FACULTY OF LIFE SCIENCES

SYLLABUS

for

M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)
(UNDER THE SCHEME OF HONOURS SCHOOL)
FOR NEW ADMISSION W.E.F. 2019-20
(SEMESTER: I - II)

M.Sc. (HS) HUMAN GENETICS
(Credit Based Evaluation & Grading System)
FOR OLD STUDENTS
(SEMESTER: III - IV)
Examinations: 2019-20

GURU NANAK DEV UNIVERSITY
AMRITSAR

M.Sc. HUMAN GENETICS
(Credit Based Evaluation & Grading System)
(UNDER THE SCHEME OF HONOURS SCHOOL)

Credit Structure

Semester	Credits			
	Core	Elective	Interdisciplinary/ Optional	Total
I	17	3	-	20
II	22	3	-	25
Total Credits	**39**	**6**	**-**	**45**
III	14	3	8	25
IV	16	-	4	20
Total Credits	**30**	**3**	**12**	**45**

SCHEME OF COURSE

Semester-I

Course No.	C/E/ I	Course Title	L	T	P	Total Credits	Total Marks		
Core Courses (20 Credits)							**Max.**	**Mid Sem**	**End Sem**
HGL-422	**C**	**Medical Genetics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-423	**C**	**Methods of Research Design**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-425	**C**	**Perspectives of Human Genome**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-426	**C**	**Human Developmental Genetics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGP-422	**C**	**Medical Genetics Practical**	**0**	**0**	**2**	**2**	**100**	**--**	**--**
Elective Course (3 Credits)									
HGP-432	**E**	**Lab Rotations in Molecular Genetics and Biochemistry**	**0**	**0**	**3**	**3**	**100**	**--**	**--**
HGP-433	**E**	**Lab Rotations in Quantitative Genetics and Bioinformatics**	**0**	**0**	**3**	**3**	**100**	**--**	**--**

NOTE : (i) The students are required to opt one ‘Elective’ paper.

M.Sc. HUMAN GENETICS

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Semester-II

Course No.	C/E/I	Course Title	L	T	P	Total Credits	Total Marks		
Core Courses (25 Credits)							**Max.**	**Mid Sem**	**End Sem**
HGL-471	**C**	**Human Molecular Genetics and Functional Genomics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-472	**C**	**Clinical Genetics and Genetic Counseling**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-473	**C**	**Bioethics and Human Genetics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-474	**C**	**Recent Concepts in Human Genetics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGS-475	**C**	**Seminar**	**0**	**0**	**1**	**1**	**100**	**20**	**80**
HGP-471	**C**	**Human Molecular Genetics and Functional Genomics Practical**	**0**	**0**	**3**	**3**	**100**	**--**	**--**
HGP-472	**C**	**Clinical Genetics and Genetic Counseling Practical**	**0**	**0**	**2**	**2**	**100**	**--**	**--**
Elective Course (3 Credits)									
HGL-481	**E**	**Genetics of Human Cancer**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-482	**E**	**Tools and Techniques in Immunology**	**3**	**0**	**0**	**3**	**100**	**20**	**80**

1. The students are required to opt one ‘Elective’ paper.

NOTE: PSL-053 ID Course Human Rights & Constitutional Duties (Compulsory Paper). Students can opt. this paper in any semester except the 1st Semester. This ID Paper is one of the total ID Papers of this course.

M.Sc. (HS) HUMAN GENETICS
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Semester-III

Course No.	C/E/ I	Course Title	L	T	P	Total Credits	Total Marks		
Core Courses (25 Credits)							**Max.**	**Mid Sem**	**End Sem**
HGL-521	**C**	**Current Trends in Medical Genetics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-523	**C**	**Genetics in Post-Genomic Era**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGP-581	**C**	**Advanced Practicals**	**0**	**0**	**6**	**6**	**100**	**--**	**--**
Elective Course (3 Credits)									
HGL-532	**E**	**Human Genetics Research and Regulation**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-533	**E**	**Advanced Course in Genetic Engineering**	**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**
	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 two paper (Interdisciplinary/Optional) of 4 credits each of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.

M.Sc. (HS) HUMAN GENETICS
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Semester-IV

Course No.	C/E/ I	Course Title	L	T	P	Total Credits	Total Marks		
Core Courses (20 Credits)							**Max.**	**Mid Sem**	**End Sem**
HGL-524	**C**	**Advanced Cytogenetics**	**3**	**1**	**0**	**4**	**100**	**20**	**80**
HGL-552	**C**	**Applied Human Molecular Genetics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-555	**C**	**Birth Defects and Dysmorphology**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGL-556	**C**	**Structural Bioinformatics and Pharmacogenomics**	**3**	**0**	**0**	**3**	**100**	**20**	**80**
HGP-552	**C**	**Applied Human Molecular Genetics Practical**	**0**	**0**	**3**	**3**	**100**	**--**	**--**
HGD-595	**–**	**Dissertation* (To be graded as satisfactory/unsatisfactory)**	**0**	**0**	**0**	**0**			
Interdisciplinary/Optional Course (Two)									
–	**I**	**To be taken from outside the department**	**4**	**0**	**0**	**4**	**100**	**20**	**80**

1. The students are required to take one papers (Interdisciplinary/Optional) of 4 credits of their choice from any other department of Guru Nanak Dev University Campus, Amritsar.
2. *Topic of dissertation and name of supervisor to be finalized during Semester-II

M.Sc. HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-I)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-422 MEDICAL GENETICS

Credits: 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.

Section A

Growth and development of medical genetics (1956-to-present), Role of genetics in medicine, Types of genetic disorders. Genetic disorders with classical Mendelian inheritance. Medical relevance of meiosis and mitosis, Consequences of consanguineous marriages.

Section B

Concepts of genetic heterogeneity, reduced penetrance, variable expressivity, Pseudoautosomal inheritance, Genomic imprinting, Mosaicism, Uniparental disomy, Anticipation and Pleiotropy with suitable examples.

Section C

Dominance and recessiveness, Concept of phenotype and relationship between genotype and phenotype in genetic disease. Mitochondrial diseases, Multifactorial Disorders, Chromosomal disorders, Sex chromosomal and differentiation anomalies.

Section D

Pharmacogenetic diseases (Hereditary disorders with altered drug response, Malignant hyperthermia, G-6-PD deficiency), 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, 6th ed.
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. Antonnarakis, S.E. and Motulsky, A.G. (2010) Vogel and Mothulsku'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

(Credit Based Evaluation & Grading System) (Semester-I)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-423: METHODS OF RESEARCH DESIGN

Credits: 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.

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, Preparing a manuscript for publication, Editing and galley proof correction of manuscript, Conflicts of interest, Publication ethics, Plagiarism, Protocol content for research project, Scientific presentation. Punctuation and abbreviations in foot notes, precaution for writing research report, outlines of technical and popular report.

Section-B

Common methods of Sampling: simple random, stratified, multistage, cluster and systematic random sampling. Non-random methods of sampling: convenience sample, snowball sampling. Questionnaire, Schedule, Advantage, Limitations and precautions 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. Statistical significance and 'P'-values. Null and alternative hypothesis, Type-I and Type-II errors.

Section-C

Risk classification of laboratory, Design and safety guidelines for biosafety level, I, II, III, IV laboratories, Disinfection and sterilization, Laboratory animal facility. Handling and storage of hazardous chemicals, Chemical incompatibility, Safe work practices and procedures.

Section-D

Health hazards of chemicals. Safe laboratory techniques for transport, storage, handling of human blood, sputum, urine, tissue samples and infectious materials, Safety equipments, Emergency procedures for fire and chemical exposure, Safety checklist

Books Recommended:

1. Ford, E.D. (2000). Scientific Methods in Ecological Research. Cambridge University Press, UK.
2. Gurumani, N. (2011). Research Methodology for Biological Sciences. M.J.P. Publishers, India.
3. Hawkins, C. and Sorgi, M. (1985). Research - How to Plan, Speak and Write about it. Springer-Verlag, Berlin.
4. Indrayan, A. (2008). Medical Biostatistics. Chapman and Hall, New York.
5. Kothari, C.R. (2009). Research Methodology: Methods and Techniques. New Age International Publication, New Delhi. 2nd ed.
6. Kothari, C.R. and Garg, G. (2014). Research Methodology: Methods and Techniques. New Age International Publication, New Delhi. 3rd. ed.
7. WHO (World Health Organization) (1993). Laboratory Biosafety Manual. AITBS Publishers, Delhi, 2nd ed.
8. WHO (World Health Organization) (2003). Laboratory Biosafety Manual. WHO, Geneva.

M.Sc. HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-I)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-425 PERSPECTIVES OF HUMAN GENOME

Credits: 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.

Section A

The Human Genome Project (HGP) – historical perspective, goals and controversies, Human Genome Project organization (HUGO), Overview of mapping strategies used in Human Genome Project, Applications and ELSI of HGP.

Section B

Basic concepts and goals of The Human Genome Diversity Project, The Environmental Human Genome Project, The Cancer Genome Anatomy Project, The SNP/HAPMap Project

Section C

Overviews of the Phenome project, toxicogenomics, transcriptomics, metabolomics/metabonomics, Human Metabolome Project.

Section D

The Human connection and model organisms in genome projects: *Escherichia coli, Saccharomyces cerevisiae, Caenorhabditis elegans, Drosophila melanogaster,* Mouse, Rat, Pufferfish, Zebrafish, Monkey, Chimpanzee.

Books Recommended:

1.Gibson, G. and Muse, S.U. (2009). A Primer of Genome Science. Sinauer.3rd ed.
2.Quackenbush, J. (2011). The Human Genome: The Book of Essential Knowledge. Imagine Publishing, New York.

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

(Credit Based Evaluation & Grading System) (Semester-I)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-426 HUMAN DEVELOPMENTAL GENETICS

Credits: 3-0-0
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.

Section-A

Molecular events during gametogenesis and fertilization in Human, Formation of placenta.Prenatal development of human embryo up to three germinal layers: Cleavage patterns, fate map during gastrulation differential gene activity and cell differentiation, Gastrulation, Somites.

Section-B

Molecular events during formation of Notochord, Neural tube, Neural crest cell. Development of spinal cord, brain, Peripheral nervous system of Human.

Section-C

Development of cardiovascular system,Genetic control of development of head and neck region, development of Limbs in human.

Section-D

Genetic and molecular control of development of gastrointestinal system and urogenital system, Genetics of sex determination in humans.

Books Recommended:

1. Gilbert, S.F. (2016). Developmental Biology. Sinnauer Association, Inc., Sunderland, MA. ,11th Ed.
2. Sadler, T.W. (2014). Langman's Medical Embryology. Lippincot Williams and Wilkins, Philadelphia, USA, 13th ed.
3. Singh, I. and Paul, G.P. (2014). Human Embryology. Jaypee Brothers Medical Publishers(P) Ltd., New Delhi, 10th ed.
4. Mueller, R.F. and Young, I.D. (2010). Emery's Elements of Medical Genetics. Churchill Livingstone, New York, 11th ed.
5. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2007). Thompson and Thompson Genetics in Medicines. W.B. Saunders and Co., New York, 7th ed.
6. Snustad, P. and Simmons, M.J. (2015) Principle of Genetics. John Wiley and Sons, Inc., N.J., USA,7th ed.

M.Sc. HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-I)

(UNDER THE SCHEME OF HONOURS SCHOOL)

Credits : 0-0-2

Max. Marks : 100

HGP-422 MEDICAL GENETICS PRACTICAL

Human Genetic variation, Pedigree interpretation for modes of inheritance, Numericals and case studies on medical genetics, Identification features of common chromosomal conditions, Karyotyping from abnormal chromosomal preparations, Genetic databases – OMIM, London dysmorphology database, Possum, Repertox, Human cytogenetics database, Online medical genetics resources – OMIM, Gene clinics, Gene tests, ESHG; Sites for patients – Genetic alliance, Family village.

M.Sc. HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-I)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGP-432: LAB ROTATIONS IN MOLECULAR GENETICS AND BIOCHEMISTRY
(Elective Paper)

Credits: 0-0-3

Max. Marks : 100

The students will visit various research labs in the department for varying time span and will be examined for different practical techniques that they have learnt during the semester.

M.Sc. HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-I)

(UNDER THE SCHEME OF HONOURS SCHOOL)

HGP-433: LAB ROTATIONS IN QUANTITATIVE GENETICS AND BIOINFORMATICS

(Elective Paper)

Credits: 0-0-3

Max. Marks : 100

The students will visit various research labs in the department for varying time span and will be examined for different practical techniques that they have learnt during the semester.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-471: HUMAN MOLECULAR GENETICS AND FUNCTIONAL GENOMICS

Credits 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.

Section-A

Gene structure, Transcriptional regulatory elements in human genome: Promoters, enhancers, silencers, insulators, locus control regions, Transcriptional regulatory elements and cofactors in human diseases, Promoter characterization: reporter gene assay, EMSA, DNA Foot printing.

Section-B

Non-coding genes, Split genes, Alternative splicing, Role of alternative splicing, Alternative splicing in human diseases and therapy, Overlapping genes and their significance, Mechanisms of post-transcriptional gene silencing.

Section-C

Human transposons, DNA Cot curves, Unique DNA and multigene families in humans, Repetitive DNA, Genetic mechanisms underlying pathogenic sequence exchanges in unique and repetitive DNA, Evolution of human mitochondrial genome, nuclear genome, sex chromosomes and DNA sequence families.

Section-D

Principles and applications of Real-Time PCR, Microarray and its applications, cDNA array, SNP array, oligonucleotide array, Traditional DNA sequencing techniques, Second generation sequencing techniques, Advantages and limitations of Next generation sequencing technologies, Whole exome sequencing, Clinical applications of exome sequencing.

Books Recommended:

1. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
2. Brown, T. A. (2005). Genetics: A Molecular Approach. Bio Scientific Publishers Ltd., Oxford, 3rded.
3. Sudbery, P. and Sudbery, I. (2009). Human Molecular Genetics. Pearson Education, UK. 3rd ed.
4. Wang, J., Tan, A C., Tian, T. (Eds.) (2012). Next Generation Microarray Bioinformatics: Methods and Protocols. Humana Press, Totowa. www.ebook3000.com
5. Kaufmann, M., and Klinger, C. (Eds.). (2012). Functioinal Genomics: Methods and Protocols. Humana Press, Totowa, 2nd ed. www.ebook3000.com

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-472 CLINICAL GENETICS AND GENETIC COUNSELLING

Credits: 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.

Section A

Clinical Applications of Medical Genetics – Prenatal diagnosis: purpose, indications and techniques – invasive and non-invasive, Amniocentesis, Chorionic villus sampling, Ultrasonography, Fetoscopy, Maternal serum screening.

Section B

Fetal cells and DNA in maternal blood, Preimplantation diagnosis, Effect of mutagenic and teratogenic exposure in early pregnancy.Gene therapy and stem cell research in clinical genetics.

Section C

Population screening for genetic disease – adult, newborn and carrier screening – Criteria for heterozygous – screening programmes, Risk calculations, Clinical utilization of presymptomatic and predispositional testing, Presymptomatic testing for genetic diseases and malignancy.

Section D

Genetic counselling – Models and process, Role of genetic counsellors, Diagnostic problems in genetic counseling, Psychosocial aspects of genetic counselling, Genetic care pathway and preventive management guidelines, Role of social workers, and of nutritional, occupational, physical, speech therapists, psychologists and school professionals in genetics.

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 ed.
7. Rimoin, D.L., Pyeritz and Korf, P.R. (2013). Emery and Rimoin's Principles and Practice of Medical Genetics, Academic Press, New York, 6thed.
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. Antonnarakis, S.E. and Motulsky, A.G. (2010) Vogel and Mothulsku'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. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-473 BIOETHICS AND HUMAN GENETICS

Credits: 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.

Section A

History, scope and development of bioethics, Prescriptive and descriptive bioethics, Basic aspects of rights'-based and duty-based ethical theories. Utilitarian ethics.

Section B

Principalism. Basic principles of bioethics: autonomy, non-maleficience, beneficience, justice, dignity, integrity, truth-telling, veracity, etc., Informed consent and confidentiality.

Section C

Bioethical maturity, Universality of bioethics, Cross-cultural bioethics, Environmental ethics.

Section D

Vignettes on ethical dilemmas in medical genetic advances: Genetic selection, Germline gene editing, Genetic screening, Genetic determinism, Genome ownership, Genetic discrimination, Genetic insurance, Genetic privacy, ART, Modern eugenics, Euthanasia.

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.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)

(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-474: RECENT CONCEPTS IN HUMAN GENETICS

Credits 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.

Section-A

MicroRNA and other non-coding RNA, microRNA and epigenetic regulatory mechanisms, Role of microRNA and noncoding RNAs in normal development, Role of microRNA in diseases: lymphocytic leukemia, prostate cancer, colitis associated cancer, microRNA as regulators of key oncogenes and tumor suppressor genes, Mitochondrial view of microRNA, RNAi,

Section-B

Epigenetic mechanisms, Histones and DNA modifications, Methods of analysis of DNA methylation, Methods of studying histone modifications, Inter-individual variations in DNA methylation,

Section-C

Segmental deletions, duplications, CNVs and their role in human diseases.MHC complex and epigenetic mechanisms in gene regulation, Role of epigenetic mechanisms in human diseases; cancer, birth defects, epileptic disorders

Section-D

Gene transfer techniques, New approaches to gene therapy for genetic disease: Spliceosome-Mediated RNA Trans-splicing (SMaRT), Triple-helix-forming oligonucleotides, Antisense gene therapy, Ribozymes, DNA drugs and vaccines, Studying human gene function through animal models.

Books Recommended:

1. Darnell, J. (2011). RNA: Life's Indispensable Molecules. Cold Spring Harbor Laboratory Press, New York.
2. Mallick, B. and Ghosh, Z. (Eds). (2012). Regulatory RNAs Basics, Methods and Applications. Springer-Verlag, Heidelberg, Germany.
3. Mráz, M. (2007). Web book: Biological Role of microRNAs in Animal Cells, Development and Cancer. http://www.microrna.ic.cz/
4. Deng, HW. and Shen, H. (eds.). (2007). Current Topics in Human Genetics: Studies in Complex Diseases. World Scientific Publishing Co. Pte. Ltd.
5. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
6. Richard, C. F. (2011) Epigenetics : The Ultimate Mystery of Inheritance. W.W. Norton & Company, Inc. New York, 1st ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGS-475: SEMINAR

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. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGP-471: HUMAN MOLECULAR GENETICS AND FUNCTIONAL GENOMICS (PRACTICAL)

Credits 0-0-3

DNA/RNA/Proteins quantification spectrophotometrically, Polymerase chain reaction (PCR), Analysis of PCR products by agarose gel electrophoresis, PCR-RFLP analysis, PCR-SSCP analysis, Demonstration of Real-Time PCR, Biological databases searches.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGP-472 CLINICAL GENETICS AND GENETIC COUNSELING (PRACTICAL)

Credits: 0-0-2

Case studies, Designing proforma (Thalassemia, Primary ammenorhea, Mental retardation, Recurrent abortions, Prenatal screening questionnaires), Taking history, Preconceptional screening and Counseling, Case management of selected genetic diseases, Genetic Registers, importance and maintenance.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-481 GENETICS OF HUMAN CANCER
(Elective Paper)

Credits: 3-0-0

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.

Section-A
Types of cancer, Origin and classification of cancer, Genetic basis of cancer, Role of tumorsuppressors and oncogenes in cancer, Role of microRNAs in cancer.

Section-B
Cytogenetics in myeloid leukemia, acute myeloid leukaemia, Myelodysplastic syndromes and myeloproliferative disorders, Cytogenetics of haematological neoplasm, Lymphomas and chronic lymphoproliferative disorders.

Section-C
Genetic basis of solid tumors - Colorectal, breast, ovarian cancer, Uterine cancer, Solid tumor cytogenetics,

Section-D
Neoplasia-associated genomic arrangements,Cancer syndromes.Gene-environment interactions, Cancer risk assessment,

Books Recommended:

1. Gersen, S.L. and Keagle, M.B. (2005). The Principles of Clinical Cytogenetics. Humana Press, 2nd ed.
2. Kakunaga, T., Sugimura, T., Tomatis, L. and Yamasaki, H. (1988). Cell Differentiation: Genes and Cancer. IAPC Scientific Publications.
3. Lawrie, C.H. (2014). MicroRNAs in Medicine. Wiley-Blackwell, Singapore,
4. Lodish, H., Berk, A., Kaiser, A.C., Krieger, M., Scott, M.P., Bretscher, A., Ploegh, H. and Matsudaira, P. (2008). Molecular Cell Biology. W.H. Freeman and Co., New York, 6th ed.
5. Nussbaum, R.L., McInnes, R.R. and Willard, H.F. (2004). Thompson and Thompson Genetics in Medicines. W.B. Saunders and Co., New York, 6thed.
6. Rooney, D.E. and Czepulkowski, B.H. (1992). Human Cytogenetics: A Practical Approach. IRL Press, Vol. 2, 2nd ed.
7. Strachan, T. and Read, A. (2011). Human Molecular Genetics. Garland Publishers, London, 4th ed.
8. Weinberg, R.A. (2014). The Biology of Cancer. Garland Science, USA, 2nd ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-II)
(UNDER THE SCHEME OF HONOURS SCHOOL)

HGL-482: TOOLS AND TECHNIQUES IN IMMUNOLOGY
(Elective Paper)

Credits: 3-0-0

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.

Section-A
Evaluation of antigen-antibody interactions: Immunoprecipitation Immunoelectrophoresis agglutination assays, western blotting, ELISA, RIA, Immunoelectron microscopy, Microarray analysis, Flow cytometry. 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
Transplantation Immunology: Types of grafts, HLA typing, Effector mechanisms of graft rejection, Prolongation of allograft survival: General and specific immunosuppressive agents. Maternal–fetal compatibility: innate immune mechanisms, adaptive immune mechanisms, fetal derived factors.

Section-D
Tumor Immunology: Oncogenes and cancer induction, Tumor-specific transplantation antigens(TSTAs) and tumor-associated transplantation antigens (TATAs). Immune response to tumors, Tumor evasion of immune system , Cancer immunotherapy

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.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-III)
(FOR OLD STUDENTS)

HGL-521 CURRENT TRENDS IN MEDICAL GENETICS

Credits 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.

Section A

General concepts of disease and homeostasis, Principles of disease diagnosis, Disease classification, Transgenerational inheritance and Developmental Orgins of health and disease,

Section B

Ageing and Ageing syndromes,Personalized Medicine and the concept of race and ethnicity, The Human Microbiome, Precision Medicine and concept of the Exposome

Section C

Personal Genomics, Incidental findings in genomic sequencing, Non-invasive prenatal testing and screening, Overview of Systems Biology and Systems Medicine.

Section D

Genetic Testing and adoptees, Gender verification in sports events, Case studies inGenomics and patents, Overview of Genetic weapons, Ecogenetics and Synthetic Biology.

Recommended Books:

1. Crowley, L.V. (2011) Essentials of Human Disease. Jones and Bartlett Publishers, USA.
2. Donaldson, P. (2015). Genetics of Complex Disease. Garland Publishers, New York, 1st ed.
3. Gardner, A. and Davies, T. (2017). Human Genetics. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited, 2nd ed.
4. Korf, B.R. and Irons, H.B. (2013). Human Genetics and Genomics. John Wiley and Sons, Hoboken, 4th ed.
5. Skirton, H. and Patch, C. (2017). Genetics for the Health Sciences. Viva Books, New Delhi, published by arrangements with Scion Publishing Limited.
6. Strachan, T. Goodship, J. and Chinnery P. (2015). Genetics and Genomics in Medicine. Garland Publishers, New York.
7. Voit, E.O. (2012). A First Course in Systems Biology, Garland Sciences, 1st ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-III)
(FOR OLD STUDENTS)

HGL-523 GENETICS IN POST-GENOMIC ERA

Credits 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.

Section-A

Introduction to post-genomic era, Scope, Tools and challenges in post-genomic era, Omics technologies, Shotgun sequencing; map-based and whole genome-based, High throughput methods; Next Generation and Third Generation sequencing technologies.

Section-B

Prokaryotic and Eukaryotic genome annotation, Sequence alignments, Comparative Genomics,
Methylome profiling, Allelic Discrimination assays; Taqman assays, Mass array, Golden gate assay, Invader assay, High throughput expression profiling using SAGE and CAGE libraries, Tilling arrays.

Section-C

Digital Karyotyping, ChIP on ChIP, ChIP sequencing, Proteomics in post-genomics era; Top down, Bottom up approaches, Tools and techniques; Affinity, Size exclusion chromatography, Mass spectrometry, Protein sequence databases.

Section-D

1000 Genome project, ENCODE project, HapMap Project, Concept of personalized medicine, Translational genomics, Genome medicine.

Books Recommended:

1. Andreas, D., Baxevanis, B.F. and Francis, O. (2005). Bioinformatics: A Practical Guide to the Analysis of Genes and Proteins. Wiley publishing Co., 3rd ed.
2. Augen, J. (2004). Bioinformatics in the Post-Genomic Era: Genome, Transcriptome, Proteome, and Information-Based Medicine. Addison-Wesley Publishers., 1sted.
3. Dassanayake, R.S. and Silva Gunawardene, Y.I.N. (2011). Genomic and Proteomic Techniques: In Post Genomics Era. Alpha Science International Ltd., 1sted.
4. Josip, L. (2011). Introducing Proteomics: From Concepts to Sample Separation, Mass Spectrometry and Data Analysis. John Wiley and Sons Ltd., UK.
5. Kwon, Y.M. and Ricke, S.C. (2011). High-Throughput Next Generation Sequencing: Methods and Applications. Humana Press, 1sted.
6. Najarian, K. (2009). Systems Biology and Bioinformatics: A Computational Approach. CRC Press, 1sted.
7. Nikolsky, Y. and Bryant, J. (2009). Protein Networks and Pathway Analysis (Methods in Molecular Biology). Humana Press, 1sted.
8. Pevsner, J. (2009). Bioinformatics and Functional Genomics. Wiley Blackwell Publishing Co., 2nd ed.
9. Twyman, R. (2013). Principles of Proteomics. Garland Science, 2nded.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-III)
(FOR OLD STUDENTS)

HGP-581: ADVANCED PRACTICALS

Credits: 0-0-6

The students will have laboratory attachments with different faculty members for varying time span and will then be examined for different practical techniques that they have learnt during the semester.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-III)
(FOR OLD STUDENTS)

HGL-532 HUMAN GENETICS RESEARCH AND REGULATION
(Elective Paper)

Credits 3-0-0
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.

Section A

Background of research on Human Genetics, The Eugenics Movement. Need for protection of human participants in research, Development of Medical oaths and research codes (*CharkhaSamitha*, Hippocrates Oath-old and new versions, Oath of Maimonides, Physician Oath, The Nuremberg Code.

Section B

InternationalGuidelines and regulations for conducting Human Genetics research-Declaration of Helsinki,Universal Declaration on the Human Genome and Human Rights, International Ethical Guidelines for Biomedical Research Involving Human Subjects(CIOMS-WHO), Declaration of Inuyama**,**Concepts of and Issues in Reprogenetics, Consumer Genetics, Personal and Population Genomics, Regenerative medicine, Intellectual Property Rights.

Section C

National regulatory frameworks regarding Genetically Modified Organisms, Human Genetic Modification, Reproductive Genetic Testing, Cloning, Surrogacy. 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 TechniquesAct, The Transplantation of Human Organs Act.

Recommended Readings:

1. Drugs and Cosmetics Act, (1940, amended 2003), http://indianmedicine.nic.in/html/pharma/adrugsnoti.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. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-III)
(FOR OLD STUDENTS)

HGL-533: ADVANCED COURSE IN GENETIC ENGINEERING
(Elective Paper)

Credits 3-0-0
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.

Section-A

Overview of two steps and one step screening for recombinants, Lambda vectors, Yeast cloning vectors based on 2µm plasmid, YACs, BACs and PACs, Expression vectors, Translational and transcriptional fusions, Lac, Trp, Tac promoters, λPL promoter, and T7 expression system, Baculovirus based expression systems in insects.

Section-B

Mammalian expression systems, Tet-on/Tet-off system, Gene Switch technology, TA and TOPO cloning, Problems of eukaryotic protein expression in *E.coli*, Codon Bias, Protein purification by tagging approach, Genomic, cDNA and subtractive libraries, library screening methods; Hybridization based, Immunoscreening, Methods to study protein interactions, Two hybrid screening, Phage display.

Section-C

Non-biological and cell mediated gene transfer, Bactofection, Alternate gene transfer, viral vectors for gene therapy: Replacement and gutless vectors, Adenovirus, Adeno associated virus, Herpes, Lentivirus and Retrovirus as vectors, Advantages and disadvantage of each type.

Section-D

Methods of production of transgenic animals; Pronuclear microinjection, Gene transfer to embryonic stem cells, Intracytoplasmic sperm injection (ICSI), SCNT. Gene targeting in animal cells by homologus and site-specific recombination, Design of targeting vectors, Tag and exchange, Hit and run strategy, General principles of transgene behaviour, position and integration effects, Cre-Lox system.

Books Recommended

1. Dale, J.W. and von Schantz, M. (2007). From Genes to Genomes: Concepts andApplications of DNA Technology. John Wiley and Sons, UK, 2nd ed.
2. Glick, B.R., Pasternak, J.J. and Patten, C.L. (2010). Molecular Biotechnology: Principlesand Applications of Recombinant DNA. ASM Press, 4th ed.
3. Kiessling, A. and Anderson, S.C. (2006). Human Embryonic Stem Cells. Jones andBartlettLearning Publishers, 2nd ed.
4. Nicoll, D.S.T. (2008). An Introduction to Genetic Engineering. Cambridge University Press,3rd ed.
5. Primrose, S.B. and Twyman, R. (2006). Principles of Gene Manipulation and Genomics.Wiley-Blackwell, Hoboken, 7th ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGL-524: ADVANCED 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

Process of cell culture, culture environment and media, Methods of culture, Types of cell culture systems, Maintenance and storage of culture, Cell-line banking, Scaling-up of cultures for tissue engineering and commercial culture, Mesenchymal stem cells, Cell quantitation methods and cytotoxicity assays.

Section-B

Methods of cell imaging, Quantification of images, Principle and applications of fluorescent microscope ,Fluorescent in situ hybridization technique and its applications: CGH, M-FISH, SKY, COBRA-FISH, CM-FISH, FIBRE-FISH, GISH, PRINS.

Section-C

Techniques and applications of Transmitted light imaging(TEM), Confocal microscope, Scanning electron microscope, Microarray, Flow cytometer,

Section-D

Human chromosome nomenclature for G-banded chromosome, Neoplasia, In situ hybridization.

Recommended Books:

1. Aruni, A., Wilson and Ramadass, P. (2011). Animal Cell Culture. MLIP Publishers, Chennai, India.
2. Freshney, R.I. (2000). Culture of Animal Cells: A Manual of Basic Techniques. Wiley Liss, Inc., New York, USA, 4th ed.
3. Gerson, S.L. (1999). Principles of Clinical Cytogenetics. Humana Press, Totowa, USA.
4. Purandaare, H. and Chakravarty, A. (2000). Human Cytogenetic Techniques and Clinical Applications. Bhalani Publishing House, Mumbai, India.
5. Roif-Dieter (1999). Diagnostic Cytogenetics. Springer-Verlag, New York.
6. Schaffer, L.G., McGowan-Jordan, J. and Schmid, M. (2016). ISCN-2016. Karger Publishers, Germany.
7. Stephens, D. (2006). Cell Imaging. Scion Publishing Limited, Oxfordshire, UK.
8. Vanjak-NovaKovic, G. and Freshney, R.I. (2006). Culture of Cells for Tissue Engineering. Wiley-Liss, New York, USA.
9. Wegner, R.D. (1999). Diagnostic Cytogenetics. Springer-Verlag, New York, USA.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGL-552 APPLIED HUMAN MOLECULAR GENETICS

Credits 3-0-0
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.

Section-A

Principles and strategies in identifying disease gene, Genetic and physical mapping of human genome, Mapping and cloning of human disease genes, Direct and indirect DNA testing and its applications.

Section-B

DNA-based diagnosis of genetic diseases, Population screening. Principles and applications of Comparative genomic hybridization (CGH), DNA foot printing, DNA microarray.

Section-C

DNA dragnets, Multiplex ligation dependent probe amplification (MLPA), Serial analysis of gene expression (SAGE), Mass spectrometry (MS), Tandem mass spectrometry (TMS) and Next generation sequencing.

Section-D

Molecular genetics of retinoblastoma, Glaucoma, Marfan syndrome, DMD, Cystic fibrosis, Huntington's disease, Complex human diseases like NIDDM, Hypertension, Cardiovascular disorders, Obesity, Molecular genetics of mitochondrial disorders.

Books Recommended

1. Brown, T.A. (2002). Genomes. Oxford Scientific Publications, London.
2. Brown, T.A. (2005). Genetics: A Molecular Approach. Bios Scientific Publishers Ltd., Oxford, 3rd ed.
3. Brown, T.A. (2006). Genomes 3. 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. Gleck, B.R. and Pasternack, J.R. (2003). Molecular Biotechnology: Principles and Applications of Recombinant DNA. ASM Press, Washington.
6. Korf, B.R. (2007). Human Genetics and Genomics. Blackwell Scientific Publication, USA, 3rd ed.
7. Krebs J.E., Goldstein, E.S. and Kilpatrick, S.T. (2012). Genes-XI. Jones and Barlett Publishers, Inc., USA.
8. Lewin, B. (2008). Genes-IX. Jones and Barlett Publishers, Inc., USA.
9. Schena, M. (2003). Microarray Analysis. Wiley-Liss, New York.
10. Strachan, T. and Read, A. (2010). Human Molecular Genetics. Garland Publishers, London, 4th ed.
11. Sudbery, P. and Sudbery, I. (2009). Human Molecular Genetics. Pearson Education, UK, 3rd ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGL-555: BIRTH DEFECTS AND DYSMORPHOLOGY

Credits 3-0-0
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.

Section-A

Syndromes, Dysmorphology and Birth defects, Evaluation of dysmorphology: Pregnancy history, Birth history, Medical history, Physical features, Diagnostic approach to dysmorphic patients.

Section-B

Indication for prenatal diagnosis, Fetal disorders, Rhesus disease, Fetal alcohol syndrome, Stillbirth and neonatal death, Spontaneous abortion, Advanced maternal age, Abnormal ultrasound findings

Section-C

Classification of birth defects, Minor and major congenital abnormalities, Teratogenic effect on development, CHARGE and VACTERAL association, Single gene defects, Multifactorial inheritance, Role of genetic counselling in dysmorphology.

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Section-D

Microcephaly, Limb as developmental model and related syndromes, Errors of morphogenesis, Disorders of sexual differentiation, Disorders of sex chromosomes, Chromosome disorders and behavioural phenotypes.

Books Recommended:

1. Fundukian, L.J. (2010). Pearls of Dysmorphology. The GALE Encyclopedia of Genetic disorders. China Translation and Printing Services Limited, China, Vol. I and II, 3rd ed.
2. Hartl, D.L. (2014). Essential Genetics: A Genomic Perspective. Jones and Barlett Learning, USA, 6th ed.
3. Jones, R.E. and Lopez, K.H. (2014). Human Reproductive Biology. Academic Press (Elsevier), 4th ed.
4. Lupski, J.R. and Stankiewicz, P. (2006). Genomic Disorders: The Genomic Basis of Disease. Humana Press, New Jersey.
5. Netter, F.H. (2003). Atlas of Human Anatomy. Icon Learning Systems, USA, 3rd ed.
6. Nussbaum, R.L., McInnes, R.R., Willard, H.I. and Boerkoel, C.I. (2001). Thompson and Thompson Genetics in medicine. W.B. Saunders and Co., 6th ed.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGL- 556 Structural Bioinformatics and Pharmacogenomics

Credits 3-0-0
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.

Section-A

Secondary and Super Secondary structures,α-helix, 3_{10}, π helix, β-sheet, turns/loops, Domains;alpha, beta, alpha+ beta domains with examples, Leucine zippers, Zn fingers, Helix bundle, β propeller, Rossman fold, DNA higher order structures, puckering and Hoogstein base pairing, Triple helices, G quadruplexes, DNA topology, Supercoiling, Role of non covalent interactions in protein and DNA structure.

Section-B

Protein folding problem and theories, Energy landscape, Structure prediction methods; homology modeling and Ab-initio, GOR, Chou-Fasman, PSI- PRED. Measures of accuracy Q_3, Sov. Overview of X-ray diffraction and NMR spectroscopy for structure determination of proteins, Principle of protein purification.

Section-C

Protein structure classification databases: HSSP, FSSP, CATH, SCOP,Motif, profile and domain databases, Introduction to HMM (Hidden Markov Models) in structure prediction, sequence and structure database, PDB, UniProt.

Section-D

Principle of drug discovery, Target identification, validation, screening, Hit to lead generation, Pharmacokinetcs and pharmacodynamics, ADME properties, Bioisosteres, Bioinformatic tools in drug discovery, insilico screening of targets, molecular docking, Pharmacogenomics with examples of Warfarin, Abacavir, Herceptin and Thiopurine drugs, CYP2D6 Pharmacogenetics, Role of SNPs in response to anti-diabetic drugs, antihypertensive drugs.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS

(Credit Based Evaluation & Grading System) (Semester-IV)

(FOR OLD STUDENTS)

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.
4. Brandon, C. and Tooze, J. (1999) Introduction to Protein Structure. Garland Publishers, London.
5. Hartl, D.L. and Jones, E.W. (2005). Genetics: Analysis of Genes and Genomes. Jones and Barlett Publishers, Massachusetts.
6. Jain, K.K. (2001). Drug Discovery: Current Trends and Future Prospects. Urch Publishers,London.
7. Leach, A.R. (2001). Molecular Modelling. Principles and Applications. Addison Wesley Longman, Essex.
8. Nelson, D.L. and Cox, M.M. (2008). Lehninger's Principles of Biochemistry. W.H. Freeman and Co., New York.

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGP-552: APPLIED HUMAN MOLECULAR GENETICS (PRACTICAL)

Credits: 0-0-3

Max. Marks : 100

DNA isolation, Amplification of genomic DNA by PCR, Agarose gel electrophoresis of amplified products, Polyacrylamide gel electrophoresis (PAGE), PCR-SSCP analysis, Demonstration of RT-PCR, Biological database searches (MEDLINE, NCBI, ENSEMBL, UCSC, GDB, PDB, HGMD, RetNet).

M.Sc. (HONS. SCHOOL) HUMAN GENETICS
(Credit Based Evaluation & Grading System) (Semester-IV)
(FOR OLD STUDENTS)

HGD-595: DISSERTATION

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