed.
6. Read, A. and Donnai, D. (2015). New Clinical Genetics. Scion, UK, 3rd edn.
7. Rimoin, D.L., Pyeritz and Korf, P.R. (2013). Emery and Rimoin's Principles and Practice of Medical Genetics, Academic Press, New York, 6th ed.
8. Skirton, H. and Patch, C. (2017). Genetics for the Health Sciences. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited.
9. Speicher, M. Antonarakis, S.E. and Motulsky, A.G. (2010) Vogel and Motulsky's Human Genetics: Problems and Approaches. Springer-Verlag, 4th ed.
10. Strachan, T. Goodship, J. and Chinnery P. (2015). Genetics and Genomics in Medicine. Garland Publishers, New York.
11. Turnpenny, P.D. and Ellard, S. (2015). Emery's Elements of Medical Genetics. Elsevier, 15th ed.

M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)

HGP- 557 EXERCISES IN CLINICAL GENETICS

Max. Marks : 100

Credits: 0-0-1.5

Case studies and pedigree construction in clinical genetics. Proforma designing for some genetic conditions, Importance of family history, Practical aspects in the case management of some genetic diseases, Types and importance of maintaining Genetic Registers.

M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)

HGL-561 REGULATION IN HUMAN GENETICS RESEARCH
(Elective Paper)

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section A

Historical overview for regulatory approaches for research in Human Genetics and Protection of Human participants (Eugenics, Experiments during and after WW-II, The Tuskegee Trial, Baby Doe Case), The Hippocratic Oath, The Nuremberg Code, Declarations of Geneva and Helsinki.

Section B

Overview of Reprogenetics, Consumer Genetics, Personal Genomics, Public health Genetics, Regenerative medicine, Organ Transplantation, Euthanasia, Intellectual Property Rights, **International** Guidelines and regulations for conducting Human Genetics research- Universal Declaration on the Human Genome and Human Rights, International Ethical Guidelines for Biomedical Research Involving Human Subjects(CIOMS-WHO).

Section C

The Indian Scenario- Genetically modified organisms, Human Genetic Modification, Reproductive Genetic Testing, Genetic Engineering Approval Committee (GEAC).

Section D

Drugs and Cosmetics Act, The Patents Act, The Medical Termination of Pregnancy Act, The Preconception and Prenatal Diagnostic Techniques Act, The Transplantation of Human Organs Act.

Recommended Readings:

1. Drugs and Cosmetics Act, (1940, amended 2003), <http://indianmedicine.nic.in/html/pharma/adrugsnote.pdf>
2. DBT and ICMR: Guidelines for Stem Cell Research and Therapy (2007): http://icmr.nic.in/stem_cell/Stem_cell_guidelines.pdf
3. Ethical Guidelines for Biomedical Research on Human Participants (2006): http://icmr.nic.in/ethical_guidelines.pdf
4. Universal Declaration on the Human Genome and Human Rights <http://www.unesco.org/new/en/social-and-human-sciences/themes/bioethics/human-genome-and-human-rights/>
5. World Medical Association: Declaration of Helsinki (Ethical Principles for Medical Research Involving Human Subjects)
6. WIPO. World Intellectual Property Organization (www.wipo.int).

**M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)**

**HGL-562 CONCEPTS OF BIOLOGICAL DEMOGRAPHY
(Elective Paper)**

Time: 3 Hours

Credits: 3-0-0

Max. Marks: 100

Mid Semester Marks : 20

End Semester Marks : 80

Mid Semester Examination: 20% weightage

End Semester Examination: 80% weightage

Instructions for the Paper Setters:

Eight questions of equal marks (Specified in the syllabus) are to be set, two in each of the four Sections (A-D). Questions may be subdivided into parts (not exceeding four). Candidates are required to attempt five questions, selecting at least one question from each Section. The fifth question may be attempted from any Section.

Section-A

Some basic concepts of demography — Family, Marriage, Reproductive span, Fertility, Fecundity, Sterility, Morbidity, Mortality, Stillbirth, Live-birth, Foetal death, Abortion, Child death, Birth order, Parity, Cohort, Growth rate, and Migration.

Section-B

Sources of demographic data, Factors affecting fertility, mortality, migration, sex ratio, Population structure, Density of population, Determinants of population growth.

Section-C

Family planning programme in India, Urbanization, Factors responsible for urban-rural population distribution

Section-D

Principles of epidemiology. Geographical distribution of genetic diseases. Distribution of complex genetic diseases. The human genetic diseases heritage.

Books Recommended:

1. Hinde, A. (1998). Demographic Methods. Oxford University Press, Inc., New York.
2. Krishna Reddy, M.M. (1996). An Introduction to Demographic Behaviour in India. Kanishka Publishers, Distributors, New Delhi.
3. Krishnan, P., Tuan, C.-H. and Mahadevan, K. (1992). Readings in Population Research. B.R. Publishing Corp., Delhi.
4. Majumdar, P.K. (2010). Fundamentals of Demography. Rawant Publications, Jaipur.
5. Raj, H. (2004). Fundamentals of Demography. Surjeet Publications, Delhi.
6. Shrivastava, O.S. (1994). Demography and Population Studies. Vikas Publishing House Pvt. Ltd., New Delhi.
7. Srinivasan, K. (1998). Basic Demographic Techniques and Applications. Sage Publications, New Delhi.

**M.Sc. HUMAN GENETICS (SEMESTER-IV)
(Credit Based Evaluation & Grading System)**

HGP-563 : RESEARCH TRAINING REPORT

(Course Category : Discipline Specific Elective Course)

Credits : 0-0-3

Marks : 100

The topic of the Research Training Report and the name of the supervisor are to be finalized during the end of Semester-II. Experimental work on project will begin in Semester-III. The dissertation is to be submitted before theory examinations of Semester-IV.