JIWAJI UNIVERSITY, GWALIOR
M.Sc. Molecular and Human Genetics
(2022-2024)

CENTRE FOR GENOMICS
JIWAJI UNIVERSITY

DISTRIBUTION OF DIFFERENT PAPERS AND CREDITS IN VARIOUS SEMESTERS

Semester	Paper Code	Name of the Paper	Type of Paper	Credits
Semester I	**MHG-101**	A. Principles of Genetic Inheritance B. Statistical Tests in Genetic Analysis	**Core**	**3**
	MHG-102	Basic Human Genetics and Human Cytogenetics	Core	3
	MHG-103	Molecular Structure & Functions of the Cell	Core	3
	MHG-104	A. Molecular Organization of Chromatin and Cytogenetics B. Cancer Biology	Core	3
	MHG-105	Practical based on papers 101 & 102	Core	3
	MHG-106	Practical based on papers103 & 104	Core	3
	MHG-107	Seminar	Core	1
	MHG-108	Assignment	Core	1
	MHG-109	Comprehensive Viva Voce	Virtual	4
		Total Credits		**24**
Semester II	MHG-201	Molecular Genetics and Genomics	Core	3
	MHG-202	Human Molecular Genetics and Human Genomics	Core	3
	MHG-203	Immunogenetics	Core	3
	MHG-204	Biochemistry: Structure, Function and Regulation of Biomolecules	Core	3
	MHG-205	Practical based on papers 201 & 202	Core	3
	MHG-206	Practical based on papers 203 & 204	Core	3
	MHG-207	Seminar	Core	1
	MHG-208	Assignment	Core	1
	MHG-209	Comprehensive Viva Voce	Virtual	4
		Total Credits		**24**
Semester III	MHG-301	Developmental and Reproductive Genetics	Core	3
	MHG-302	Clinical Genetics and Genetic Counseling	Core	3
	MHG-303	Population Genetics, Human Evolutionary and Behavior Genetics	Elective: Centric	3
	MHG-304	A. Recombinant DNA Technology B. Molecular Diagnostic Methods	Elective: Generic/Centric	3
	MHG-305	Practical based on papers 301 & 302	Core	3
	MHG-306	Practical based on papers 303 & 304	Elective: Generic/Centric	3
	MHG-307	Seminar	Core	1
	MHG-308	Assignment	Core	1
	MHG-309	Comprehensive Viva Voce	Virtual	4
		Total Credits		**24**
Semester IV	MHG-401	Bio-informatics and Bio-techniques	Core	3
	MHG-402	Practical based on papers 401	Core	3
	MHG-403	Seminar	Core	1
	MHG-404	Assignment	Core	1
	MHG-405	Dissertation Work	Core	12
	MHG-406	Comprehensive Viva Voce	Virtual	4
		Total Credits		**24**
Minimum Number of Credits to be earned for the award of degree (Valid:80 + Virtual: 16)				**96**

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The course for Master of Science (M. Sc.) in Molecular & Human Genetics shall comprise of four semesters of six months duration each. Each theory and practical paper will be of 3 credits. The first 3 semesters shall include 4 theory papers and 2 practical courses, while the 4th semester, will include 1 theory paper and 1 practical paper. The total marks for assessment in these papers are 100 marks, out of which 60 marks are for final examinations and 40 marks for internal assessments. All the theory papers are divided into 40 classes of 1 hr. After the completion of each topic in a particular paper, there will be a test and the maximum marks will be equivalent to the number of hours allotted to that topic. These marks will comprise the internal assessment marks (40) for each paper. The students will participate in weekly seminars (on any topic from the syllabus allotted to them by the faculty) and journal clubs (seminar on a research paper of interest), to meet the needs in their aim to become an interdisciplinary researcher. For this they will be awarded 1 credit each. Also a comprehensive viva voce examination will be held during the practical exams (4 credits).

In the last semester, the students shall formulate a short project proposal (dream project designed by the students themselves) in the subject related to the course under the supervision of the faculty involved and submit the proposal along with presentation for evaluation (2 credits). In addition, the students are required to undergo a 3 month dissertation work, to obtain professional exposure in well reputed Research Institutes/Universities or Industries and submit the final report along with a presentation for evaluation in the 4th semester (12 credits).

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

Semester I

Paper MHG-101: A. Principles of Genetic Inheritance
B. Statistical tests in genetic analysis

(No. of classes of 60 mins each)

A. Principles of Genetic Inheritance

Unit I

1. Mendel's laws of inheritance — 2
   - 1.1 Law of segregation
   - 1.2 Law of independent assortment
2. Chromosomal theory of inheritance — 1
3. Extensions of Mendelism — 5
   - 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 — 2

Unit II

5. Sex chromosomes and sex-linked inheritance — 2
   - 5.1 Sex chromosomes and their meiotic behaviour
   - 5.2 Sex-linked inheritance in *Drosophila* and *human*
6. Linkage and crossing over — 4
   - 6.1 Concept
   - 6.2 Cytological demonstration of crossing Over in *Drosophila (Genetic* and cytological crossing over)
   - 6.3 Genetic distance and physical distance
7. Linkage and crossing over: Preparation of Linkage map — 3
   - 7.1 Genetic recombination & construction of genetic maps in *Drosophila* (3-point test Cross) & yeast (Tetrad analysis).
   - 7.2 Interference and coincidence
   - 7.3 Mitotic recombination
8. Inheritance of quantitative traits — 4
   - 8.1 Continuous and discontinuous variation
   - 8.2 Genetic variance and heritability. (Narrow sense and broad sense); Quantitative trait loci (QTL)

Unit III

9. Polygenic inheritance, Environmental effects on gene expression — 2
10. Extranuclear inheritance & maternal effects — 3
    - 10.1 Organelle heredity (mitochondria & chloroplast); Petite mutations
    - 10.2 Infectious heredity (Cytoplasmic inheritance) in symbionts (*Paramecium*) &*Drosophila*
    - 10.3 Maternal inheritance: Ephestia pigmentation and snail coiling

B. Statistical tests in genetic analysis

Unit IV

11. Application of laws of probability (product rule, sum rule. binomial probability) — 1
12. Measures of central tendency: Mean, Median, Mode — 1
13. Measures of dispersion: Standard deviation, standard error, Variance, Coefficient of variation — 2
14. Hypothesis testing and analysis of Genetic data — 4
    - 14.1 Statistical & Scientific hypothesis
    - 14.2 The null and alternative hypothesis
    - 14.3 F-tests & Chi square test, Student's t test, Z test, Q test
15. General idea of Correlation and Regression Analysis — 2
16. ANOVA: General idea of one way & two way analysis — 2

Paper MHG-102: Basic Human Genetics and Human Cytogenetics

(No. of classes of 60 mins each)

Unit I

1. History of Human Genetics 1
2. Pedigrees- gathering family history. pedigree symbols, construction of pedigrees; presentation of molecular genetic data in pedigrees 1
3. Pedigree analysis of monogenic traits: 2
   - 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 pattems I: Non-penetrance, variable expressivity, pleiotropy, onset, dominance problem; anticipation, compound heterozygosity 2
5. Complications to the basic pedigree pattems II: Genomic imprinting and uniparentaldisomy, spontaneous mutations, mosaicism and chimerism, male lethality, X- inactivation, Consanguinity and its effects in the pedigree pattern, allele frequency in population. 2

Unit II

6. Complex traits- polygenic and multifactorial 6
   - 6.1 Approaches to analysis of complex traits- 'Nature vs nurture', role of family and shared 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 2
8. Estimation of genetic components of multifactorial traits: emperic risk, heritability, coefficient of relationship, application of Baye's theorem 2
9. Pharmacogenomics: 3
   - 9.1 Concept
   - 9.2 Polymorphism relating to drug metabolism and disposition
   - 9.3 Polymorphism affecting drug targets

Unit III

10. Human Cytogenetics 3
    - 10.1 Origins and developments in the study of human cytogenetics
    - 10.2 Chromosome banding: Principle, methods and application (G, C, Q, R, T and NOR banding)
    - 10.3 Molecular techniques in human chromosome analysis (FISH, GISH, CGH, SKY), In situ Restriction enzyme banding (Chromosomal hybridization)
11. Human chromosomal pathologies: 2
    - 11.1 Numerical aberrations and their common syndromes
    - 11.2 Structural aberrations and their common syndromes (translocations, duplications, deletions, micro deletion syndromes, fragile sites, etc.)
12. Human cytogenetics: Karyotype and Nomenclature 2
    - 12.1 Human karyotype: banding patterns, ideogram, nomenclature of banding
    - 12.2 Nomenclature of aberrant karyotypes

Unit IV

13. Tissue culture methods: Lab preparation, sterilization, culture media, sera & growth factors, 3
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. 4
15. Stem cells: Origin, culture, properties and therapeutic applications 2
16. General idea of Pharmacogenetics, Ecogenetics, Teratogenetics and Biochemical genetics (Blood groups & Serology; Protein polymorphism & its significance 2
17. Conventions of nomenclature of genes and gene products in different model systems (Bacteria, Viruses, Yeast, mouse and human: HGNC recommendations). 1

Paper MHG-103: Cell Biology: Molecular Structure & Functions of the Cell

(No. of classes of 60 mins each)

Unit I

1. Plasma Membrane: 3
   - 1.1 Molecular organization
   - 1.2 Transport across membrane
2. Mechanisms of Endocytosis and Exocytosis. 1
3. Endomembrane system: Ultrastructure of EPR & transport through EPR 2
4. Endomembrane system: Ultrastructural organization of Golgi complex & Transport through GC. 2
5. Mitochondria: 2
   - 5.1 Ultrastructure
   - 5.2 Mitochondrial transport
   - 5.3 Chemiosmotic theory and respiratory chain complexes

Unit II

6. Ultrastructure of nucleus & nucleolus 1
7. Mechanisms of intracellular digestion: Structure & functions of Lysosomes. 1
8. Structure and functions of Peroxisomes 1
9. Structure and biosynthesis of Ribosomes 1
10. Signaling 4
    - 10.1 Intracellular receptor and cell surface receptors
    - 10.2 Signaling via G-protein linked receptors (PKA, PKC, CaM kinase)
    - 10.3 Enzyme linked receptor signaling (Growth factor receptor signaling; JACK-STAT pathway)
    - 10.4 Network and cross-talk between different signal mechanisms
    - 10.5 Role of NO & CO in cell signaling.

Unit III

11. Cytoskeletons: 3
    - 11.1 Microfilaments: Structural organization. Cell motility and cell shape
    - 11.2 Intermediate filaments
12. Microtubule: Ultra structure and functional organization 2
13. Structure and functions of cilia, flagella, and centriole 1
14. Cell cycle and its regulation 4
    - 14.1 Overview of the Cell ycle
    - 14.2 Cell cycle control system
    - 14.3 Control of cell division and cell growth

Unit IV

15. Mitotic Cell Division: Molecular mechanisms 3
    - 15.1 Mitotic spindle and arrangement of chromosomes on equator
    - 15.2 Regulation of exit from metaphase
    - 15.3 Chromosome movement at anaphase
16. Meiotic Cell division 2
    - 16.1 Overview of the process
    - 16.2 Meiosis specific cellular changes: Molecular & Biochemical
    - 16.3 Genetic consequences of meiosis
17. Programmed cell death and Senescence: 4
    - 17.1 Definition and General Characteristics; Necrosis & PCD
    - 17.2 Morphological and Biochemical changes
    - 17.3 Molecular pathways of PCD
    - 17.4 Inhibitors of PCD and survival factors
18. Cell-Cell Interaction 3
    - 18.1. Cell adhesions
    - 18.2. Cell junctions (Occluding, Anchoring & Gap junctions)
    - 18.3. Extracellular matrix: Organization & Functions; Integrins

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 at 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-104: A. Molecular Organization of Chromatin and Cytogenetics
B. Cancer Genetics

A. Molecular Organization of Chromatin and Cytogenetics

(No. of classes of 60 mins each)

Unit I

1. Prokaryotic and eukaryotic chromosome — 1
2. Chromatin Structure — 3
   - 2.1. Chemical constituents: histones & DNA
   - 2.2. Nucleosome and higher order organization
   - 2.3 Chromatin remodelling
3. Chromatin Organization — 2
   - 3.1 Metaphase chromosome: centromere and kinetochore, telomere and its maintenance
   - 3.2 Chromosomal domains (matrix, loop domain) and their functional significance
4. Structural and functional organization of interphase nucleus — 1
5. Functional states of chromatin and alterations in chromatin organization: DNAse I hypersensitivity — 1
6. Giant chromosomes: Structural and functional characteristics of Polytene and lampbrush chromosomes — 2

Unit II

7. Heterochromatin and euchromatin; position effect variegation. — 1
8. Chromosomal abnormalities — 3
   - 8.1 Types
   - 8.2 Meiosis in inversion and translocation heterozygotes; breakage-fusion-bridge cycles
   - 8.3 Cell cycle specific chromosomal aberrations in somatic chromosomes
   - 8.4 Sister chromatid exchanges and somatic crossing over
9. Dosage compensation in mammals — 1
   - 9.1 Lyon's Hypothesis
   - 9.2 Sex chromatin
10. Molecular mechanism of X-chromosome inactivation — 2
11. Dosage compensation in *Drosophila* — 1
12. General idea of dosage compensation in nematode, *C. elegans.* — 1

B. Cancer Genetics

Unit III

13. Mutagenesis & Mutation — 3
    - 13.1 Types & origin
    - 13.2 Mechanisms
    - 13.3 Detection and isolation
14. DNA damage and repair mechanisms — 2
15. Chromosomal Instability and DNA damage response — 2
16. Cancer Biology — 3
    - 16.1 Cancer & environment
    - 16.2 Biochemical & structural Changes in cancer cells
    - 16.3 Tumor progression: angiogenesis & metastasis

Unit IV

17. General idea of Oncogenes and Tumor suppressor genes — 1
18. Molecular mechanisms of tumorogenesis: — 2
    - 18.1 Cell cycle check-point defects
    - 18.2 Tumor specific markers
19. Chromosomal basis of Cancer — 3
    - 19.1Philadelphia chromosome, Retinoblastoma, Burkitt's lymphoma
    - 19.2 Oncogene amplification (HSR & DM)
    - 19.3 Aneuploidy in neoplasia
20. Epigenetic Mechanisms: Methylation, Acetylation, Histone modification — 3
21. Epigenetics and Cancer — 2
    - 21.1 Epigenetic inheritance and gene expression
    - 21.2 Epigenetic regulation in cancer

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
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.Press1988

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

Principles of Genetic Inheritance, Statistical tests in genetic analysis, Basic Human Genetics & Human Cytogenetics

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. Experiments on biostatistics problems: Mean, median, mode, T-test, Chi square test, correlation test, etc.
9. Preparation of pedigree charts for common phenotypic characters of Human
11. Tissue culture: Lymphocyte culture and chromosome preparations
12. C, G and Fluorescence banding

Scheme of paper 105:
Total marks: 60

1. Analysis of Mono hybrid / Dihybrid / Sex-linkage crosses	7
2. Linkage & Crossing over / Three Point Test cross & Gene mapping	7
3. Karyotyping and Idiogram preparation/ G- or C-banding of mammalian chromosomes	7
4. Lymphocyte culture and chromosome preparations	7
5. Spotting (8): (*Spots based on genetic inheritance, Basic Human Genetics, Human Cytogenetics*)	16
6. Viva Voce (Experiment related)	10
7. Practical record	6
Total	**60**

Paper MHG-106: (Based on Theory Papers MHG-103 & MHG-104)
Molecular Structure & Functions of the Cell, Molecular Organization of Chromatin and Cytogenetics
Cancer Genetics

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 cells
3. Study of Meiosis in grasshopper testis
4. Study of meiosis from super ovulated oocytes of female and male testis mice/rat
5. Study of polytene chromosomes in *Drosophila / Chironomous* larval salivary glands
6. Study of endocytosis by trypan blue ingestion
8. Study of permanent slides of various tissue-types (e.g., epithelial, connective, blood, muscle, nervous, etc.)
9. Electron micrographs & Photomicrographs related to cellular structures, etc.
10. Methods of histology & histochemistry for localization of biomolecules.
11. Detection of chromosome anomalies in blood cancers.
12. Experiments related to cell structure and function (Apoptosis, Signaling, cancer, etc.)

Scheme for Paper 106:

1. Metaphase plate preparation from rat bone marrow	7
2. Study of mitosis/meosis from onion root tips/grasshopper testis	7
3. Histological preparation and histochemical staining to show biomolecules	7
4. Endocytosis/Study of polytene chromosome	7
5. Spotting (8): (*Cell Biology, Chromosome Organization, Cancer Biology*).	16
6. Viva Voce (Experiment related)	10
7. Practical Record	6
Total	**60**

Paper MHG-107: Seminar (From Syllabus) Credit: **1**
Paper MHG-108: Research Paper Presentation Credit: **1**
Paper MHG-109: Comprehensive Viva Voce Virtual Credit: **4**
(2 credit each for 105 and 106; to be conducted by an external examiner on the day of practical exam)

Semester II

Paper MHG-201: Molecular Genetics & Genomics

(No. of classes of 60 mins each)

Unit I

1. Properties & evolution of genetic material, Flow of genetic information — 1
2. Organization of viral and bacterial genomes — 1
3. Eukaryotic genome — 3
   - 3.1. Repetitive DNA
   - 3.2. General concept of a gene
   - 3.3 Non-coding genes
   - 3.4 DNA Renaturation & Denaturation and Complexity of genome; Cot value
4. Replication: Prokaryotic and Eukaryotic — 3
   - 4.1 DNA Polymerases
   - 4.2 Replicons, origin & termination
   - 4.3 Replisomes; Genes controlling replication

Unit II

5. Recombination — 3
   - 5.1 Homologous recombination
   - 5.2. Gene conversion
   - 5.3 Site-specific recombination
6. Transcription in Prokaryotes: Prokaryotic RNA polymerase, sigma factors, initiation and termination. — 2
7. Eukaryotic RNA polymerases and their promoters; Transcription units. — 2
8. Transcription activators and repressors: Identification of transcription factors, DNA binding motifs, reporter assay & repressors — 2

Unit III

9. Processing of transcripts: — 2
   - 9.1 5' capping.
   - 9.2 3' Polyadenylation
   - 9.3 Splicing
   - 9.4 RNA editing
10. Post-transcriptional regulation — 2
    - 10.1. Alternative splicing
    - 10.2. Transport and targeting of RNA
    - 10.3. Post-transcriptional gene silencing
11. Regulation of gene expression — 4
    - 11.1.Concept of Operons (lac and trp as examples) and regulon
    - 11.2. Positive and negative regulation
    - 11.3. Enhancers and promoters
    - 11.4 Regulation by attenuation and anti-termination
12. Mechanisms of steroid hormone & stress induced gene expression — 2

Unit IV

13. Gene mapping in bacteria — 2
    - 13.1. Transformation
    - 13.2. Conjugation
    - 13.3. Transduction
14. Recombination, deletion and complementation mapping in T4 phage (rII locus) — 2
    - 14.1 Intragenic recombination in Bacteriophage
    - 14.2 Deletion & complementation mapping in T4 phage
15. Translation — 3
    - 15.1 General mechanism
    - 15.2 Role of rRNA & tRNA in translation
    - 15.3 Translational control of mRNA and targeting of proteins
16. Transposable Genetic Elements — 3
    - 16.1Mechanism of transposition in prokaryotes
    - 16.2 Transposable genetic elements in: Yeast, *Drosophila*, maize and Retrotransposons
17. Structural and Functional Genomics — 3

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 Barlett 1994
8 Modem 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 & Trempy, Blackwell, 2004
14. Genomes 3, TA Brown, Garland, 2007

Paper MHG-202: Human Molecular Genetics and Human Genomics

(No. of classes of 60 mins each)

Unit I

1. Genetic mapping of Mendelian characters: 2
   - 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: 3
   - 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
   - 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 3
   - 4.1 Low resolution mapping- Cell hybrids, mini- and microcells, synteny of genes,
   - 4.2 Radiation hybrid mapping.
   - 4.3 Assembly of clone contigs and identifying genes in cloned DNA
5. Integration of cytogenetic, genetic and physical maps 1

Unit II

6. History, HGP organization and goals of human genome project 1
7. The Genome projects: 2
   - 7.1 Mapping strategies, current status of various maps; DNA segment nomenclature
   - 7.2 ELSI
   - 7.3 Benefits & patenting of genetic materials
8. Human genome diversity project (HGDP): General idea on 1000 Genome Project, Encode project 2
9. Organization of human genome: 2
   - 12.1 Mitochondrial genome
   - 12.2 Nuclear genome -Gross base composition, gene density, CpG islands
10. Comparative genomics -Characteristics of genomes of human and other model organisms (Sacchromyces cerevisea, *Caenorhabditis elegans, Drosophila melanogaster* and mouse) 2

Unit III

11. Human genome structure: 2
    - 11.1 RNA-encoding genes, functionally identical/similar genes
    - 11.2 Diversity in size and organization of genes
    - 11.3 Pseudogenes
12. Gene families in human genome 2
    - 12.1 Multigene families -Classical gene families, families with large conserved domains, families with small conserved domains, evolutionary concepts
    - 12.2 Gene super families
    - 12.3 Gene families in clusters
13. Small RNAs: RNAi, siRNA and miRNA: General idea and applications 2
14. Functional genomics -ESTs, Transcriptosome, Proteome, Multiplex and DNA microarray (chip) based analysis, LC-MS 2

Unit IV

15. Molecular pathology 3
    - 15.1 Nomenclature of mutations and their databases
    - 15.2 Loss of function and gain of function mutations in diseases
16. Molecular pathology: Human genome instability & pathogenicity associated with repeated sequences 2
    - 16.1 Slipped strand mispairing
    - 16.2 Unequal crossover and unequal sister chromatid exchange
    - 16.3 Gene conversion
    - 16.4 Retrotransposition
    - 16.5 Illegitimate recombination
17. Identifying human disease genes 3
    - 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. DNA testing

18.1 Direct and indirect testing (gene tracking) in individuals

18.2 DNA tests for identity and relationships including forensic applications

18.3 Population screening- ethics, organization and advantages

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-203: Immunogenetics

Unit I **(No. of classes of 60 mins each)**

1. General introduction to immune system — 2
    - 1.1 Innate and adaptive immunity
    - 1.2 cells and organs of the immune system
    - 1.3 Immune responses
2. Antigens, antibodies and T cell receptors — 4
    - 2.1 Antigens: Immunogenicity vs antigenicity
    - 2.2 Structure and function of antibody: Ig G, Ig M, Ig A, Ig E & Ig D
    - 2.3 Monoclonal Antibodies
    - 2.4 B and T cell receptors and coreceptors
    - 2.5 Antigen-antibody interactions
3. Immunoglobulin — 5
    - 3.1 Organization of Ig gene loci
    - 3.2 Molecular mechanisms of generation of antibody diversity
    - 3.3 Expression of Ig genes
    - 3.4 Regulation of Ig gene transcription
    - 3.5 Antibody Engineering

Unit II

4. T cell receptor — 2
    - 4.1 Organization of TCR gene loci
    - 4.2 Generation of TCR diversity
5. The HLA Complex — 4
    - 5.1 General organization & inheritance
    - 5.2 MHC molecules & genes
    - 5.3 Expression of HLA genes
    - 5.4 Regulation of HLA Expression
6. Role of HLA in disease susceptibility — 1
    - 6.1 HLA polymorphism
    - 6.2 Mechanism of disease association and HLA associated diseases

Unit III

7. Generation and regulation of immune responses-I — 6
    - 7.1 Antigen processing and presentation and MHC restriction
    - 7.2 Cytokines and Leukocyte, activation and migration
    - 7.3 T cell maturation, activation and differentiation
    - 7.4 B cell maturation, activation and differentiation
8. Generation and regulation of immune responses-II — 4
    - 8.1 Cell mediated cytotoxic responses
    - 8.2 Clonal selection and immunological memory
    - 8.3 Complement system
    - 8.4 Regulation of immune responses and Immunological tolerance
9. Introduction to immunosenescence — 1

Unit IV

10. Human Immune system disorders — 4
    - 10.1Primary and Secondary Immunodeficiencies
    - 10.2 Auto immunity & auto immune disorders (e.g., RA/SLE/MS)
    - 10.3 Hypersensitive reactions
    - 10.4 Cytokine related diseases
11. Immune system in human health — 4
    - 11.1 Immune response to infectious diseases and malignancy (Immunity to tumors)
    - 11.2 Concept of immunotherapy
    - 11.3 Vaccines
    - 11.4 Transplantation immunology: (Allograft, Xenograft, Syngraft, Graft versus host and host versus graft rejections).
12. Basics of Host-Pathogen interaction, evolution of pathogenicity and regulation of virulence — 2
13. Mechanism of drug resistance in pathogens: Viruses & Bacteria — 1

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 Immunolgy, 6th Ed., Virrela, Informa Health Care, 2007
7. Immunolgy, Janeway & Travers, Garland Publishing Inc, 1994
8. Essential Immunology, Roitt Blackwell 1994
9. Immunology, Roitt et al Mosloy 1993
10. hmnunology -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. Fundamaentals of Immumnology, William Paul, Freeman

Paper MHG-204: Biochemistry: Structure, Function and Regulation of Biomolecules

(No. of classes of 60 mins each)

Unit I

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

Unit II

6. Lipids — 3
   - 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 — 1
8. Prostaglandins: structure and function — 1
9. Disorders of lipid metabolism — 1
10. Hormones — 2
    - 10.1 Characteristics
    - 10.2 Mechanism of action of peptide and steroid hormones

Unit III

11. Hormone receptors and diseases — 1
12. Amino acids and peptides — 2
    - 12.1 Essential and non-essential amino acids
    - 12.2 Porphyrins and bile pigments
13 Metabolism of essential amino acids and related disorders — 2
14 Small peptides and their biomedical importance — 1
15 Structure- conformation-function relationship of proteins: Insulin, Hemoglobin and Collagen — 2

Unit IV

16 Protein folding and Protein degradation — 2
17. Enzymes: — 4
    - 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(Types of helical structures) — 2
19. Nucleotide Metabolism: Synthesis and degradation of pyrimidine and purine nucleotides — 2
20. Disorders of nucleotide metabolism — 1

Paper MHG-205: (Based on Theory Papers MHG-201 & MHG-202)

Molecular Genetics & Genomics, Human Molecular Genetics and Human Genomics

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. PCR-based detection of allelic inheritance of a DNA marker
6. Molecular detection of genetic diseases

Scheme of Practical MHG-205

Maximium Marks: 60

1. Experiments on bacterial culture/determination of Growth curve	9
2.transformation, selection of clones, etc.	9
3. PCR based detection of allelic inheritance of a DNA marker	9
4. Spots (8): (*Molecular genetics, Human Molecular Genetics, Human Genomics, etc.*)	16
5. Viva Voce (Experiment related)	10
6. Practical record	7
Total	**60**

Paper MHG-206: (Based on Theory Papers MHG-203 & MHG-204)
Immunogenetics and Biochemistry

1. Precipitation and agglutination reactions
2. Study of cell types of immune system
3. Immunodiffusion
4. Antibody titration
5. Enzyme linked immuno-absorbent assay (ELISA)
6. Blood grouping & Rh factor determination
7. Immuno-localization of antigens
8. Buffers, pH, preparation of solutions
9. Spectrophotometric estimation of glucose, cholesterol and protein
10. Sugar estimations in normal and diabetic patients
11. Assay and kinetics of Alkaline phosphatase/Esterase

SCHEME:

Time: 5 hrs MM: 60	7
1. Buffer & PH measurement/ prepration of solutions/enzyme kinetics	7
2. Spectrophoyometric estimation of proteins/cholesterol/glucose	7
3. ELISA/Immunodiffusion/ Immunoelectrophoresis/ Immunodetection	7
4. Blood group & Rh detection/ Immune cells	16
5. Spot (8): (*Biochemistry & Immunogenetics related*	10
6. Viva Voce (Experiment related)	6
7. Practical Record	**60**
Total	

Paper MHG-207: Seminar (From Syllabus) Credit: **1**
Paper MHG-208: Research Paper Presentation Credit: **1**
Paper MHG-209: Comprehensive Viva Voce Virtual Credit: **4**

Semester III

Paper MHG-301: Developmental and Reproductive Genetics

A. Developmental Genetics **(No. of classes of 60 mins each)**

Unit I

1 Early development 4
- 1.1 Fertilization
- 1.2 Types of cleavage
- 1.3 Gastrulation: Cell movement and formation of germ layers in frog, chick and mouse
- 1.4 Concept of determination, competence and differentiation

2. Development of vertebrate nervous system 3
- 2.1 Formation of neural tube
- 2.2Formation of brain region
- 2.3Tissue architecture of central nervous system

3. Genetics of pattem formation in *Caenorhabditis:* Vulva formation 1
4. Genetics of pattem formation in Vertebrates: 2
- 4.1. Axes formation and HOX genes
- 4.2. Limb formation in chick

Unit II

5. Genetics of pattem formation in *Drosophila* 4
- 5.1 Maternal effect genes and formation of body axes
- 5.2 Segmentation genes
- 5.3 Homeotic genes' function
- 5.4 Imaginal disc development

6. Regeneration: Types of regeneration; Regeneration in Hydra, Salamander & liver regeneration 2
7. Senescence: Concept and theories of Ageing; Age related disorders
8. Sex determination: 3
- 8.1 Mechanisms of sex determination in eukaryotes: Heterogamatic & Homogametic, Haplodiploidy
- 8.2 Role of Environmental factors; Mosaics and Gynandromorphs;
- 8.3 Sex determination in Melandrium.

Unit III

9. Sex determination in *Drosophila* 3
- 9.1 Genic Balance theory
- 9.2 Molecular mechanism of sex determination
- 9.3 Mechanism of Sexual dimorphism

10. Sex determination in Mammals: Endocrine & Molecular Mechanism of sex determination 2
11. Sex determination in Humans: 2
- 11.1Human Y chromosome- evolution, structure,
- 11.2 Molecular organization and its role in sex determination

12. Sex determination in *Caenorhabditis elegans*. 2

B. Reproductive Genetics

Unit IV

13. Human Development 3
- 13.1 Differentiation of Germ cells and Gametogenesis,
- 13.2 Fertilization, ovulation and implantation
- 13.3 Stages of Human embryonic development

14. Human Developmental Disorders 5
- 14.1Abnormal implantation: contribution of maternal and paternal genes
- 14.2 Teratogenesis and tumors associated with gastrulation
- 14.3 Birth defects: erythroblastsis fetalis, fetal hydrops and twin defects
- 14.4 Neural crest, Craniofacial and skeletal dysplasias
- 14.5 Vertebral defects: spina bifidia and scoliosis
- 14.6 Defects in sex differentiation

15. Human Reproductive Issues
    15.1 Abnormal gametes and infertility
    15.2 Spontaneous abortions and still birth (etiology, pathogenesis, genetic characterstics, clinical notes, diagnosis and management) 1
    15.3 Reproductive options: Assisted reproductive techniques (ARTs), IVF

Recommended Books

1. Developmental Biology, 8th Ed., Gilbert, Sinauer, 2006
2. Principles of Developmental Genetics, Moody, Elsevier, 2007
3. Principles of Developrnent, 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'Rahi1ly and Muller Wiley 1992

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

(No. of classes of 60 mins each)

Unit I

1. An overview of the genetic basis of syndromes and disorders — 1
2. Monogenic diseases with well known molecular pathology — 3
   - 2.1. Cystic fibrosis
   - 2.2. Tay-Sachs Syndrorne
   - 2.3. Marfan syndrome
3. Inborn errors of metabolism and their genetic bases — 3
   - 3.1 Phenylketonuria
   - 3.2 Mucopolysaccharidosis
   - 3.3 Galactosemia
4. Neurogenetic disorders — 3
   - 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 — 1
6. Muscle genetic disorders — 3
   - 6.1 Dystrophies (Duchenne Muscular dystrophy and Becker Muscular Dystrophy)
   - 6.2 Myotonias
   - 6.3 Myopathies
7. Genetic disorders of Haemopoitic systems — 3
   - 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 — 4
   - 8.1 Colour Blindness
   - 8.2 Retinitis pigmentosa
   - 8.3 Glaucoma
   - 8.4 Cataracts

Unit III

9. Syndromes: — 2
   - 9.1 Genomic syndromes: Neurofibromatosis I syndrome
   - 9.2 Genome imprinting: Prader-Willi and Angelman syndromes, Beckwith-Wiedeman syndrome
10. Cancers and cancer-prone syndromes — 3
    - 10.1 Haematological malignancies
    - 10.2 Retinoblastoma, Wilm's turnour, Colorectal cancer
    - 10.3 DNA-repair deficiency syndromes
    - 10.4 Breast cancer
11. Complex polygenic syndromes — 3
    - 11.1 Hyperlipidemia
    - 11.2 Atherosclerosis
    - 11.3 Diabetes mellitus
12. Mitochondrial syndromes — 1
13. Management of genetic disorders — 1

Unit IV

14. Historical overview of genetic counseling I: — 2
    - 14.1 Models of Eugenic, Medical/Preventive, decision making, Psychotherapeutic Counseling; current definition and goals
    - 14.2 Philosophy and Ethos of genetic services and counseling
15. Components of genetic counseling II: — 2
    - 15.1 Indications for and purpose
    - 15.2 Information gathering and construction of pedigrees
    - 15.3 Medical Genetic evaluation: Basic components of Medical, Past medical, social & family history
    - 15.4 Physical examination: General and dysmorphology examination
    - 15.5 Documentation, Legal and ethical considerations

16. Patterns of inheritance, risk assessment and counseling in common Mendelian and Multifactor syndromes 2
17. Prenatal and Preimplantation diagnosis 2
    17.1 Indications for prenatal diagnosis and for chromosomal testing
    17.2 Noninvasive and Invasive methods 1
18. Genetic testing: biochemical & molecular tests
    18.1 In children
    18.2 Presymptomatic testing for late onset diseases (predictive medicine)

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., Churchil 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: Population Genetics, Human Evolutionary and Behavior Genetics

(No. of classes of 60 mins each)

Unit I

1. Concept and theories of evolution — 1
2. Microevolution in Mendelian population — 2
   - 2.1 Mendelian Population
   - 2.2 Allele frequencies and genotype frequencies
   - 2.3 Hardy-Weinberg equilibrium and conditions for its maintenance — 2
3. Elemental forces of evolution
   - 3.1 Mutation
   - 3.2 Selection
   - 3.3 Genetic drift
   - 3.4 Migration — 2
4. Nonrandom and random breeding
   - 4.1 Inbreeding and assortative mating
   - 4.2 Inbreeding coefficient, allelic identities by descent
   - 4.3Heterosis — 2
5. Isolating mechanisms: Geographic and reproductive isolation — 2
6. Concept of species and modes of speciation: sympatry, allopatry, stasipatry & parapatry — 2

Unit II

7. Genetic variability in natural population I:
   - 7.1 Chromosomal polymorphism
   - 7.2 Enzyme polymorphism
   - 7.3 DNA polymorphism — 2
8. Genetic variability in natural population II:
   - 8.1 Adaptive genetic polymorphism
   - 8.2 Balanced polymorphism
   - 8.3 Linkage disequilibrium — 2
9. Molecular population genetics
   - 9.1 Molecular evolution (neutral theory, punctuated equilibrium)
   - 9.2 Molecular clock — 3
10. Molecular Phylogenetics:
    - 10.1 Construction of phylogenetic tree using nucleotide sequence data
    - 10.2 Amino acid sequence and phylogeny (globin gene, cytochrome b gene, etc.)
    - 10.3 DNA-DNA hybridization
    - 10.4 Restriction enzyme sites
    - 10.5 Nucleotide sequence comparison and homologies — 3

Unit III

11. Human phylogeny
    - 11.1 Hominid evolution. anatomical, Geographical, Cultural
    - 11.2 Molecular phylogenetics of Homo sapiens — 1
12. Peopling of continents (Europe, Africa, Asia) — 3
13. Admixture:
    - 13.1 Meeting of human populations & its genetic imprint
    - 13.2 Detection of admixture (based on allele frequencies & DNA data)
    - 13.3 Y Chromosome & mitochondrial DNA markers in genealogical studies — 2
14. Culture and human evolution
    - 14.1 Learning, society and culture
    - 14.2 Relative rates of cultural and biological evolution
    - 14.3 Social Darwinism
    - 14.4 Sociobiology

Unit IV — 4

15. Nature-nurture and behaviour
    - 15.1 Genetic experiments to investigate animal and human behaviour
    - 15.2 Identifying genes for behavior (induced mutations, QTL, synteny homology)
    - 15.3 Environmental influence- shared and non-shared environment
    - 15.4 Investigating genetics of human behaviour (twin & adoption study designs, Interpretating heritability, linkage and association studies)

16. Psychopathology
    16.1 Signs and symptoms
    16.2 Schizophrenia
    16.3 Mood disorders
    16.4 Anxiety disorders
    16.5 Disorders of childhood
    16.6 Personality and personality disorders- antisocial personality, criminal behaviour 2
17. Cognitive abilities and Disabilities
    17.1 Mental retardation
    17.2 Learning disorders 2
    17.3 Communication disorders
18. Neurogenetics:
    18.1 Study design: Genetic and environmental manipulations
    18.2 Circadian rhythms
    18.3 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. 33

Paper MHG-304: A. Recombinant DNA Technology
B. Molecular Diagnostic methods

(No. of classes of 60 mins each)

A. Recombinant DNA Technology

Unit I

1. Enzymes used in DNA technology — 2
   - 1.1 Restriction and modification enzymes
   - 1.2 Other nucleases
   - 1.3 Polymerases
   - 1.4 Ligase, kinases and phosphatases , — 3
2. Cloning vectors
   - 2.1. Plasmids
   - 2.2 Phages
   - 2.3 Cosmids
   - 2.4 Artificial chromosomes
   - 2.5 Shuttle vectors — 2
   - 2.6 Expression vectors
3. Construction of genomic and cDNA libraries — 5

Unit II

4. Screening and characterization of clones
   - 4.1 Preparation of probes
   - 4.2 Principles of hybridizations and hybridization based techniques (Colony, plaque, Southern, Northern and in situ hybridizations)
   - 4.3 Expression based screening — 5
   - 4.4 Interaction based screening: yeast two-hybrid system
5. Basic Principles and Applications of the following techniques
   - 5.1 DNA sequencing
   - 5.2 Oligonucleotide synthesis
   - 5.3 Polymerase Chain Reaction
   - 5.4 DNA Fingerprinting
   - 5.5 Microarray — 2
6. Promoter characterization: promoter analysis through reporter genes, electrophoretic mobility shift assay, DNA foot-printing. — 2

Unit III

7. Microcloning and Positional cloning: RFLP mapping, chromosome walking and jumping — 3
8. Mutagenesis
   - 8.1 Site directed mutagenesis
   - 8.2 Transposon mutagenesis
   - 8.3 Construction of knockout mutants — 3
9. Gene transfer techniques
   - 9.1 Microinjection
   - 9.2 Transfection of cells: Principles and methods
   - 9.3 Germ line transformation in *Drosophila,* transgenic and knock out mice: Strategies and methods

B. Molecular Diagnostic Methods

Unit IV

10. Testing DNA variation for diseases association — 3
    - 10.1SNPs & Diseases
    - 10.2 Methods of SNP Typing: Brief idea of Traditional approach, Taqman
    - 10.3 Next generation sequencing, exome sequencing — 3
11. Microarray approach to gene expression analysis (Brief idea)
    - 11.1 DNA microarray platforms
    - 11.2 cDNA array
    - 11.3 SAGE, Array CGH — 3
12. HLA Typing using molecular methods (Brief idea)
    - 12.1 PCR with sequence-specific primer

12.2 Sequence-specific oligonucleotide probe hybridization
12.3 Sequenced-based HLA typing

13. Methods for analysis of DNA Methylation (Brief idea)
13.1 Bisulphite modification
13.2 Methylation-specific PCR, Bisulfite sequencing
13.3 Real time PCR methods, Pyro-sequencing

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-305: (Based on Theory Papers MHG-301 & MHG-302)
Developmental and Reproductive Genetics; Clinical Genetics and Genetic Counseling

1. Facial landmarks and dermatoglyphia
2. Risk assessment (Binomial probability and Bayesian calculation)
3. Detection of mutations in Thalassemia patients
4. Preparation of Human Karyotype and making idiogram of the banded chromosomes
5. Micrographs demonstrating examples of molecular methods, development stages etc.
6. Study of expression of segmentation genes in *Drosophila*
7. Observation of homeotic mutants of *Drosophila*
8. Study of Frog development
9. Study of Chick embryo development (whole mounts & permanent slides)
10. Study of various stages of human fetal development (Observation of models)

SCHEME:
Time: 5 hrs MM: 60

1. Demonstration of chick embryo development	7
2. Diagnosis of Thalassemia mutations by multiplex PCR-based methods/ Karyotyping	7
3. Pedigree construction & Risk assessment in pedigree	7
4. Facial landmark/Dermatoglyphia	7
5. Spots (8): (*Development, Reproductive and Clinical genetics related*)	16
6. Viva Voce (Experiment related)	10
7. Practical Record	6
Total	**60**

Paper MHG-306: (Based on Theory Papers MHG-303 & MHG-304)
Population Genetics, Human Evolutionary and Behavior Genetics; Recombinant DNA Technology and Molecular Diagnostic methods

1. Genomic DNA extraction from mouse/human/fly
2. Quantification of DNA on gel and by spectrophotometer
3. Isolation of plasmid DNA and Restriction mapping of plasmid DNA
4. Southern Hybridization
5. Western blotting
6. Bisulfite modification for methylation analysis
7. PCR-RFLP for detection of allelic inheritance of a DNA marker
8. Microsatellite/RAPD/mitochondrial marker-based detection of molecular polymorphism in populations.
9. Study of courtship behavior in *Drosophila*
10. Personality analysis (through questionnaires); IQ measurement
11. Visit to mental hospital to study behavior of Schizophrenia & other mental disorders patients
12. Hardy-Weinberg Genetic equilibrium: PTC Tasters & non-tasters; Calculation of gene & genotype frequencies
13. Study of Models/Photographs on molecular genetics, evolutionary principles and examples of molecular methods

SCHEME:
Time: 5 hrs MM: 60

1. Plasmid/genomic DNA isolation and agarose gel electrophoresis of DNA and separation on gel	7
2. PCR-RFLP for detection of allelic inheritance	7
3. Restriction mapping / Hybridization methods	7
4. Experiments on population genetics/evolutionary principles	7
5. Spot (8): (*RDT, Population, Evolutionary and Behavior Genetics*)	16
6. Viva Voce (Experiment related)	10
7. Practical Record	6
Total	**60**

Paper MHG-307: Seminar (From Syllabus) Credit: **1**
Paper MHG-308: Research Paper Presentation Credit: **1**
Paper MHG-309: Comprehensive Viva Voce Virtual Credit: **4**

Semester IV

Paper MHG-401: Bioinformatics and Biotechniques

(No. of classes of 60 mins each)

A. Bioinformatics

Unit I

1. Bioinformatics: Introduction and scope of Bioinformatics: An Overview — 1
2. Biological databases — 3
   - 2.1 Nucleotide sequence databases (GenBank, EMBL, DNA data bank of Japan)
   - 2.2Genome databases (Ensembl, Flybase, MGI Mouse Genome),
   - 3.3 Protein databases (UniProt, PIR/Protein Identification Resource, SWISS-PROT, Human Proteinpedia, etc.)
3. Sequence analysis (Brief idea): — 2
   - 3.1 Methods for alignment (dot matrices)
   - 3.2 Tools for sequence alignment – Fasta, BLAST, PSI-Blast,
4. Bioinformatics tools for multiple sequence alignment and phylogenetic analysis (PHYLIP, CLUSTAL W) — 1

Unit II

5. Gene prediction (Brief ideas): Methods & gene prediction tools — 1
6. Bioinformatic Tools for Protein Research (Brief ideas): — 3
   - 6.1 Peptide Sequence notation and applications
   - 6.2 Protein Structure predictions
   - 6.3 Protein function prediction
7. Haplotype and linkage analysis using Haploview, Arlequin software — 2
8. Introductory ideas on virtual libraries: — 2
   - 8.1 MEDLINE, Science Citation Index, SCOPUS
   - 8.2 Electronic Journals and retrieval of other information related to research (PubMed, PMC)
9. Introduction to Nanotechnology and its applications — 2

B. Biotechniques

Unit III

10. Basic principles and applications of: — 2
    - 10.1 Light & Dark-field Microscopy
    - 10.2 Phase-contrast Microscopy
11. General idea on the principles and applications of: Transmission & Scanning Electron Microscope — 3
12. General idea on the principles and applications of: — 2
    - 12.1 Fluorescence Microscopy
    - 12.2 Confocal Microscopy
13. Basic Principles and applications of Absorption and Fluorescence spectrophotometer — 2
14. General principle, applications and methods of: Partition and Adsorption Chromatography — 2

Unit IV

15. Cell Imaging: Photomicrography & Image analysis system — 2
16. General Principles and Application of Flow Cytometry — 2
    - 16.1 Fluorescent activated cell sorter
    - 16.2 Fluorescent Labels
    - 16.3 Cytometric Bead Array
17. Centrifugation: Types, rotors and applications — 2
18. General principles, methods and applications of PAGE and 2D gel electrophoresis — 2
19. Principle, methods and applications of Auto-radiography — 2

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
4. Electrophoresis in Practice, 4th Ed., Westermeir, Wiley, 2005
5. Basic Methods in Microscopy, Spector, CSHL Press, 2006
6. Laboratory Protocols in Applied Life Sciences, Bisen PS, CRC Press, 2014
7. Introduction to Instrumentation in Life Sciences, Bisen & Sharma, CRC Press, 2013
8. Current Topics in Computational Molecular Biology, Jiang et al, Anne Books, 2004
9. Bioinformatics, Brown, Eaton, 2000
10. Bioinformatics for Geneticists, 2nd Ed., Barnes, Wiley, 2007
11. Bioinformatics Computing, Bergeron, Pearson, 2003
12. Bioinformatics: Methods and Protocols, Misener & Krawetz, Human Press, 2004
13. Internet and e-mail, 2nd Ed., Bangia, Khanna, 2002
14. Proteomics, O'Connor & Hames, Scion, 2008
15. Proteomics in Practice, 2nd Ed., Westermeir et al, Wiley, 2008