



**M.Sc. in Molecular and Human  
Genetics**

**DISTRIBUTION OF DIFFERENT PAPERS AND CREDITS IN VARIOUS SEMESTERS  
(2015-2017)**

Semester	Paper Code	Name of the Paper	Type of Paper	Credits
<i>Semester I</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 papers 103 & 104	Core	3
	MHG-107	Seminar	Core	1
	MHG-108	Assignment	Core	1
	MHG-109	Comprehensive Viva Voce	Virtual	4
	<b>Total Credits</b>			
<i>Semester II</i>	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
	<b>Total Credits</b>			
<i>Semester III</i>	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
	<b>Total Credits</b>			
<i>Semester IV</i>	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
	<b>Total Credits</b>			



**JIWAJI UNIVERSITY, GWALIOR**



**M.Sc. in Molecular and Human  
Genetics**

**(2015-2017)**

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*
  - 6.3 Genetic distance and physical distance
  - 6.4 Genetic and cytological crossing over
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

**Recommended Books**

1. An Introduction to Genetic Analysis, 7<sup>th</sup> Ed., Griffiths et al, Freeman, 2000
2. Genetics, 3<sup>rd</sup> Ed., Strickberger, Macmillan, 1985
3. Genetics: Analysis of Genes and Genomes, 6<sup>th</sup> Ed., Hartl and Jones, Jones & Bartlett, 1998
4. Concepts of Genetics, 9<sup>th</sup> Ed., Klug and Cummings, Pearson, 2009
5. Principles of Genetics, 7<sup>th</sup> Ed., Tamarin, Tata McGraw Hill, 2002
6. Principles of Genetics, 3<sup>rd</sup> Ed., Snustad and Simmons, Wiley, 2003
7. Schaum's Outline of Genetics, 4<sup>th</sup> Ed., Elrod and Stansfield, McGraw Hill, 2002
8. Principles of Genetics, Gardner et al., John Wiley 1991
9. Mathematical & Statistical Methods for Genetic Analysis, 2<sup>nd</sup> Ed., Lange, Springer, 2004
10. Methods in Biostatistics, 6<sup>th</sup> Ed., B. K. Mahajan, Jaypee, 2004

## 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 patterns I: Non-penetrance, variable expressivity, pleiotropy, onset, dominance problem; anticipation, compound heterozygosity 2
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. 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)
11. Human chromosomal pathologies: 2
  - 11.1 Numerical aberrations and their common syndromes
  - 11.2 Structural aberrations and their common syndromes (translocations, duplications, deletions, microdeletion 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

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



# Paper MHG-104: A. Molecular Organization of Chromatin and Cytogenetics

## B. Cancer Genetics

(No. of classes of 60 mins each)

### A. Molecular Organization of Chromatin and Cytogenetics

#### 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 tumorigenesis: 2
  - 18.1 Cell cycle check-point defects
  - 18.2 Tumor specific markers
19. Chromosomal basis of Cancer 3
  - 19.1 Philadelphia 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, 6<sup>th</sup> Ed., Watson et al, CSH Press, 2008
2. Genes IX, Benjamin Lewin, Jones and Bartlett, 2008
3. Human Chromosomes, 4<sup>th</sup> Ed., Miller and Therman, 2001
5. First years of Human Chromosomes, Harper, Scion, 2006
6. Molecular Biology of the Cell, 4<sup>th</sup> Ed., Alberts et al, Garland, 2002
7. Human Cytogenetics- A practical Approach, 3<sup>rd</sup> 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: 100**

1. Analysis of Mono hybrid / Dihybrid / Sex-linkage crosses	14
2. Linkage & Crossing over / Three Point Test cross & Gene mapping	12
3. Karyotyping and Idiogram preparation/ G- or C-banding of mammalian chromosomes	16
4. Facial landmarks/Dermatoglyphia	14
5. Spotting (8): ( <i>Spots based on genetic inheritance, Basic Human Genetics, Human Cytogenetics</i> )	24
6. Viva Voce (Experiment related)	10
7. Practical record	10
<b>Total</b>	<b>100</b>

**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	16
2. Study of mitosis/meosis from onion root tips/grasshopper testis	12
3. Histological preparation and histochemical staining to show biomolecules	16
4. Endocytosis/Study of polytene chromosome	12
5. Spotting (8): ( <i>Cell Biology, Chromosome Organization, Cancer Biology</i> ).	24
7. Viva Voce (Experiment related)	10
8. Practical Record	10
<b>Total</b>	<b>100</b>

**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
  - 16.1 Mechanism 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 Modern genetic Analysis. Griffith et al Freeman 1999
9. Molecular Biology of the Gene, 6<sup>th</sup> 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, 6<sup>th</sup> 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 2  
(*Saccharomyces cerevisiae*, *Caenorhabditis elegans*, *Drosophila melanogaster* and mouse)

### 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, 7<sup>th</sup> Ed, Nussbaum et al, Elsevier, 2007
7. Human Genetics and Genomics, 3<sup>rd</sup> Ed, Korf, Blackwell, 2007
8. Molecular Diagnosis, 2<sup>nd</sup> 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, 3<sup>rd</sup> Ed, Vogel, Springer, 1997



## Paper MHG-203: Immunogenetics

(No. of classes of 60 mins each)

### Unit I

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
  - 10.1 Primary 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, 6<sup>th</sup> Ed., Abbas et al, Elsevier, 2007
2. Immunology, 6<sup>th</sup> Ed Roitt, Mosby, 2002
3. Immunology, 5<sup>th</sup> Ed., Kuby, Freeman, 2002
4. Microbiology, 6<sup>th</sup> Ed., Prescott et al, McGraw Hill, 2005
5. Microbiology: A Human Perspective, 4<sup>th</sup> Ed., Nester et al, McGraw Hill, 2004
6. Medical Immunology, 6<sup>th</sup> 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, Benjamin 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-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	2
19. Nucleotide Metabolism: Synthesis and degradation of pyrimidine and purine nucleotides	2
20. Disorders of nucleotide metabolism	1

**Recommended Books**

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

**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  $\alpha$ -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**

**Maximum Marks: 100**

1. Experiments on bacterial culture (Growth curve)/ transformation, selection of clones, etc.	16
2. PCR based detection of allelic inheritance of a DNA marker	14
3. Molecular detection of genetic diseases	14
4. Gene induction in <i>Drosophila</i> (heat shock treatment)/Transgenic for hsp70-lacZ gene	12
5. Spots (8): ( <i>Molecular genetics, Human Molecular Genetics, Human Genomics, etc.</i> )	24
6. Viva Voce (Experiment related)	10
7. Practical record	10
<b>Total</b>	<b>100</b>

**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: 100**

1. Spectrophotometric estimation of proteins	05
2. Sugar estimation of diabetic patient/Enzyme kinetics	05
3. ELISA/Immunodiffusion	05
4. Blood group & Rh detection/ Immune cells/ Immunodiffusion/ Immunoelectrophoresis/ Immunodetection	05
5. Spot (8): ( <i>Biochemistry &amp; Immunogenetics related</i> )	16
6. Seminar	10
7. Viva Voce (Experiment related)	10
8. Practical Record	05
<b>Total</b>	<b>60</b>

**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**

(No. of classes of 60 mins each)

**A. Developmental Genetics**

**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  |   |
| 2.2 Formation of brain region   |   |
| 2.3 Tissue architecture of central nervous system                                     |   |
| 3. Genetics of pattern formation in <i>Caenorhabditis</i> : Vulva formation           | 1 |
| 4. Genetics of pattern formation in Vertebrates:                                      | 2 |
| 4.1. Axes formation and HOX genes   |   |
| 4.2. Limb formation in chick  |   |

**Unit II**

- |  |   |
|--|---|
| 5. Genetics of pattern formation in <i>Drosophila</i>  | 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: Heterogametic & Homogametic, Haplodiploidy  |   |
| 8.2 Role of Environmental factors; Mosaics and Gynandromorphs;                                 |   |
| 8.3 Sex determination in Melandrium.   |   |

**Unit III**

- |  |   |
|--|---|
| 9. Sex determination in <i>Drosophila</i>  | 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 |
| 11.1 Human Y chromosome- evolution, structure,   |   |
| 11.2 Molecular organization and its role in sex determination                          |   |
| 12. Sex determination in <i>Caenorhabditis elegans</i> .                               |   |

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 |
| 14.1 Abnormal implantation: contribution of maternal and paternal genes      |   |
| 14.2 Teratogenesis and tumors associated with gastrulation                   |   |
| 14.3 Birth defects: erythroblastosis fetalis, fetal hydrops and twin defects |   |
| 14.4 Neural crest, Craniofacial and skeletal dysplasias                      |   |
| 14.5 Vertebral defects: spina bifida and scoliosis                           |   |
| 14.6 Defects in sex differentiation  |   |

15. Human Reproductive Issues	3
15.1 Abnormal gametes and infertility	
15.2 Spontaneous abortions and still birth (etiology, pathogenesis, genetic characteristics, clinical notes, diagnosis and management)	
15.3 Reproductive options: Assisted reproductive techniques (ARTs), IVF	1

### **Recommended Books**

1. Developmental Biology, 8<sup>th</sup> Ed., Gilbert, Sinauer, 2006
2. Principles of Developmental Genetics, Moody, Elsevier, 2007
3. Principles of Development, 2<sup>nd</sup> Ed., Wolpert, Oxford 2002
4. The Cellular & Molecular Biology of Pattern Formation, Stocum & Karr, 1990
5. Larsen's Human Embryology, 4<sup>th</sup> Ed., Churchill Livingstone, 2009
6. Langman's Medical Embryology, 10<sup>th</sup> Ed., Sadler, LMW, 2006
7. Human Embryology, 8<sup>th</sup> Ed., Singh & Pal, McMillan, 2007
8. Smith's Recognizable Patterns of Human Malformations, 6<sup>th</sup> Ed, Jones, Elsevier, 2006
9. Neural tube defects, Oppenheimer, Informa, 2007
10. Essential Medical Genetics, Conner & Ferguson-Smith, 5<sup>th</sup> Ed., Blackwell Science, 1997
11. Emery's Element of Medical Genetics, 11<sup>th</sup> 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-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 Syndrome
  - 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 Haemopoietic 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 tumour, 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	
18. Genetic testing: biochemical & molecular tests	1
18.1 In children	
18.2 Presymptomatic testing for late onset diseases (predictive medicine)	

### **Recommended Books**

1. Thompson & Thompson, Genetics in Medicine, 7<sup>th</sup> Ed., Nuusbaum et al, Elsevier, 2007
2. Emery & Remoin's Principles & Practice of Medical Genetics, Vol I-III, 5<sup>th</sup> Ed., Churchill Livingstone, 2007
3. New Clinical Genetics, Read & Donnai, Scion, 2007
4. Emery's Element of Medical Genetics, 11<sup>th</sup> 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, 6<sup>th</sup> Ed, Jones, Elsevier, 2006
10. Neural tube defects, Oppenheimer, Informa, 2007
11. Essential Medical Genetics, Conner & Ferguson-Smith, 5<sup>th</sup> 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
3. Elemental forces of evolution 2
  - 3.1 Mutation
  - 3.2 Selection
  - 3.3 Genetic drift
  - 3.4 Migration
4. Nonrandom and random breeding 2
  - 4.1 Inbreeding and assortative mating
  - 4.2 Inbreeding coefficient, allelic identities by descent
  - 4.3 Heterosis
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: 2
  - 7.1 Chromosomal polymorphism
  - 7.2 Enzyme polymorphism
  - 7.3 DNA polymorphism
8. Genetic variability in natural population II: 2
  - 8.1 Adaptive genetic polymorphism
  - 8.2 Balanced polymorphism
  - 8.3 Linkage disequilibrium
9. Molecular population genetics 2
  - 9.1 Molecular evolution (neutral theory, punctuated equilibrium)
  - 9.2 Molecular clock
10. Molecular Phylogenetics: 3
  - 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

### Unit III

11. Human phylogeny 3
  - 11.1 Hominid evolution. anatomical, Geographical, Cultural
  - 11.2 Molecular phylogenetics of Homo sapiens
12. Peopling of continents (Europe, Africa, Asia) 1
13. Admixture: 3
  - 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
14. Culture and human evolution 2
  - 14.1 Learning, society and culture
  - 14.2 Relative rates of cultural and biological evolution
  - 14.3 Social Darwinism
  - 14.4 Sociobiology

### Unit IV

15. Nature-nurture and behaviour 4
  - 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	3
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	
17. Cognitive abilities and Disabilities	2
17.1 Mental retardation	
17.2 Learning disorders	
17.3 Communication disorders	
18. Neurogenetics:	2
18.1 Study design: Genetic and environmental manipulations	
18.2 Circadian rhythms	
18.3 Learning & memory	

### **Recommended Books**

1. Evolution, 4<sup>th</sup> Ed., Strickberger, Jones and Barlett, 2008
2. Human Evolution, 5<sup>th</sup> Ed, Roger Lewin, Blackwell, 2005
3. Evolutionary Analysis, 4<sup>th</sup> 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, 4<sup>th</sup> Ed., Plomin et al, Worth, 2001
7. Genetics: Analysis of Gene and Genomes, 6<sup>th</sup> Ed., Hartl & Jones, Jones and Bartlett, 2005
8. Neurogenetics of Psychiatric Disorders, Sawa & McInnes, Informa Healthcare, 2007
9. Synopsis of Psychiatry, 9<sup>th</sup> Ed, Kaplan & Sadock, LMW, 2003
10. Genetics of Population, 2<sup>nd</sup> Ed., Heidrick, Jones and Bartlett, 2000
11. Human Evolutionary Genetics, 1<sup>st</sup> 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 ,
2. Cloning vectors 3
  - 2.1 Plasmids
  - 2.2 Phages
  - 2.3 Cosmids
  - 2.4 Artificial chromosomes
  - 2.5 Shuttle vectors
  - 2.6 Expression vectors
3. Construction of genomic and cDNA libraries 2

##### Unit II

4. Screening and characterization of clones 5
  - 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
  - 4.4 Interaction based screening: yeast two-hybrid system
5. Basic Principles and Applications of the following techniques 5
  - 5.1 DNA sequencing
  - 5.2 Oligonucleotide synthesis
  - 5.3 Polymerase Chain Reaction
  - 5.4 DNA Fingerprinting
  - 5.5 Microarray
6. Promoter characterization: promoter analysis through reporter genes, electrophoretic mobility shift assay, 2 DNA foot-printing. 2

##### Unit III

7. Microcloning and Positional cloning: RFLP mapping, chromosome walking and jumping 2
8. Mutagenesis 3
  - 8.1 Site directed mutagenesis
  - 8.2 Transposon mutagenesis
  - 8.3 Construction of knockout mutants
9. Gene transfer techniques 3
  - 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
  - 10.1 SNPs & Diseases
  - 10.2 Methods of SNP Typing: Brief idea of Traditional approach, Taqman
  - 10.3 Next generation sequencing, exome sequencing
11. Microarray approach to gene expression analysis (Brief idea) 3
  - 11.1 DNA microarray platforms
  - 11.2 cDNA array
  - 11.3 SAGE, Array CGH
12. HLA Typing using molecular methods (Brief idea) 3
  - 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) 3
  - 13.1 Bisulphite modification
  - 13.2 Methylation-specific PCR, Bisulfite sequencing
  - 13.3 Real time PCR methods, Pyro-sequencing

**Recommended Books**

1. Recombinant DNA, 2<sup>nd</sup> 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, 5<sup>th</sup> Ed., Brown, Taylor & Francis, 2005
6. Principles of Gene Manipulation & Genomics, 7<sup>th</sup> Ed., Primrose & Twyman, Blackwell, 2006
7. Genetics: A Molecular Approach, 3<sup>rd</sup> Ed., Brown, Taylor & Francis, 2005
8. Molecular Cloning: A Laboratory Manual, 3<sup>rd</sup> 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: 100**

1. Demonstration of chick embryo development	16
2. Diagnosis of Thalassemia mutations by multiplex PCR-based methods/ Karyotyping	14
3. Pedigree construction & Risk assessment in pedigree	14
4. Facial landmark/Dermatoglyphia	12
5. Spots (8): ( <i>Development, Reproductive and Clinical genetics related</i> )	24
6. Viva Voce (Experiment related)	10
7. Practical Record	10
<b>Total</b>	<b>100</b>

**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: 100**

1. Plasmid/genomic DNA isolation and agarose gel electrophoresis of DNA and separation on gel	16
2. PCR-RFLP for detection of allelic inheritance	14
3. Restriction mapping / Hybridization methods	14
4. Experiments on population genetics/evolutionary principles	12
5. Spot (8): ( <i>RDT, Population, Evolutionary and Behavior Genetics</i> )	24
6. Viva Voce (Experiment related)	10
7. Practical Record	10
<b>Total</b>	<b>100</b>

**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)
  - 2.2 Genome databases (Ensembl, Flybase, MGI Mouse Genome),
  - 2.3 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, 2<sup>nd</sup> Ed., Freifelder, Freeman, 1999
3. Practical Biochemistry, 5<sup>th</sup> Ed., Wilson and Walker, Cambridge, 2003
4. Electrophoresis in Practice, 4<sup>th</sup> 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, 2<sup>nd</sup> 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, 2<sup>nd</sup> Ed., Bangia, Khanna, 2002
14. Proteomics, O'Connor & Hames, Scion, 2008
15. Proteomics in Practice, 2<sup>nd</sup> Ed., Westermeir et al, Wiley, 2008

## MHG 402: Bioinformatics and Biotechniques

1. Principles and handling of bright field, phase contrast and fluorescence microscope
2. Principles of Spectrophotometry and applications
3. Principles of centrifugation and applications
4. Separation of biomolecules by gel filtration
5. Chromatography- Paper & TLC
6. Agarose gel electrophoresis for separation of DNA
7. SDS-Polyacrylamide gel electrophoresis for separation of proteins
8. Use of Internet: Pubmed, Entrez, EMBL databases for literature search and for comparison of protein and DNA sequences.
9. Applications of BLAST, FASTA, CLUSTALW, GENSCAN, RASMOL, Phylodendron.
10. Primer Designing and Insilico PCR
11. Use of NEB cutter
12. Haplotype and Linkage Analysis

### Scheme of the Practical

1. Experiments on Biostatistics problem	10
2. Chromatography: Paper/TLC	10
3. Biochemical estimations of glucose/cholesterol/proteins	12
4. Separation of biomeolecules: protein/DNA	14
5. Experiments on Bioinformatic application: Primer designing/Haplotype analysis	10
6. Spots (8) ( <i>Bioinformatics, Bioinformatics</i> )	24
7. Viva Voce (Experiment related)	10
8. Practical Record	10
<b>Total `</b>	<b>100</b>

**Paper MHG-403:** Seminar (From Syllabus)

Credit:1

**Paper MHG-404:** Formulation of Research Project and Presentation

Credit:1

**Paper MHG-405:** Dissertation and Viva-voce Examination

Credit: 12

**Paper MHG-406:** Comprehensive Viva Voce of all semesters

Virtual Credit: 4