



JIWAJI UNIVERSITY, GWALIOR
Master of Science in
MOLECULAR & HUMAN GENETICS
(2012-2014)



The course for Master of Science (M. Sc.) in Molecular & Human Genetics shall comprise of four semesters of six months duration each. Semester I-III shall include four theory papers and two practical courses each of 100 marks, out of which 85 marks are for final examinations and 15 marks for internal examinations. Each practical course will include one seminar on topics related to the paper/course work for the semester. In IInd and IIIrd semesters each, the students shall carry out a short project work in the subject related to the course under the supervision of the faculty involved and submit a report along with a presentation/seminar for evaluation. In IVth semester there are 2 theory papers and one practical paper. There shall be a related internal assessment for each theory and practical course, involving 15 marks each. In addition, the students are required to undergo about 2-3 months of summer training, preferably at the parent institution and submit the dissertation work as part of their M. Sc. degree in the IVth semester. For internal examinations (internal assessment tests) 3 tests on each paper in each semester shall be conducted and average marks of best of two shall be considered for final awards. Head of the Department/Co-ordinator shall send the marks of internal assessments of theory and laboratory courses to the Registrar before respective University examination. The outline of the course structure is as follows:

Marks Allotted: 100 each
(85 Ext + 15 Int)

Semester I

Paper MHG-101: Principles of Genetic Inheritance

Paper MHG-102: Basic Human Genetics and Model Organisms in Genetic Studies

Paper MHG-103: Molecular Organization of Chromatin and Cytogenetics

Paper MHG-104: Tools & Techniques

Paper MHG-105: Practical based on papers 101 & 102

Paper MHG-106: Practical based on papers 103 & 104

Semester II

Paper MHG-201: Molecular Genetics and Genomics

Paper MHG-202: Molecular Structure & Functions of the Cell and Cancer Biology

Paper MHG-203: Biomolecules, Cellular Metabolism and Human Metabolic Disorders

Paper MHG-204: Developmental Genetics and Genetics of Human Development & Developmental Defects

Paper MHG-205: Practical based on papers 201 & 202

Paper MHG-206: Practical based on papers 203 & 204

Semester III

Paper MHG-301: Human Molecular Genetics and Human Genomics

Paper MHG-302: Molecular and Clinical Genetics of Human Diseases and Genetic Counseling

Paper MHG-303: Recombinant DNA Technology and Molecular Diagnostics in Human Diseases

Paper MHG-304: Immunogenetics and Molecular Genetics of Human Pathogens

Paper MHG-305: Practical based on papers 301 & 302

Paper MHG-306: Practical based on papers 303 & 304

Semester IV

Paper MHG-401: Population Genetics, Human Evolutionary Genetics and Behaviour Genetics

Paper MHG-402: Biostatistics & Bioinformatics

Paper MHG-403: Practical based on theory papers.

Paper MHG-404: Dissertation (200) & Viva-voce Examination (100)

300

Detailed Syllabus for M. Sc. in Molecular & Human Genetics

Semester I

Paper MHG-101: Principles of Genetic Inheritance

Unit I

1. Mendel's laws of inheritance
 - 1.1 Law of segregation
 - 1.2 Law of independent assortment
2. Chromosomal theory of inheritance
3. Extensions of Mendelism
 - 3.1 Allelic variation and gene function- Dominance relationships and Complications in the concept of dominance
 - 3.2 Multiple allelism, allelic series
 - 3.3 Testing gene mutations for allelism: complementation test
 - 3.4 Visible, sterile and lethal mutations
 - 3.5 Pleiotropy
4. Gene interactions and modifying genes

Unit II

5. Sex chromosomes and sex-linked inheritance
 - 5.1 Sex chromosomes and their meiotic behaviour
 - 5.2 Sex-linked inheritance in *Drosophila* and *human*
6. Linkage and crossing over
 - 6.1 Concept
 - 6.2 Cytological demonstration of crossing Over in *Drosophila*
 - 6.3 Genetic distance and physical distance
 - 6.4 Genetic and cytological crossing over
7. Linkage and crossing over: Preparation of Linkage map
 - 7.1 Genetic recombination and construction of genetic maps in *Drosophila* (Three-point test Cross) and yeast (Tetrad analysis).
 - 7.2 Interference and coincidence
 - 7.3 Mitotic recombination
8. Gene mapping in bacteria
 - 8.1. Transformation
 - 8.2. Conjugation
 - 8.3. Transduction

Unit III

9. Concept of marker and balancer chromosomes in *Drosophila*
10. Fine structure of gene (rII locus)
 - 10.1 Intragenic recombination in Bacteriophage
 - 10.2 Deletion & complementation mapping in T4 phage
11. Extranuclear inheritance & maternal effects
 - 11.1 Organelle heredity (mitochondria & chloroplast); Petite mutations
 - 11.2 Infectious heredity (Cytoplasmic inheritance) in symbionts (*Paramecium*) & *Drosophila*
 - 11.3 Maternal inheritance: Ephestia pigmentation and snail coiling
12. Statistical tests in genetic analysis
 - 12.1 Application of laws of probability (product rule, sum rule. binomial probability)
 - 12.2 Chi square test and its application in the analysis of genetic data

Unit IV

13. Inheritance of quantitative traits
 - 13.1 Continuous and discontinuous variation
 - 13.2 Genetic variance and heritability. (Narrow sense and broad sense); Quantitative trait loci (QTL)
14. Polygenic inheritance: Johansen's pure lines, Polygenes
15. Environmental effects and gene expression I
 - 15.1 Effect of external and internal environment
 - 15.2 Phenocopies
16. Environmental effects and gene expression II
 - 16.1 Twin studies and IQ
 - 16.2 Concordance – Discordance
 - 16.3 Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity

Unit V

17. Sex determination:
 - General concepts and variations in the mechanisms of sex determination in eukaryotes: Heterogametic & Homogametic, Haplodiploidy, role of Environmental factors; Mosaics and Gynandromorphs; Sex determination in Melandrium.
18. Sex determination in *Drosophila*
 - 18.1 Genic Balance theory
 - 18.2 Molecular mechanism of sex determination
 - 18.3 Mechanism of Sexual dimorphism
19. Sex determination in Mammals: Human Y chromosome- evolution, structure, molecular organization and its role in sex determination
20. Sex determination in mammals: Endocrine & Molecular Mechanism of sex determination

Recommended Books

1. An Introduction to Genetic Analysis, 7th Ed., Griffiths et al, Freeman, 2000
2. Genetics, 3rd Ed., Strickberger, Macmillan, 1985
3. Genetics: Analysis of Genes and Genomes, 6th Ed., Hartl and Jones, Jones & Bartlett, 1998
4. Concepts of Genetics, 9th Ed., Klug and Cummings, Pearson, 2009
5. Principles of Genetics, 7th Ed., Tamarin, Tata McGraw Hill, 2002
6. Principles of Genetics, 3rd Ed., Snustad and Simmons, Wiley, 2003
7. Encyclopaedia of Genetics, Vol I and II, Reeve, Pentagon Press, 2002
8. Schaum's Outline of Genetics, 4th Ed., Elrod and Stansfield, McGraw Hill, 2002
9. Principles of Genetics, Gardner et al., John Wiley 1991

Paper MHG-102: Basic Human Genetics, Cytogenetics and Model Organisms in Genetic Study

Unit I

1. History of Human Genetics
2. Pedigrees- gathering family history. pedigree symbols, construction of pedigrees; presentation of molecular genetic data in pedigrees
3. Pedigree analysis of monogenic traits:
 - 3.1 Autosomal inheritance-dominant, recessive
 - 3.2 Sex-linked inheritance- X-linked recessive, dominant; Y -linked
 - 3.3 Sex-limited and sex-influenced traits
 - 3.4 Mitochondrial inheritance
 - 3.5 MIM number
4. Complications to the basic pedigree patterns I: Non-penetrance, variable expressivity, pleiotropy, onset, dominance problem; anticipation, compound heterozygosity

Unit II

5. Complications to the basic pedigree patterns II: Genomic imprinting and uniparental disomy, spontaneous mutations, mosaicism and chimerism, male lethality, X- inactivation, Consanguinity and its effects in the pedigree pattern, allele frequency in population.
6. Complex traits- polygenic and multifactorial
 - 6.1 Approaches to analysis of complex traits- 'Nature vs nurture', role of family and environment, monozygotic and dizygotic twins and adoption studies
 - 6.2 Polygenic inheritance of continuous (quantitative) traits, normal growth charts, Dysmorphology
 - 6.3 Polygenic inheritance of discontinuous (dichotomous) traits- threshold model, liability and recurrence risk
7. Genetic susceptibility in complex traits. alcoholism, cardiovascular disease, diabetes mellitus and obesity
8. Estimation of genetic components of multifactorial traits: emperic risk, heritability, coefficient of relationship, application of Baye's theorem

Unit III

9. Application & roles of probability in prediction analysis of genetic data
10. Pharmacogenomics:
 - 10.1 Concept
 - 10.2 Polymorphism relating to drug metabolism and disposition
 - 10.3 Polymorphism affecting drug targets
11. Human cytogenetics
 - 11.1 Origins and developments in the study of human cytogenetics
 - 11.2 Techniques in human chromosome analysis (including molecular techniques)
 - 11.3 Human karyotype: banding, nomenclature of banding
 - 11.4 Nomenclature of aberrant karyotypes
12. Human chromosomal pathologies:
 - 12.1 Common syndromes due to numerical chromosome changes
 - 12.2 Common syndromes due to structural alterations (translocations, duplications, deletions microdeletion syndromes, fragile sites, etc.)

Unit IV

13. Tissue culture methods: Lab preparation, sterilization, culture media, sera & growth factors,
14. Principles and methods of tissue culture: lymphocyte and fibroblast culture; culture of cancer/tumor cells/tissues; cell-lines; applications of tissue culture techniques in clinical cytogenetics.
15. Stem cells: Origin, culture, properties and therapeutic applications

16. General idea of Pharmacogenetics, Ecogenetics, Teratogenetics and Biochemical genetics (Blood groups & Serology; Protein polymorphism & its significance)

Unit V

17. Life cycles and advantages of model organisms used in genetic studies I:
17.1 T4 and lambda phages
17.2 *E. coli*
17.3 *Saccharomyces cerevisiae* & *S. pombe*
17.4 *Neurospora*
18. Life cycles and advantages of model organisms used in genetic studies II:
18.5 *Trypanosoma*
18.6 *Caenorhabditis*
18.7 Zebra fish
18.7 Mouse
19. Conventions of nomenclature of genes and gene products in different model systems I:
Bacteria and viruses
20. Conventions of nomenclature of genes and gene products in different model systems II:
Yeast, mouse and human (HGNC recommendations).

Recommended Books

1. Human Molecular Genetics 3, Strachen & Read, Blackwell, 2004
2. An Introduction to Molecular Human Genetics, Pasternak, Wiley, 2000
3. Molecular Biology of the Gene, 6th Ed., Watson et al, CSH Press, 2008
4. Human Genetics, Lewis, McGraw Hill, 2007
5. Human Genetics: Problems and Approaches, 3rd Ed, Vogel, Springer, 1997
6. Genetic Nomenclature Guide Trends in Genetics Elsevier 1998
7. Molecular and Genetic Analysis of Human Traits, Maroni, Blackwell, 2001
8. Thompson & Thompson's Genetics in Medicine, 7th Ed, Nussbaum et al, Elsevier, 2007
9. An Atlas of Drosophila Genes: Sequences & Molecular Features, Maroni, Oxford, 1993
10. Culture of Animal Cells, 4th Ed., Freshney, Wiley, 2000
11. Animal Cell Culture & Technology, 2nd Ed, Butler, Bios, 2008
12. Animal Cell Culture, 3rd Ed, Masters, Oxford, 2000
13. Human Cytogenetics- A practical Approach, 3rd Ed., Rooney et al, 2001
14. Human Chromosome: Structure, Behavior and Effects, 3rd Ed., Therman and Susman, 1993
15. Foundations of Comparative Genomics, Mushegian, Elsevier, 2007
16. Basic Human Genetics Mange and Mange Sinauer Assoc 1999
17. Essentials of Medical Genetics Smith
18. Human Genetics Vogel and Motulsky Springer Verlag 1982
19. *Drosophila* .A Laboratory Handbook Ashburner Cold Spring Harbor 1989

Paper MHG-103: Molecular Organization of Chromatin and Cytogenetics

Unit I

1. Chromatin: Prokaryotic and eukaryotic chromosome
2. Structural Organization of Eukaryotic chromatin
 - 2.1. Chemical constituents: histones & DNA
 - 2.2. Nucleosome and higher order organization
 - 2.3 Chromatin remodelling
3. Metaphase chromosome: centromere and kinetochore, telomere and its maintenance
4. Structural and functional organization of interphase nucleus

Unit II

5. Functional states of chromatin and alterations in chromatin organization: DNase I hypersensitivity
6. Methylation: DNA methylation, epigenetic inheritance and gene expression
7. Giant chromosomes: Structural and functional characteristics of Polytene chromosomes
8. Heterochromatin and euchromatin; position effect variegation.

Unit III

9. DNA Renaturation & Denaturation and Complexity of genome; Cot value
10. Chromosome bandings: Principles, methods and applications:
 - 10.1 G, C, Q, R & T banding
 - 10.2 Chromosome painting (FISH, GISH, CGH, SKY.)
11. Chromosomal anomalies: Numerical aberrations & their genetic consequences
12. Chromosomal abnormalities: Structural
 - 12.1 Types
 - 12.2 Meiosis in inversion and translocation heterozygotes; breakage-fusion-bridge cycles
 - 12.3 Cell cycle specific chromosomal aberrations in somatic chromosomes
 - 12.4 Sister chromatid exchanges and somatic crossing over

Unit IV

13. Cell cycle and its regulation: *Xenopus*, Yeast and mammals
14. Mitotic Cell Division: Molecular mechanisms
 - 14.1 Mitotic spindle and arrangement of chromosomes on equator
 - 14.2 Regulation of exit from metaphase
 - 14.3 Chromosome movement at anaphase
15. Meiotic Cell division
 - 15.1 Overview of the process
 - 15.2 Meiosis specific cellular changes: Molecular & Biochemical
 - 15.3 Genetic consequences of meiosis
16. Structural organization and genetics of organelle genome
 - 16.1 Mitochondrial genome
 - 16.2 Chloroplast genome

Unit V

17. Dosage compensation in mammals
 - 17.1 Lyon's Hypothesis
 - 17.1 Sex chromatin
 - 17.2 Human X chromosome
18. Molecular mechanism of X-chromosome inactivation
19. Dosage compensation in *Drosophila*
20. General idea of dosage compensation in nematode, *C. elegans*.

Recommended Books

1. Molecular Biology of the Gene, 6th Ed., Watson et al, CSH Press, 2008
2. Genes IX, Benjamin Lewin, Jones and Bartlett, 2008
3. Human Chromosomes, 4th Ed., Miller and Therman, 2001
4. Human Chromosome: Structure, Behavior and Effects, 3rd Ed., Therman and Susman, 1993
5. First years of Human Chromosomes, Harper, Scion, 2006
6. Molecular Biology of the Cell, 4th Ed., Alberts et al, Garland, 2002
7. Human Cytogenetics- A practical Approach, 3rd Ed., Rooney et al, IRL, 2001
8. Chromosome aberrations -Basic and Applied Aspects, Obe and Natarajan Springer 1990
9. Structure and Function of Eukaryotic Chromosomes, Hennig, Springer 1987
10. The Chromosome Hamsew and Flavell Bios 1993
11. The Eukaryotic Chromosome Bostoc and Surnoer Elsevier 1980
12. The Principles of Clinical Cytogenetics Gersen and Keagle Hwnana 1999
13. Heterochromatin: Molecular & Structural aspects R. S. Verma Cambridge Uni.Press 1988

Paper MHG-104: Tools & Techniques

Unit I

1. Basic principles and applications of:
 - 1.1 Light & Dark-field Microscopy
 - 1.2 Phase-contrast Microscopy
2. General idea on the principles and applications of: Transmission & Scanning Electron Microscope
3. General idea on the principles and applications of:
 - 3.1 Fluorescence Microscopy
 - 3.2 Confocal Microscopy
4. Atomic force microscopy

Unit II

5. Introduction to principle and applications of Mass spectrometry
6. Basic Principles and applications of Absorption spectrophotometer
7. Basic Principles and applications of Fluorescence spectrophotometer
8. Introduction to principle and applications of Infra Red (IR) spectrometry

Unit III

9. Centrifugation: Types, rotors and applications
10. General principles, methods and applications of: Paper, starch and Agarose gel electrophoresis
11. General principles, methods and applications of Polyacrylamide gel electrophoresis
12. Isoelectric focusing and 2D gel electrophoresis

Unit IV

13. General principle, applications and methods of: Partition Chromatography
14. General principle, applications and methods of: Adsorption Chromatography
15. General idea about principle and applications of X-ray diffraction
16. General idea about ESR & NMR

Unit V

17. Gel Documentation system
18. Cell Imaging: Photomicrography & Image analysis system
19. Radiotracer techniques:
 - 19.1 Properties and units of radioactivity & Half-life
 - 19.2 Working and applications of GM counter & liquid scintillation counter
20. Principle, methods and applications of Auto-radiography

Recommended Books

1. Cell Imaging, Stephans, Scion, 2006
2. Physical Biochemistry, 2nd Ed., Freifelder, Freeman, 1999
3. Practical Biochemistry, 5th Ed., Wilson and Walker, Cambridge, 2003
7. Electrophoresis in Practice, 4th Ed., Westermeir, Wiley, 2005
8. Basic Methods in Microscopy, Spector, CSHL Press, 2006

Practical Paper MHG-105: (Based on Theory Papers MHG-101 & MHG-102)

Principles of Genetic Inheritance, Basic Human Genetics and Model Organisms in Genetic Studies

1. *Drosophila*: Collection, handling and laboratory culture
2. Life cycle study of *Drosophila*
3. Structural identification of *Drosophila*: wild type and phenotypic mutants
4. Monohybrid and dihybrid crosses in *Drosophila*
5. Sex linked inheritance in *Drosophila*
6. Linkage and crossing over in *Drosophila*
7. Gene mapping by making three point test cross
8. Study of chromosomal disorders (Phenotypes & karyotypes) and preparation of Pedigree chart of some common phenotypic characters of human
9. Facial landmarks and dermatoglyphia
10. Study of Sex-chromatin from buccal smear and hair root cells
11. C, G and Fluorescence banding
12. Study of life cycles of some model systems: Yeast, mice/rat, *Dictyostelium*, etc.
13. Calculation of the coefficient of relationship (r) in pedigree
14. Culture (lymphocyte, fibroblast, etc.) of human tissues for chromosomal Studies

Scheme of paper 105:

Total marks: 85

1. Analysis of Mono hybrid / Dihybrid / Sex-linkage crosses	10
2. Linkage & Crossing over / Three Point Test cross & Gene mapping	10
3. Preparation of pedigree/ Sex chromatin/ Facial landmarks/ Dermatoglyphia	10
4. Lymphocyte culture & chromosome preparation	15
5. Spotting (8): (<i>Drosophila</i> mutants, pedigrees, life cycle studies, Photomicrographs, etc.)	20
6. Viva Voce	10
7. Practical record	10
Total	85

Paper MHG-106: (Based on Theory Papers MHG-103 & MHG-104)
Molecular Organization of Chromatin, Cytogenetics and Tools & Techniques

1. Study of metaphase chromosomes from rat/mice bone marrow
2. Study of mitosis and effect of microtubule inhibitor on mitosis in onion root tip
3. cells
4. Study of Meiosis in grasshopper testis
5. Study of meiosis from super ovulated oocytes of female and male testis
6. mice/rat
7. Preparation of synaptonemal complex from mouse testis
8. Study of polytene chromosomes in *Drosophila/Chironomous* larval salivary
9. glands
10. Other practicals related to cytogenetics.
11. Principles and handling of bright field, phase contrast and fluorescence microscope
12. Principles of Spectrophotometry and applications
13. Principles of centrifugation and applications
14. Separation of biomolecules by gel filtration
15. Electrophoresis- SDS-PAGE & Agarose gel
16. Chromatography- Paper & TLC

Scheme for Paper 106:

Total marks: 85

1. Study of mitosis / meiosis from insect/ animal models	10
2. Study of polytene chromosomes from insect model	10
3. Study of working of tools in cytogenetic analysis/ biochemical estimations (spectrophotometer / Centrifugation /Microscopy / Gel filtration/Chromatography/Electrophoresis, etc.)	15
4. Spotting (8)	20
6. Seminar	10
7. Viva Voce	10
8. Practical Record	10
Total	85

Semester II

Paper MHG-201: Molecular Genetics & Genomics

Unit I

1. Properties & evolution of genetic material
 - 1.1 Flow of genetic information
 - 1.2 Organization of viral and bacterial genomes
2. Eukaryotic genome
 - 2.1. C-value paradox
 - 2.2. Repetitive DNA
 - 2.3. General concept of a gene
 - 2.4 Non-coding genes
3. Gene families: Molecular organization and evolutionary significance of globin, histone and actin gene families.
4. Replication:
 - 4.1 DNA Polymerases
 - 4.2 Replicons, origin & termination
 - 4.3 Replisomes; Genes controlling replication

Unit II

5. Molecular mechanisms of replication in bacterial & Eukaryotic chromosomes, rDNA and mitochondrial DNA
6. Mutagenesis & Mutation
 - 6.1 Types & origin
 - 6.2 Mechanisms
 - 6.3 Detection and isolation
7. DNA damage and repair mechanisms
8. Recombination
 - 8.1 Homologous recombination
 - 8.2. Gene conversion
 - 8.3 Site-specific recombination

Unit III

9. Transcription in Prokaryotes: Prokaryotic RNA polymerase, sigma factors, initiation and termination.
10. Eukaryotic RNA polymerases and their promoters; Transcription units.
11. Transcription activators and repressors: Identification of transcription factors, DNA binding motifs, reporter assay & repressors
12. Processing of transcripts:
 - 12.1 5' capping.
 - 12.2 3' Polyadenylation
 - 12.3 Splicing
 - 12.4 RNA editing

Unit IV

13. Regulation of gene expression- I : transcription initiation
 - 11.1. Concept of Operons (lac and trp as examples) and regulon
 - 11.2. Positive and negative regulation
 - 11.3. Enhancers and promoters
14. Regulation of gene expression-II: Regulation by attenuation and anti-termination
15. Mechanisms of steroid hormone & stress induced gene expression
16. Post-transcriptional regulation
 - 16.1. Alternative splicing
 - 16.2. Transport and targeting of RNA
 - 16.3. Post-transcriptional gene silencing

Unit V

17. Translation

17.1 General mechanism

17.2 Role of rRNA & tRNA in translation

17.3 Translational control of mRNA and targeting of proteins

18. Transposable Genetic Elements in prokaryotes & Mechanism of transposition

19. Transposable genetic elements in: Yeast, *Drosophila*, maize and Retrotransposons

20. Genomics concepts

20.1 Structural Genomics

20.2 Functional Genomics

Recommended Books

1 An Introduction to Genetic Analysis, Griffiths et al Freeman 2000

2 Applied Molecular Genetics. Meesfeld Wiley-Liss 1999

3 Gene Regulation Latchman. Chapman and Hall 1995

5 Genes and Genome. Berg and Singer 1998

6 Genetic Switch. Ptashne Cell & Blackwell 1986

7 Microbial Genetics. Maloy and Freifelder Jones and Bartlett 1994

8 Modern genetic Analysis. Griffith et al Freeman 1999

9. Molecular Biology of the Gene, 6th Ed., Watson et al, CSH Press, 2008, 1987

10. Genes IX (VII, VIII), Benjamin Lewin, Jones and Bartlett, 2008

11. Genes and Genome, Singer & Berg, USB, 1991

12. Genetic Analysis of Genes and Genomes, 6th Ed, Hartl & Jones, Jones and Bartlett, 2005

13. Fundamental Bacterial Genetics, Trun & Trempey, Blackwell, 2004

14. Genomes 3, TA Brown, Garland, 2007

Paper MHG-202: Molecular Structure & Functions of the Cell and Cancer Biology

Unit I

1. Plasma Membrane:
 - 1.1 Molecular organization
 - 1.2 Transport across membrane
2. Mechanisms of Endocytosis and Exocytosis.
3. Endomembrane system: Ultrastructure of EPR & transport through EPR
4. Endomembrane system: Ultrastructural organization of Golgi complex & Transport through GC.

Unit II

5. Mitochondria:
 - 5.1 Ultrastructure
 - 5.2 Mitochondrial transport
 - 5.3 Chemiosmotic theory and respiratory chain complexes
6. Ultrastructure of nucleus & nucleolus
7. Mechanisms of intracellular digestion: Structure & functions of Lysosomes.
8. Cytoskeletons:
 - 6.1 Microfilaments: Structural organisation. Cell motility and cell shape
 - 6.2 Intermediate filaments

Unit III

9. Microtubule: Ultrastructure and functional organization
10. Structure and functions of cilia, flagella, and centriole
11. Cell-Cell Interaction
 - 11.1. Cell adhesions
 - 11.2. Cell junctions (Occluding, Anchoring & Gap junctions)
 - 11.3. Extracellular matrix: Organization & Functions; Integrins
12. Structure and biosynthesis of Ribosome

Unit IV

13. Structure and functions of Peroxisomes
14. Programmed cell death:
 - 14.1 Definition and General Characteristics; Necrosis & PCD
 - 14.1 Morphological and Biochemical changes
 - 14.2 Molecular pathways of PCD
15. Signal transduction
 - 15.1 Intracellular receptor and cell surface receptors
 - 15.2 Signalling via G-protein linked receptors (PKA, PKC, CaM kinase)
16. Signaling:
 - 16.1 Enzyme linked receptor signalling (Growth factor receptor signaling; JACK-STAT pathway)
 - 16.2 Network and cross-talk between different signal mechanisms
 - 16.3 Role of NO & CO in cell signaling.

UNIT V

17. Cancer Biology
 - 17.1 Cancer & environment
 - 17.2 Biochemical & structural Changes in cancer cells
 - 17.3 Tumor progression: angiogenesis & metastasis
18. General idea of Oncogenes and Tumor suppressor genes
19. Molecular mechanisms of tumorigenesis:
 - 19.1 Cell cycle check-point defects, centrosome defects & telomerase defects.
 - 19.2 Tumor specific markers

- 19.3 Epigenetic regulation: Methylation
- 19.4 Genomic instability (MSI & LOH)
- 20. Chromosomal basis of cancer:
 - 20.1 Philadelphia chromosome, Retinoblastoma, Burkitt's lymphoma
 - 20.2 Oncogene amplification (HSR & DM)
 - 20.3 Aneuploidy in neoplasia

Recommended Books

1. Molecular Biology of the Cell, 4th Ed., Alberts et al, Garland, 2002
2. Molecular Cell Biology, 6th Ed., Lodish et al, Freeman & Co. 2008
3. Cell and Molecular Biology, Karp, Wiley, 2002
4. Developmental Biology, 8th Ed., Gilbert, Sinauer, 2006
5. Essential Cell Biology Alberts et al Garland 1998
6. Cell and Molecular Biology, 8th Ed., De Robertis, Lea & Febiger, 1987.
7. The Cell, Cooper, ASM Press, 2004.
8. Molecules of Death, 2nd Ed., Waring et al, ICP, 2007
9. Principles of Anatomy and Physiology, 11th Ed., Tortora & Derrickson, Wiley, 2006.

Paper MHG-203: Biochemistry and Metabolic Disorders

Unit I

1. Bioenergetics
 - 1.1. Second law of thermodynamics
 - 1.2. Free energy
 - 1.3. High-energy compounds
 - 1.4. Water
 - 1.5. Oxidative phosphorylation
2. Carbohydrates
 - 2.1. Introduction
 - 2.2. Mucopolysaccharides and related disorders
 - 2.3. Glycolysis
 - 2.4. Krebs cycle
3. Carbohydrate metabolism
 - 3.1. Gluconeogenesis
 - 3.2. Pentose phosphate pathway
 - 3.3. Glycogenesis and glycogenolysis.
4. Disorders of glycogen metabolism

Unit II

5. Structure and function of water- and lipid- soluble vitamins
6. Lipids
 - 6.1. Fatty acids: synthesis and oxidation of fatty acid
 - 6.2. Ketogenesis
 - 6.3. Metabolism of cholesterol
7. Lipoproteins: role in lipid transport and storage
8. Prostaglandins: structure and function

Unit III

9. Disorders of lipid metabolism
10. Hormones
 - 10.1. Characteristics
 - 10.2. Mechanism of action of peptide and steroid hormones
11. Hormone receptors and diseases
12. Amino acids and peptides
 - 12.1. Essential and non-essential amino acids
 - 12.2. Porphyrins and bile pigments

Unit IV

13. Metabolism of essential amino acids and related disorders
14. Small peptides and their biomedical importance
15. Structure- conformation-function relationship of proteins: Insulin, Hemoglobin and Collagen
16. Protein folding and Protein degradation

Unit V

17. Enzymes:
 - 17.1. General properties; Ribozymes
 - 17.2. Enzyme kinetics: derivation of Michaelis-Menten equation and calculations based on it & L-B plot
 - 17.3. Enzyme inhibition
 - 17.4. Mechanism of action (lysozyme & chymotrypsin)
 - 17.5. Regulation of enzyme activity
18. Nucleic Acids: structure and conformations
19. Nucleotide Metabolism: Synthesis and degradation of pyrimidine and purine nucleotides
20. Disorders of nucleotide metabolism

Recommended Books

1. Lehninger Principles of Biochemistry, 5th Ed., Nelson & Cox, Freeman, 2008
2. Harper's Illustrated Biochemistry, 27th Ed, Murray et.al. McGraw Hall 2006
3. Biochemistry, 3rd Ed., Zubay et.al, WCB 1993
4. Biochemistry, 5th Ed., Stryer et al, Freeman, 2002
5. Biochemistry, 3rd Ed., Voet & Voet, Wiley, 2004
6. Biochemistry and Molecular Biology, 2nd Ed., Elliot & Elliot, Oxford, 2004
7. Clinical Biochemistry, 6th Ed, Smith et al, Blackwell, 2004
8. Textbook of Medical Biochemistry, 6th Ed, Chatterjee & Shinde, Jaypee, 2005.
9. Text book of Clinical Biochemistry, Davlin
10. Biochemistry, Rawn, J. D.
11. Biochemistry, Mathews

Paper MHG-204. Developmental Genetics and Genetics of Human Development & Development Defects

Unit I

- 1 Early development
 - 1.1 Fertilization
 - 1.2 Types of cleavage
 - 1.3 Concept of determination, competence and differentiation
2. Gastrulation: Cell movement and formation of germ layers in frog, chick and mouse
3. Genetics of pattern formation: *Caenorhabditis*: Vulva formation
4. Pattern formation in Vertebrates:
 - 4.1. Axes formation and HOX genes
 - 4.2. Limb formation in chick

Unit II

5. Embryonic development in *Drosophila* I:
 - 5.1 Cleavage
 - 5.2 Gastrulation
 - 5.3 Anterior-posterior polarity
6. Embryonic development in *Drosophila* II:
 - 6.1 Maternal effect genes
 - 6.2 Segmentation genes
7. Embryonic development in *Drosophila* III:
 - 7.1 Homeotic selector genes: organization, functions and evolutionary significance.
 - 7.2 Generation of Dorsal-ventral polarity
8. Regeneration and Aging:
 - 8.1 Types of regeneration; Regeneration in Hydra, Salamander & liver regeneration
 - 8.2 Concept and theories of Ageing and Senescence; Age related disorders

Unit III Human Embryology & Developmental defects (Brief idea)

9. Human embryology I:
 - 9.1 Germ cell differentiation, Gametogenesis, fertilization, ovulation and implantation
 - 9.2 Brief account of embryonic development: Blastulation, Gastrulation, formation of notochord and establishment of body axis.
10. Human embryology II:
 - 10.1 Organogenesis: Formation of embryonic germ layers and their derivatives
 - 10.2 Fetal development and placentation (development, structure and function)
Fetal membranes in twins.
11. Molecular regulations of development and defects I:
 - 11.1 Human Hox genes and genetic defects due to mutation in Hox genes
 - 11.2 Somite differentiation and homeobox genes (anterior-posterior patterning)
 - 11.3 Limb development & limb defects
12. Molecular regulations of development and defect II:
 - 12.1 Eye development and eye defects
 - 12.2 Development of spinal cord and neural tube defects
 - 12.3 Brain development and cranial defects

Unit IV Human Embryology & Developmental defects (Brief idea)

13. Molecular regulations of development and defect III:
 - 13.1 Cardiac development and heart defects
 - 13.2 Kidney development and kidney defects
14. Molecular regulations of development and defect IV:
 - 14.1 Facial development and facial cleft defects
 - 14.2 Muscle development
15. Genetic defects due to abnormalities in developmental processes I:
 - 15.1 Abnormal implantation: contribution of maternal and paternal genes

- 15.2 Teratogenesis and tumors associated with gastrulation
- 15.3 Birth defects: erythroblastosis fetalis, fetal hydrops and twin defects
- 16. Genetic defects due to abnormalities in developmental processes -II
 - 16.1 Craniofacial and skeletal dysplasias
 - 16.2 Defects in sex differentiation
 - 16.3 Neural crest and craniofacial defects
 - 16.4 Vertebral defects: spina bifida and scoliosis

Unit V Human Developmental & Reproductive issues (General idea)

- 17. Environmental factors: Types and their effects on development
- 18. Reproductive failure:
 - 18.1 Infertility
 - 18.2 Abnormal gametes and infertility
- 19. Spontaneous abortions and still birth (etiology, pathogenesis, genetic characteristics, clinical notes, diagnosis and management)
- 20. Reproductive options: Assisted reproductive techniques (ARTs)

Recommended Books

1. Developmental Biology, 8th Ed., Gilbert, Sinauer, 2006
2. Principles of Developmental Genetics, Moody, Elsevier, 2007
3. Principles of Development, 2nd Ed., Wolpert, Oxford 2002
4. The Cellular & Molecular Biology of Pattern Formation, Stocum & Karr, 1990
5. Larsen's Human Embryology, 4th Ed., Churchill Livingstone, 2009
6. Langman's Medical Embryology, 10th Ed., Sadler, LMW, 2006
7. Human Embryology, 8th Ed., Singh & Pal, McMillan, 2007
8. Smith's Recognizable Patterns of Human Malformations, 6th Ed, Jones, Elsevier, 2006
9. Neural tube defects, Oppenheimer, Informa, 2007
10. Essential Medical Genetics, Conner & Ferguson-Smith, 5th Ed., Blackwell Science, 1997
11. Emery's Element of Medical Genetics, 11th Ed., Mueller & Young, Churchill Livingstone, 2003
12. Developmental Stages in Human Embryos O'Rahilly and Muller Carnegie 1987
13. Human Embryology Made Easy Rana Harwood 1998
14. Human Embryology and Teratology O'Rahilly and Muller Wiley 1992

Paper MHG-205: (Based on Theory Papers MHG-201 & MHG-202)
Molecular Genetics & Genomics, Molecular Structure & Functions of the Cell and Cancer Biology

1. Laboratory culture of bacterial (*E. coli*) cells
2. Plotting of growth curve for the determination of bacterial growth
3. Demonstration of bacterial transformation: Preparation of competent cells, transformation, and selection by antibiotics or α -complementation.
4. Gene induction in *Drosophila* (heat shock treatment)/Transgenic for hsp70-lacZ gene)
5. Study of endocytosis by trypan blue ingestion
6. Study of permanent slides of various tissue-types (e.g., epithelial, connective, blood, muscle, nervous, etc.); cancer cytogenetics, etc.
7. Electron micrographs & Photomicrographs related to cellular structures, etc.
8. Methods of histology & histochemistry for localization of biomolecules.
9. Experiments related to cell structure and function (Apoptosis, Signaling, cancer, etc.)

Scheme of Practical MHG-205

Maximum Marks: 85

1. Experiments on bacterial culture (Growth curve)/ transformation, selection of clones, etc.	15
2. Demonstration of endocytosis/ Heat stress response	10
3. Histological preparation and Histochemical staining to show biomolecules (10+10)	20
4. Spots (8): Molecular genetics,, cell biology, cancer, etc.	20
6. Viva voce	10
7. Practical record	10
Total	85

Paper MHG-206: (Based on Theory Papers MHG-203 & MHG-204)
Biochemistry and Metabolic Disorders, Developmental Genetics, Genetics of Human Development and Developmental Defects

1. Isolation of chromatin, its digestion by micrococcal nuclease and separation by agarose gel electrophoresis.
2. Extraction of Histones and analysis by SDS-PAGE
3. Buffers, pH, preparation of solutions
4. Spectrophotometric estimation of glucose, cholesterol and protein
5. Sugar estimations in normal and diabetic patients
6. Assay and kinetics of Alkaline phosphatase/Esterase
7. Study of expression of segmentation genes in *Drosophila*
8. Observation of homeotic mutants of *Drosophila*
9. Study of Frog development
10. Study of Chick embryo development (whole mounts & permanent slides)
11. Study of various stages of human foetal development (Observation of models)
12. Maintenance of mouse colony (observation of vaginal plug)

Scheme of Practical MHG-206

Maximum Marks: 85

1. Developmental Genetics: -Drosophila segmentation genes/ Homeotic mutations/Frog/Chick Embryology/ Human embryology	10
3. Buffer & pH measurement/ Biochemical estimations	10
4. Enzyme kinetics/ SDS-PAGE to separate polypeptides	15
5. Spotting (8): -Biochemical/ Physiological, Developmental Genetics, etc.	20
6. Seminar -	10
7. Viva voce -	10
8. Practical record -	10
Total -	85

Semester III

Paper MHG-301: Human Molecular Genetics and Human Genomics

Unit I

1. Genetic mapping of Mendelian and complex characters:
 - 1.1 Identifying recombinants and non-recombinants in pedigrees
 - 1.2 Genetic and physical map distances
 - 1.3 Genetic markers
2. Mapping of genetic traits:
 - 2.1 Two-point mapping- LOD score analysis
 - 2.2 Multipoint mapping
 - 2.3 Homozygosity mapping
3. Genetic mapping of complex traits; Difficulties in mapping
 - 3.1 Allele sharing methods- affected sib pair analysis
 - 3.2 Allelic association, Linkage disequilibrium mapping, Transmission disequilibrium test
4. Physical mapping of the human genome: Low resolution mapping- Cell hybrids, mini- and microcells, synteny of genes, Radiation hybrid mapping.

Unit II

5. Integration of cytogenetic, genetic and physical maps
6. Human genome mapping: Assembly of clone contigs and identifying genes in cloned DNA
7. History, HGP organization and goals of human genome project
8. The Genome projects:
 - 8.1 Mapping strategies, current status of various maps; DNA segment nomenclature
 - 8.2 ELSI
 - 8.3 Benefits & patenting of genetic materials

Unit III

9. Human genome diversity project (HGDP): Concept and goals
10. Bioethics: Definition, history, principles & Theories
11. Comparative genomics -Characteristics of genomes of human and other model organisms (yeast, *Caenorhabditis elegans*, *Drosophila*, *Fungus* and mouse)
12. Organization of human genome:
 - 12.1 Mitochondrial genome
 - 12.2 Nuclear genome -Gross base composition, gene density, CpG islands

Unit IV

13. Human genome structure:
 - 13.1 RNA-encoding genes, functionally identical/similar genes
 - 13.2 Diversity in size and organization of genes
 - 13.2 Pseudogenes
14. Functional genomics -ESTs, Transcriptosome, Proteome, Multiplex and DNA microarray (chip) based analysis
15. Gene families in human genome
 - 15.1 Multigene families -Classical gene families, families with large conserved domains, families with small conserved domains
 - 15.2 Gene super families
 - 15.3 Gene families in clusters
16. DNA testing
 - 16.1 Direct and indirect testing (gene tracking) in individuals
 - 16.2 DNA tests for identity and relationships including forensic applications
 - 16.3 Population screening- ethics, organization and advantages

Unit V

17. Identifying human disease genes
 - 17.1 Principles and strategies
 - 17.2 Position-independent and positional cloning
 - 17.3 Candidate gene approaches
 - 17.4 Confirming a candidate gene, mutation screening, testing in animal models
18. Molecular pathology
 - 18.1 Nomenclature of mutations and their databases
 - 18.2 Loss of function and gain of function mutations in diseases
19. Molecular pathology: Instability of the human genome and diseases- pathogenicity associated with repeated sequences:
 - 19.1 Slipped strand mispairing
 - 19.2 Unequal crossover and unequal sister chromatid exchange
 - 19.3 Gene conversion
 - 19.4 Retrotransposition
 - 19.5 Illegitimate recombination
20. Small RNAs:
 - 20.1 RNAi/si RNA: General idea and applications
 - 20.1 miRNA: General idea and applications

Recommended Book

1. Human Molecular Genetics 3, Strachen & Read, Blackwell, 2004
2. An Introduction to Molecular Human Genetics, Pasternak, Wiley, 2000
3. Human Genetics, Lewis, McGraw Hill, 2007
4. Molecular and Genetic Analysis of Human Traits, Maroni, Blackwell, 2001
5. Human Genetics, Gardner et al, Viva, 2008
6. Thompson & Thompson's Genetics in Medicine, 7th Ed, Nussbaum et al, Elsevier, 2007
7. Human Genetics and Genomics, 3rd Ed, Korf, Blackwell, 2007
8. Molecular Diagnosis, 2nd Ed., Coleman and Tsongalis, Humana Press, 2006
9. Current Topics in Human Genetics: Studies in Complex Diseases, Deng et al, World, 2007
10. Human Genetics: Problems and Approaches, 3rd Ed, Vogel, Springer, 1997

Paper MHG-302: Clinical Genetics and Genetic Counseling

Unit I

1. An overview of the genetic basis of syndromes and disorders
2. Monogenic diseases with well known molecular pathology
 - 2.1. Cystic fibrosis
 - 2.2. Tay-Sachs Syndrome
 - 2.3. Marfan syndrome
3. Inborn errors of metabolism and their genetic bases
 - 3.1 Phenylketonuria
 - 3.2 Mucopolysaccharidosis
 - 3.3 Galactosemia
4. Neurogenetic disorders
 - 4.1 Major regions of human brain and nerve conduction
 - 4.2 Charcot-Marie tooth syndrome. Spino-muscular atrophy
 - 4.3 Alzheimer's disease

Unit II

5. Syndromes due to triplet nucleotide expansion
6. Muscle genetic disorders
 - 6.1 Dystrophies (Duchenne Muscular dystrophy and Becker Muscular Dystrophy)
 - 6.2 Myotonias
 - 6.3 Myopathies
7. Genetic disorders of Haemopoitic systems
 - 7.1 Overview of Blood cell types and haemoglobin
 - 7.2 Sickle cell anemia
 - 7.3 Thalassemias
 - 7.4 Hemophilias
8. Genetic disorders of eye
 - 8.1 Colour Blindness
 - 8.2 Retinitis pigmentosa
 - 8.3 Glaucoma
 - 8.4 Cataracts

Unit III

9. Genetic disorders of skeleton
10. Genetic disorders of skin
11. Syndromes:
 - 11.1 Genomic syndromes: Neurofibromatosis I syndrome
 - 11.2 Genome imprinting: Prader-Willi and Angelman syndromes, Beckwith-Wiedeman syndrome
12. Cancers and cancer-prone syndromes
 - 12.1 Haematological malignancies
 - 12.2 Retinoblastoma, Wilm's tumour, Colorectal cancer
 - 12.3 DNA-repair deficiency syndromes
 - 12.4 Breast cancer

Unit IV

13. Complex polygenic syndromes
 - 13.1 Hyperlipidemia
 - 13.2 Atherosclerosis
 - 13.3 Diabetes mellitus
14. Mitochondrial syndromes
15. Management of genetic disorders
16. Historical overview (philosophy & ethos) and Components of genetic counseling I:
 - 16.1 Indications for and purpose
 - 16.2 Information gathering and construction of pedigrees

- 16.3 Medical Genetic evaluation
 - 16.3.1 Basic components of Medical History
 - 16.3.2 Past medical history, social & family history

Unit V

- 17. Components of genetic counseling II:
 - 17.1 Physical examination, General and dysmorphology examination
 - 17.2 Documentation, Legal and ethical considerations
- 18. Patterns of inheritance, risk assessment and counseling in common Mendelian and Multifactor syndromes
- 19. Genetic testing: biochemical & molecular tests
 - 19.1 In children
 - 19.2 Presymptomatic testing for late onset diseases (predictive medicine)
- 20. Prenatal and Preimplantation diagnosis 4
 - 20.1 Indications for prenatal diagnosis
 - 20.2 Indications for chromosomal testing
 - 20.3 Noninvasive methods
 - 20.4 Invasive methods

Recommended Books

1. Thompson & Thompson, Genetics in Medicine, 7th Ed., Nuusbaum et al, Elsevier, 2007
2. Emery & Remoin's Principles & Practice of Medical Genetics, Vol I-III, 5th Ed., Churchill Livingston, 2007
3. New Clinical Genetics, Read & Donnai, Scion, 2007
4. Emery's Element of Medical Genetics, 11th Ed., Mueller & Young, Churchill Livingstone, 2003
5. Genetics for Healthcare Professionals, Skirton & Patch, Bios, 2002
6. Medical Genetics at a Glance, Pritchard & Korf, Blackwell,2003
7. A Guide to Genetic Counseling, Baker et al, Wiley, 1998
8. Prenatal Medicine, Vugt & Shulman, Informa Healthcare, 2006
9. Smith's Recognizable Patterns of Human Malformations, 6th Ed, Jones, Elsevier, 2006
10. Neural tube defects, Oppenheimer, Informa, 2007
11. Essential Medical Genetics, Conner & Ferguson-Smith, 5th Ed., Blackwell Science, 1997
12. An Introduction to Molecular Human Genetics Pastemak Fritzgarald 2000
13. Genes in Medicine Rasko and Downes, Chapman & Hall (1996)
14. Introduction to Risk Calculation in Genetic Counselling, Young Oxford 1999
15. Human Molecular Genetics Strachen and Read Bio Sci. Publish. 2007
16. Color Atlas in Genetics Passarge Thieme 2001

Paper MHG-303: Recombinant DNA Technology and Molecular Diagnostics in Human Diseases

Unit I

1. Enzymes used in DNA technology
 - 1.1 Restriction and modification enzymes
 - 1.2 Other nucleases
 - 1.3 Polymerases
 - 1.4 Ligase, kinases and phosphatases ,
2. Cloning vectors
 - 2.1 Plasmids
 - 2.2 Phages
 - 2.3 Cosmids
 - 2.4 Artificial chromosomes
 - 2.5 Shuttle vectors
 - 2.6 Expression vectors
3. Cloning Techniques
 - 3.1 isolation & purification of genomic & plasmid DNA & RNA
 - 3.2 Gel electrophoresis of nucleic acids (RNA & DNA); Pulse field gel electrophoresis
 - 3.3 Construction of genomic libraries
 - 3.4 construction of cDNA libraries
4. Microcloning and Positional cloning: RFLP mapping, chromosome walking and jumping

Unit II

5. Screening of clones from libraries
 - 5.1 Expression based screening
 - 5.2 Interaction based screening: yeast two-hybrid system
 - 5.3 Preparation of probes
 - 5.4 Restriction mapping
- 6 Principles of hybridizations and hybridization based techniques:
 - 6.1 Colony, plaque, Southern, Northern and *in situ* hybridizations
 - 6.2 ELISA, western and southwestern blotting
 - 6.3 Microarray based detections
7. Characterization of clones
 - 7.1 DNA sequencing methods
 - 7.2 S1 nuclease and RNase mapping of nascent RNAs
8. Promoter characterization: promoter analysis through reporter genes, electrophoretic mobility shift assay, DNA foot-printing and linker scanning mutagenesis.

Unit III

9. Oligonucleotide synthesis
10. Principles & applications of Polymerase Chain Reaction (Types)
- 11 DNA fingerprinting
- 12 Mutagenesis
 - 12.1 Site directed mutagenesis
 - 12.2 Transposon mutagenesis
 - 12.3 Construction of knockout mutants

Unit IV

13. Gene transfer techniques
 - 13.1 Microinjection
 - 13.2 Transfection of cells: Principles and methods
14. Germ line transformation in *Drosophila*, transgenic and knock out mice: Strategies and methods
15. Applications of Recombinant DNA Technology
 - 15.1. Monitoring of gene expression in live cells
 - 15.2 Molecular genetic analysis of human diseases
 - 15.3 Biosafety & ethical considerations

- 16. Gene therapy & Stem cells:
 - 17.1 Somatic and germ line gene therapy
 - 17.2 DNA drugs and vaccines
 - 17.3 Stem Cells: Type, sources, culture and applications in therapy

Unit V Molecular Diagnostics (General ideas)

- 17. Testing DNA variation for diseases association
 - 17.1 SNPs; SNPs & Diseases
 - 17.2 Methods of SNP Typing: Brief idea of Traditional approach, Microchip (Affymetrix) & Taqman
- 18. Microarray approach to gene expression analysis (Brief idea)
 - 18.1 DNA microarray platforms
 - 18.2 cDNA array
 - 18.3 oligonucleotide arrays
 - 18.4 Concept of genome-wide association studies (GWAS)
 - 18.5 SAGE, CGH, Array CGH, SNP arrays
- 19. HLA Typing using molecular methods (Brief idea)
 - 19.1 PCR with sequence-specific primer
 - 19.2 Sequence-specific oligonucleotide probe hybridization
 - 19.3 Sequenced-based HLA typing
 - 19.4 Methods based on determination of conformation: SSC polymorphism, Heteroduplex analysis
- 20. Methods for analysis of DNA Methylation (Brief idea)
 - 20.1 Bisulphite modification
 - 20.2 Methylation-specific PCR
 - 20.3 Real time PCR methods
 - 20.4 Methylation-sensitive SSC analysis
 - 20.5 Profiling and arrays

Recommended Books

- 1. Recombinant DNA, 2nd Ed., Watson et al, Scientific American, 1998
- 2. Genes and Genome, Singer & Berg, USB, 1991
- 3. PCR, Hughes & Moody, Scion, 2007
- 4. Genomes 3, TA Brown, Garland, 2007
- 5. Gene Cloning & DNA Analysis: An Introduction, 5th Ed., Brown, Taylor & Francis, 2005
- 6. Principles of Gene Manipulation & Genomics, 7th Ed., Primrose & Twyman, Blackwell, 2006
- 7. Genetics: A Molecular Approach, 3rd Ed., Brown, Taylor & Francis, 2005
- 8. Molecular Cloning: A Laboratory Manual, 3rd Ed., Sambrook & Russell, CSH Press, 2001
- 9. Laboratory Manual, Human Molecular Biology, Suzycki, Blackwell, 2003
- 10. Current Protocols in Molecular Biology Ausubel et al Wiley. 1989
- 11. DNA Science Micklos and Freyer Cold Spring Harbor 1990

Paper MHG-304: Immunogenetics and Molecular Genetics of Human Pathogens

Unit I

1. General introduction to immune system
 - 1.1 Innate and adaptive immunity
 - 1.2 Immune responses
 - 1.3 Antigens & antibodies
 - 1.4 cells and organs of the immune system
2. Antigens & antibodies
 - 2.1 Immunogenicity vs antigenicity
 - 2.2 Factors influencing immunogenicity
 - 2.3 Structure and function of antibody: Ig G, Ig M, Ig A, Ig E & Ig D
 - 2.4 Antigen-antibody interactions
3. Immunoglobulin -I
 - 3.1 The immunoglobulin super family
 - 3.2 Organization of Ig genes
4. Immunoglobulin -II
 - 4.2 Expression of Ig genes
 - 4.3 Regulation of Ig gene transcription

Unit II

5. Generation of antibody diversity and Antibody engineering
6. T-Cell receptor
 - 6.1 The T-cell receptor
 - 6.2 Organization of TCR gene loci
 - 6.3 Generation of TCR diversity
7. Major Histocompatibility Complex molecules (MHC)
 - 7.1 General organization & inheritance
 - 7.2 MHC molecules & genes
 - 7.3 Regulation of MHC Expression
8. The HLA complex I:
 - 8.1 Organization of HLA complex
 - 8.2 Structure of class I and II HLA molecules

Unit III

9. The HLA Complex II:
 - 9.1 Expression of HLA genes
 - 9.2 HLA polymorphism
10. Antigen processing and presentation
11. Generation and regulation of immune responses
 - 11.1 Cytokines and activation of T & B cells
 - 11.2 Clonal selection
 - 11.3 Complement system
 - 11.4 Regulation of immune responses
 - 11.5 Immunological tolerance
12. Transplantation immunology: general idea of
 - 12.1 Allograft
 - 12.2 Xenograft
 - 12.3 Syngraft
 - 12.4 Graft versus host and host versus graft rejections.

Unit IV

13. Immune disorders – I
 - 13.1 HLA associated diseases
 - 13.2 Immunodeficiencies: HIV
 - 13.3 Auto immunity & auto immune disorders (e.g., RA/SLE/MS)
14. Immune disorders – II
 - 14.1 Hypersensitive reactions
 - 14.2 Cytokine-related diseases
 - 14.3 Role of MHC in disease susceptibility
15. Immune system in human health
 - 15.1 Immune response to infectious diseases and malignancy
 - 15.2 Concept of immunotherapy
 - 15.3 Vaccines
16. Hybridoma Technology: Production and applications of monoclonal and polyclonal antibodies

Unit V

17. Basics of Host-Pathogen interaction, evolution of pathogenicity and regulation of virulence;
Mechanism of drug resistance in pathogens: Viruses & Bacteria
18. Molecular pathology of following pathogens: HIV, Hepatitis virus *Mycobacterium tuberculosis*,
19. Molecular pathology of *Plasmodium*, *Leishmania*, & *Trypanosoma*
20. Development and application of molecular methods in diagnosis of infectious diseases and pathogen detection
 - 20.1 Immunoproteomics
 - 20.2 Immunoprecipitation, Immunoblotting and Immunofluorescence.
 - 20.3 Expression cloning
 - 20.4 PCR-based detection methods, etc.

Recommended Books

1. Cellular and Molecular Immunology, 6th Ed., Abbas et al, Elsevier, 2007
2. Immunology, 6th Ed Roitt, Mosby, 2002
3. Immunology, 5th Ed., Kuby, Freeman, 2002
4. Microbiology, 6th Ed., Prescott et al, McGraw Hill, 2005
5. Microbiology: A Human Perspective, 4th Ed., Nester et al, McGraw Hill, 2004
6. Medical Immunology, 6th Ed., Virrela, Informa Health Care, 2007
7. Immunology, Janeway & Travers, Garland Publishing Inc, 1994
8. Essential Immunology, Roitt Blackwell 1994
9. Immunology, Roitt et al Mosloy 1993
10. Immunology -A Short Course, Benjamini Wiley-Liss 2000
11. Text Book of Immunology, Barrett Mosloy 1988
12. Biology of Microorganisms, Madigen et al Prentice Hall 1997
13. Introductory Microbiology, Heritage et al Cambridge Univ. 1996
14. Microbiology, Pel czar et al Tata 1993
15. Molecular Diagnosis of Infectious Diseases, Reischel Humana 1998
16. Fundamentals of Immunology, William Paul, Freeman

Paper MHG-305: (Based on Theory Papers MHG-301 & MHG-303)
Human Molecular Genetics and Human Genomics and Clinical Genetics and Genetic Counseling

1. Extraction of DNA from human lymphocytes & agarose gel
2. PCR-based detection of allelic inheritance of a DNA marker
3. Micrographs demonstrating examples of molecular methods, etc.
4. Study of sex chromatin from mammalian buccal epithelium, hair root cells and lymphocytes.
5. Lymphocyte culture and chromosome preparations
6. Chromosome banding, karyotyping and making idiogram of the banded chromosomes
7. Detection of chromosome anomalies in cancer tissues
8. Molecular detection of genetic diseases
9. Preparation of pedigree charts for common genetic disorders
10. Risk assessment (Binomial probability and Bayesian calculation)
11. Calculation of allelic identity by descent (IBD) in inbred families

SCHEME:

Time: 5 hrs MM: 85

1. Isolation of DNA from blood lymphocytes/tissue & agarose gel electrophoresis	10
2. DNA Polymorphism study by PCR-based methods	15
3. Pedigree construction & Risk assessment in pedigree	10
4. Preparation of sex chromatin / G- or C-banding of mammalian chromosomes	10
5. Spots (8): (Human molecular and clinical genetics related)	20
6. Viva voce	10
7. Practical Record	10
Total	85

**Completion of a short in-house Research Project with the faculty involved as part of the practical papers. Student shall also interact with medical specialists/ Doctors at the local medical centers/hospitals in order to collect clinical information on various genetic disorders through direct observation and personal interview with the patients. Students shall submit the project report and deliver seminar for evaluation.*

Practical Paper MHG-306: (Based on Theory Papers MHG-303 & MHG-304)
Recombinant DNA Technology, Diagnostic Technologies, Immunogenetics and Molecular Genetics of Human Pathogens

1. Isolation of plasmid DNA
2. Quantitation of DNA on gel and by spectrophotometer
3. Restriction mapping of plasmid DNA
4. Southern Hybridization
5. In situ hybridization
6. Genomic DNA extraction from mouse/human/fly
7. Western blotting
8. PCR
9. Precipitation and agglutination reactions
10. Study of cell types of immune system
11. Immunodiffusion
12. Antibody titration
13. Enzyme linked immuno-absorbent assay (ELISA)
14. Blood grouping & Rh factor determination
15. Immuno-localization of antigens

SCHEME:

Time: 5 hrs MM: 85

1. Molecular genetics: Plasmid/genomic DNA isolation and agarose gel electrophoresis/Restriction digestion of DNA and separation on gel	12 ^{1/2}
2. Blood group & Rh detection/ Immune cells/PCR based diagnosis (RFLP)/ Hybridization methods	12 ^{1/2}
3. Immunodiffusion/ Immunoelectrophoresis/ Immunodetection	10
4. Spot (8): Recombinant DNA & Immunogenetics related	20
5. Seminar	10
6. Viva voce	10
7. Practical Record	10
Total	85

Semester IV

Paper MHG-401: Population Genetics, Human Evolutionary Genetics and Behavior Genetics

Unit I

1. Concept and theories of evolution
2. Microevolution in Mendelian population
 - 2.1 Mendelian Population
 - 2.2 Allele frequencies and genotype frequencies
 - 2.3 Hardy-Weinberg equilibrium and conditions for its maintenance
3. Elemental forces of evolution
 - 3.1 Mutation
 - 3.2 Selection
 - 3.3 Genetic drift
 - 3.4 Migration
4. Isolating mechanisms: Geographic and reproductive isolation

Unit II

5. Genetic variability in natural population I:
 - 5.1 Chromosomal polymorphism
 - 5.2 Enzyme polymorphism
 - 5.3 DNA polymorphism
6. Genetic variability in natural population II:
 - 6.1 Adaptive genetic polymorphism
 - 6.2 Balanced polymorphism
 - 6.3 Linkage disequilibrium
7. Concept of species and modes of speciation: sympatry, allopatry, stasipatry & parapatry
8. Nonrandom and random breeding
 - 8.1 Inbreeding and assortative mating
 - 8.2 Heterosis

Unit III

9. Molecular population genetics
 - 9.1 Molecular evolution (neutral theory, punctuated equilibrium)
 - 9.2 Molecular clock
10. Molecular evolution and Phylogenetic tree
 - 10.1 Construction of phylogenetic tree using nucleotide sequence data
 - 10.2 Amino acid sequence and phylogeny (globin gene, cytochrome b gene, etc.)
11. DNA-based phylogeny
 - 11.1 DNA-DNA hybridization
 - 11.2 Restriction enzyme sites
 - 11.3 Nucleotide sequence comparison and homologies
12. Concepts of Eugeics, Euthenics & Euphenics

Unit IV Human Evolution

13. Human phylogeny
 - 13.1 Hominid evolution. anatomical, Geographical, Cultural
 - 13.2 Molecular phylogenetics of *Homo sapiens*
14. Peopling of continents (Europe, Africa, Asia)
15. Admixture:
 - 15.1 Meeting of human populations & its genetic imprint
 - 15.2 Detection of admixture (based on allele frequencies & DNA data)
 - 15.3 Y Chromosome & mitochondrial DNA markers in genealogical studies

- 16. Culture and human evolution
 - 16.1 Learning, society and culture
 - 16.2 Relative rates of cultural and biological evolution
 - 16.3 Social Darwinism
 - 16.4 Sociobiology

Unit IV Behaviour Genetics

- 17. Nature-nurture and behaviour
 - 17.1 Genetic experiments to investigate animal and human behaviour
 - 17.2 Identifying genes for behavior (induced mutations, QTL, synteny homology)
 - 17.3 Environmental influence- shared and non-shared environment
 - 17.4 Investigating genetics of human behaviour (twin & adoption study designs, Interpreting heritability, linkage and association studies)
- 18. Psychopathology
 - 17.1 Signs and symptoms
 - 17.2 Schizophrenia
 - 17.3 Mood disorders
 - 17.4 Anxiety disorders
 - 17.5 Disorders of childhood
 - 17.6 Personality and personality disorders- antisocial personality, criminal behaviour
- 19. Cognitive abilities and Disabilities
 - 19.1 Mental retardation
 - 19.2 Learning disorders
 - 19.3 Communication disorders
- 20. Neurogenetics:
 - 20.1 Study design: Genetic and environmental manipulations
 - 20.2 Circadian rhythms
 - 20.3 Learning & memory

Recommended Books

1. Evolution, 4th Ed., Strickberger, Jones and Barlett, 2008
2. Human Evolution, 5th Ed, Roger Lewin, Blackwell, 2005
3. Evolutionary Analysis, 4th Ed, Freeman & Herron, Pearson, 2007
4. Genetics and the Origin of Species, Dobzhansky, Oxford, 1976.
5. Organismic Evolution, Verne Grant, Freeman, 1977
6. Behavioral Genetics, 4th Ed., Plomin et al, Worth, 2001
7. Genetics: Analysis of Gene and Genomes, 6th Ed., Hartl & Jones, Jones and Bartlett, 2005
8. Neurogenetics of Psychiatric Disorders, Sawa & McInnes, Informa Healthcare, 2007
9. Synopsis of Psychiatry, 9th Ed, Kaplan & Sadock, LMW, 2003
10. Genetics of Population, 2nd Ed., Heidrick, Jones and Bartlett, 2000
11. Human Evolutionary Genetics, 1st Ed., Jobling and Smith, Garland, 2004.

Paper MHG402. Biostatistics & Bioinformatics

Unit I

1. Biostatistics:
 - 1.1 Overview
 - 1.2 Measures of central tendency: Mean, Median, Mode
 - 1.3 Measures of dispersion: Standard deviation, standard error, Variance, Coefficient of variation
2. Tabulation and presentation of data
3. Hypothesis testing and Test of Hypothesis:
 - 3.1 Statistical & Scientific hypothesis
 - 3.2 The null and alternative hypothesis
 - 3.3 Procedure (steps) for Hypothesis testing
 - 3.4 F-tests & Chi square test
4. Test of significance: Student's t-test

Unit II

5. General idea of Correlation and Regression Analysis
6. ANOVA: General idea of one way & two way analysis
7. Nonparametric methods for statistical inference
 - 7.1 Wilcoxon Rank-sum test
 - 7.2 Mann-Whitney test
8. Sampling: Types and methods of sampling

Unit III

9. Computers and computer languages:
 - 9.1 Introduction to computers,
 - 9.2 Components of a computer system (CPU, I / O units
 - 9.3 Computer languages: Introductory concepts of computer languages;
10. Introduction to operating systems, Linux/ UNIX
11. Elementary idea on the applications of common DOS- and WINDOWS based software packages
12. Computer Networking:
 - 12.1 Introduction to: E-mail and Internet, WWW, HTTP, HTML, URLs, Websites, Web Pages, Web browser;
 - 12.2 Applications in library search & information retrieval

Unit IV

13. Bioinformatics: Introduction and scope of Bioinformatics: An Overview
14. Biological databases I:
 - 14.1 Introduction
 - 14.2 Types of databases (Entrez, SRS or Sequence Retrieval System, PIR or Protein Identification Resource, GENE BANK, SWISS-PROT and other databases, etc.).
15. Biological databases II:
 - 15.1 Nucleotide and protein sequence databases,
 - 15.2 Major Bioinformatics databases,
16. Sequence analysis (Brief idea):
 - 16.1 Methods for alignment (dot matrices)
 - 16.2 Tools for sequence alignment – Fasta, BLAST, PSI-Blast,
 - 16.3 Multiple Sequence Alignment (MSA) - tools and applications.

Unit V

17. Bioinformatics tools for phylogenetic analysis
18. Proteomics (Brief ideas):
 - 18.1 Proteome & Tools for proteome analysis,
 - 18.2 protein function prediction,
19. Gene prediction (Brief ideas): Methods & gene prediction tools,

20. Elementary ideas on the analysis of genetic data: PHYLIP, haplotype analysis (Hep cervus), Linkage analysis,

Recommended Books

1. Current Topics in Computational Molecular Biology, Jiang et al, Anne Books, 2004
2. Bioinformatics, Brown, Eaton, 2000
3. Bioinformatics for Geneticists, 2nd Ed., Barnes, Wiley, 2007
4. Genome Mapping and Sequencing, Dunham, Horizon, 2003
5. Microarray Technology Through Applications, Falciani, Taylor and Francis, 2007
6. Bioinformatics Computing, Bergeron, Pearson, 2003
7. Introduction to Bioinformatics, Lesk, Oxford, 2002
8. Bioinformatics: Methods and Protocols, Misener & Krawetz, Human Press, 2004
9. Internet and e-mail, 2nd Ed., Bangia, Khanna, 2002
10. Proteomics, O'Connor & Hames, Scion, 2008
11. Proteomics in Practice, 2nd Ed., Westermeir et al, Wiley, 2008
12. DNA Microarrays, Schena, Scion, 2008
13. Bioinformatics-Sequence and Genome Analysis, Mount, CBS, 2003
14. A primer of Genome Science. G.Gibson & SV Muse, Sinauer Associates, Inc.Publishers, 2002.
15. Fundamental Concepts of Bioinformatics. Krane and Raymer, Pearson Education, 2003
16. Instant Notes: Bioinformatics. Westhead, Parish and Twyman, Viva Book Pvt. Ltd., 2003
17. Introduction to Bioinformatics. Attwood and Parry-Smith, Pearson Education, 2003
18. Mathematical & Statistical Methods for Genetic Analysis, 2nd Ed., Lange, Springer, 2004
19. Fundamentals of Biostatistics, 2nd Ed., Khan & Khanum, Ukaaz, 2004
20. Choosing and Using Biostatistics, 2nd Ed., Calvin Dytham, Blackwell, 2006
21. The Analysis of Biological Data, Whitlock & Schutler, Robert & Co., 2009
22. Biostatistics, Dr. Rajeev Goswami, MD, 2009
23. Methods in Biostatistics, 6th Ed., B. K. Mahajan, Jaypee, 2004

MHG 403: Population Genetics, Human Evolutionary Genetics and Behavior Genetics, Biostatistics and Bioinformatics

1. Microsatellite/RAPD/mitochondrial marker-based detection of molecular polymorphism in populations.
2. Study of Models/Photographs on molecular genetics & evolutionary principles
3. Study of courtship behaviour in *Drosophila*
4. Personality analysis (through questionnaires)
5. IQ measurement
6. Schizophrenia & other disorders & visit to mental hospital
7. Hardy-Weinberg Genetic equilibrium: PTC Tasters & non-tasters
8. Hardy-Weinberg Genetic equilibrium and Enzyme polymorphism: Calculation of gene & genotype frequencies
9. Experiments on biostatistics problems: Mean, median, mode, T-test, Chi square test, correlation test, etc.
10. Computer handling, familiarization with window based software (e.g., MSWORD,
11. MS-EXEL, MS-POWERPOINT, PHOTOSHOP, ACROBAT etc.)
12. Use of Internet: Pubmed, Entrez, EMBL databases for literature search and for comparison of protein and DNA sequences.
13. Applications of BLAST, FASTA, CLUSTALW, GENSCAN, RASMOL, Phylogendron.

Scheme of the Practical Maximum Marks: 85

1. Experiments on population genetics/ evolutionary principles	10
3. Experiment on behavior	10
4. Experiment on biostatistics problem	10
5. Bioinformatics/Computer application	15
6. Spots (8)	20
7. Viva Voce	10
8. Practical Record	10
Total	85

MHG 403: Dissertation (Training) work

Dissertation work -	200
Viva-	100
Total	300