

# **SYLLABUS**

**M. Sc. Course In**

**MOLECULAR BIOLOGY AND HUMAN GENETICS**

*(With effect from the session 2021–2023)*

**[CHOICE BASED CREDIT SYSTEM]**



**THE UNIVERSITY OF BURDWAN  
BURDWAN, WEST BENGAL  
713104, INDIA**

The M.Sc. syllabus (2021-2023) has been endorsed in the meeting of Ad-hoc Board of Post Graduate Studies in Molecular Biology and Human Genetics held on 25/09/2021 and 28/01/2022. Subsequently approved the in the meeting of Faculty Council of Science on \_\_\_\_\_.

*(Teacher-in-charge)*

Dept. of Human Genetics & Molecular Biology, BU

## Department of Molecular Biology and Human Genetics

### *Summary of the course and credits*

#### Semester I [Credits – 24]

Course				Lect. Hr /Week	Dur. of Exam (in H)	Marks			Credit
Course code	Type	T/P	Name			I.A.	E.T	Total	
MSMHG101	Core	T	Transmission Genetics & Basic Human Genetics	4T	2T	10	40	50	4
MSMHG102	Core	T	Biochemistry & Cell Biology	4T	2T	10	40	50	4
MSMHG103	Core	T	Computational Biology, Biostatistics & Evolutionary Genetics	4T	2T	10	40	50	4
MSMHG104	Core	T	Molecular Biology & Bio techniques	4T	2T	10	40	50	4
MSMHG105	Core	P	Lab work based on courses MSMHG101 & MSMHG102	4P	4P	10	40	50	4
MSMHG106	Core	P	Lab work based on courses MSMHG103 & MSMHG104	4P	4P	10	40	50	4
Total credit									24

T/P: Theory/Practical

#### Semester II [Credits – 24]

Course				Lect. Hr /week	Dur. of Exam (in H)	Marks			Credit
Course code	Type	T/P	Name			I.A.	E.T	Total	
MSMHG201	Core	T	Reproductive Biology & Developmental Genetics	4T	2T	10	40	50	4
MSMHG202	Core	T	Immunology & Immunogenetics	4T	2T	10	40	50	4
MSMHG203	Core	T	Molecular Human Genetics & Human Genome	4T	2T	10	40	50	4
MSMHG204	Core	T	Genetic Engineering Model Genetic Systems	4T	2T	10	40	50	4
MSMHG205	Core	P	Lab work based on courses MSMHG201 & MSMHG202	4P	4P	10	40	50	4
MSMHG206	Core	P	Lab work based on courses MSMHG203 & MSMHG204	4P	4P	10	40	50	4
Total credit									24

T/P: Theory/Practical

**Semester III [Credits – 24]**

Course				Lect. Hr /week	Dur. of Exam (in H)	Marks			Credit
Course code	Type	T/P	Name			I.A.	E.T	Total	
MSMHG301	Core	T	Bioinformatics & Omics	4T	2T	10	40	50	4
MSMHG302	Core	T	Clinical Genetics & Genetic Counselling	4T	2T	10	40	50	4
MSMHG303	Core	P	Lab work based on courses MSMHG301 & MSMHG302	4T	4P	10	40	50	4
MSMHG304	GE	T	Basic Principle of Genetics	2T	1T	5	20	25	2
MSMHG305	DEC	T	Discipline-centric Elective	4T	2T	10	40	50	4
MSMHG306	DEC	P	Lab work based on courses MSMHG305	4T.	4P	10	40	50	4
MSMHG307	CE	NA	Community Engagement Or Clinical Assignments	N.A.	N.A.	5	20	25	2
Total credit									24

T/P: Theory/Practical

CE: Community Engagement Activities; DE: Discipline-centric Elective; GE: Generic elective

**\* Discipline-centric Elective (Student need to take any one of these Discipline-centric Elective)**

1. CANCER BIOLOGY (CORSE CODE: MSMHG 305-DE1)
2. INFECTION BIOLOGY (CORSE CODE: MSMHG 305-DE2)
3. MOLECULAR DIAGNOSTICS (CORSE CODE: MSMHG 305-DE3)
4. NANOBIO TECHNOLOGY (CORSE CODE: MSMHG 305-DE4)
5. STEM CELL AND REGENERATIVE MEDICINE (CORSE CODE: MSMHG 305-DE5)
6. One course of 4 credits from SWAYAM may be opted

*\*Based on the Departmental resource, student may be offered all, some or one DE for the particular session. If more than One DE courses are offered students may opt any of these DE course as per given selection criteria. New discipline-centric elective may be included, as per Departmental resource and appropriate steps. The concerned teachers and Department committee will formulate the detail content and take necessary modality for this purpose at that time.*

**\*\* CORSE CODE: MSMHG 306-DE1, 306-DE2, 306-DE3, 306-DE4, 306-DE5,**

**## Community Engagement:** Based on Discipline-centric Elective

### Semester IV [Credits – 24]

Course				Lect. Hr /week	Dur. of Exam (in H)	Marks			Credit
Course code	Type	T/P	Name			I.A.	E.T	Total	
MSMHG401	Core	T	Neurogenetics and Pharmacogenetics	4T	2T	10	40	50	4
MSMHG402	Core	T	Intellectual Property Rights, Biosafety and Bioethics	4P	2P	10	50	50	4
MSMHG403	Core	T	Project Proposal Preparation	2T	1T	10	40	50	4
MSMHG404	DE	T	Journal Club/ Communication Skills and Seminar Presentation	NA	NA	10	40	50	4
MSMHG405	DE	P	Visit of a National Lab or Biotech Industry	NA	NA	10	40	50	4
MSMHG406	Project/Term paper	NA	Dissertation and Comprehensive Viva	NA	NA	10	40	50	4
Total credit									24

#### Notes on marks distribution:

- In each course, 20% marks is allotted for Internal Assessment (for both theory and practical's), i.e., 10 marks for a paper of 50 marks and 5 marks for a paper of 25 marks.
- Marks distribution for each paper will be as follows:
  - For **40** marks of **NON-UNIT** based paper:

*Four questions (out of six) of 2 marks each, four questions (out of six) of 4 marks each and two questions (out of four) of 8 marks each are to be answered*
  - For **40** marks of **UNIT** based paper:

**UNIT I** (Total Marks 20): *Four questions (out of six) of 1 mark each, two questions (out of four) of 4 marks each and one question (out of two) of 8 marks are to be answered*

**UNIT II** (Total Marks 20): *Four questions (out of six) of 1 mark each, two questions (out of four) of 4 marks each and one question (out of two) of 8 marks are to be answered*
  - For 20 marks of **NON-UNIT** based paper:

*Four questions (out of six) of 1 mark each, two questions (out of four) of 4 marks each and one question (out of two) of 8 marks are to be answered*

# SEMESTER-I

## MSMHG-101: Core Course Transmission Genetics & Basic Human Genetics (Credit 4)

**Time: 2h**

**Full Marks: 50**

**Unit I: Transmission Genetics (Credit 2)**

**Full Marks: 25**

**Lecture hours**

- |   |   |
|---|---|
| 1. Introduction to Genetics   | 1 |
| 2. Mendelism  | 5 |
| 2.1. Mendel's and His Experiments   |   |
| 2.2. Mendel's Laws of Inheritance with examples   |   |
| 2.3. Chromosomal basis of segregation and independent assortment  |   |
| 2.4. Application of laws of probability (product rule, sum rule), Chi-square test and its application in analysis of genetic data |   |
| 3. Extensions of Mendelism  | 8 |
| 3.1. Allelic variation and gene function- Dominance relationships, basis of dominant and recessive mutations                      |   |
| 3.2. Multiple allelism, allelic series  |   |
| 3.3. Testing gene mutations for allelism: complementation test, intragenic complementation  |   |
| 3.4. Visible, sterile and lethal mutations  |   |
| 3.5. Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity, phenocopy            |   |
| 3.6. Gene interactions and modifying genes  |   |
| 3.7. Pleiotropy   |   |
| 4. Sex-linked inheritance, Linkage and crossing over  | 4 |
| 4.1. Genetic recombination and construction of genetic maps in Drosophila   |   |
| 4.2. Interference and coincidence   |   |
| 4.3. Cytological demonstration of crossing over in Drosophila   |   |
| 5. Inheritance of quantitative traits   | 4 |
| 5.1. Continuous and discontinuous variation   |   |
| 5.2. Polygenic inheritance  |   |
| 5.3. Genetic variance, heritability (narrow sense and broad sense)  |   |
| 6. Cytoplasmic inheritance, maternal effects, inheritance due to parasites and symbionts  | 3 |

### Suggested Readings

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

**Unit II: Basic Human Genetics (Credit 2)**

**Full Marks: 25**

**Lecture hours**

1. History of Human Genetics	1
2. Pedigrees- gathering family history, pedigree symbols, construction of pedigrees, presentation of molecular genetic data in pedigrees Mendel's and His Experiments	2
3. Monogenic traits	10
3.1. Autosomal inheritance-dominant, recessive	
3.2. Sex-linked inheritance	
3.3. Sex-limited and sex-influenced traits	
3.4. Mitochondrial inheritance	
3.5. MIM number	
3.6. Complications to the basic pedigree patterns- nonpenetrance, variable expressivity, pleiotropy, late onset, dominance problems, anticipation, genetic heterogeneity, genomic imprinting and uniparentaldisomy, spontaneous mutations, mosaicism and chimerism, male lethality, X-inactivation	
3.7. Risk assessment; application of Bayes' theorem	
3.8. Allele frequency in population	
3.9. Consanguinity and its effects	10
4. Complex traits	
4.1. Approaches to analysis of complex traits- 'Nature -nurture' concept, role of Family and shared environment, monozygotic and dizygotic twins and adoption studies	
4.2. Polygenic inheritance of continuous (quantitative) traits, normal growth charts, Dysmorphology	
4.3. Polygenic inheritance of discontinuous (dichotomous) traits- threshold model, liability and recurrence risk	
4.4. Genetic susceptibility in multifactorial disorders (alcoholism, diabetes mellitus, obesity)	
4.5. Estimation of genetic components of multifactorial traits: empiric risk, heritability, coefficient of relationship	
5. Basics of Pharmacogenetics and ecogenetics	2

**Suggested Readings**

1. Mange and Mange (2005). Basic Human Genetics. Sinauer Assoc
2. Gersen & Keagle (2005). The Principles of Clinical Cytogenetics. Humana
3. Connor & Smith (1997). Essentials of Medical Genetics. Blackwell
4. Davies (1993). Human Genetic Disease Analysis. IRL
5. Emery and Mueller (1992). Elements of Medical Genetics. ELBS
6. Jorde et al (2005). Medical Genetics. Elsevier
7. Korf (2006). Human Genetics. Blackwell

**MSMHG-102: Core Course  
Biochemistry & Cell Biology  
(Credit 4)**

**Time: 2h**

**Full Marks: 50**

**Unit I: Biochemistry (Credit 2)**

**Full Marks: 25**

**Lecture hours**

1. Bioenergetics	4
1.1. Second law of thermodynamics and its application	
1.2. Concept of free energy and calculations based on free energy change	
1.3. Oxidative phosphorylation: Biological oxidation-reduction reactions	
2. Protein structure and functions	5
2.1. Primary structure, peptide bond	
2.2. Secondary structure and Prediction of secondary structure, Ramachandran plot	
2.3. Tertiary structure: Forces stabilizing tertiary structure, Domains and motifs	
2.4. Quaternary structure	
3. Enzymes	5
3.1. Enzyme kinetics	
3.1.1. Lowering of activation energy	
3.1.2. Derivation of Michaelis-Menten equation, related calculations and MM & LB plots	
3.2. Mechanism of action	
3.2.1. Active site, substrate binding, transition state analogues and Abzyme	
3.2.2. Acid-base and covalent catalysis (Chymotrypsin, Carboxypeptidase)	
3.2.3. Concepts of regulation of enzyme activity	
4. Hormones	4
4.1. Characteristics	
4.2. Mechanism of action of peptide hormones	
4.3. Mechanism of action of steroid hormones and gene expression	
4.4. Hormonal regulation of fuel metabolism	4
5. Vitamins	
5.1. Structure, Dietary sources, biochemical functions, requirements of water- and lipid-soluble vitamins (vitamin B complex, C and A, D, E & K vitamins)	
5.2. Recommended dietary allowances, nutritive value, Requirement of vitamins under different physiological states- infancy, childhood, adolescence, pregnancy, lactation and ageing	
5.3. Deficiency diseases	
6. Integration of metabolic pathways	3

**Suggested Readings**

1. Nelson et al (2009). Lehninger's Principles of Biochemistry. McMillan
2. Devlin (2008). Biochemistry: with clinical correlations. Wiley
3. Berg et al (2006). Biochemistry. Freeman
4. Voet & Voet (2006). Biochemistry Vol. 1 and 2. Wiley
5. Zubay et al (1995). Principles of Biochemistry. WCB
6. 4. Rodney Boyer (2006): Concepts in Biochemistry, Wiley



## Unit II: Cell Biology (Credit 2)

**Full Marks: 25**

**Lecture hours**

1. Plasma Membrane: organization and dynamics transport across membrane; mechanisms of endocytosis and exocytosis	3
2. Endomembrane system	3
2.1. General organization of protein transport within and outside the cell	
2.2. Protein sorting and secretion	
2.3. Mechanism of intracellular digestion	
3. Cytoskeleton: Structural and functional organization	3
4. Mitochondria: Ultrastructure and Chemiosmotic theory and respiratory chain complexes	3
5. Structure and function of peroxisome	
6. Nucleolus and biosynthesis of ribosome	1
7. Cell divisions and Cell cycle regulation	1
8. Cell-Cell Interaction: Cell adhesion molecules, Cellular junctions, Extracellular matrix	3
9. Signal transduction	2
9.1. Intracellular receptor and cell surface receptors	5
9.2. Signalling via G-protein linked receptors (PKA, PKC, CaM kinase)	
9.3. Enzyme linked receptor signaling pathways	
9.4. Network and cross-talk between different signal mechanisms	
10. Programmed cell death	1

### Suggested Readings

1. Alberts et al (2008). Molecular Biology of the Cell. Garland
2. Cooper (2007). The Cell: A molecular Approach. ASM Press
3. Gilbert (2007). Developmental Biology. Sinauer
4. Jeremy et al (2002). Biochemistry. Freeman
5. Karp (2010). Cell and Molecular Biology. John Wiley
6. Lewin et al. (2009). Lewin's Genes X. Jones & Bartlett Learning, 2009
7. Lodish et al (2004). Molecular Cell Biology. Freeman
8. Pollard & Earnshaw (2002). Cell Biology. Saunders
9. Tobin and Morcel (1997). Asking about Cells. Saunders

**MSMHG-103: Core Course**  
**Computational Biology, Biostatistics & Evolutionary Genetics**  
**(Credit 4)**

**Time: 2h**

**Full Marks: 50**

**Unit I: Computational Biology, Biostatistics (Credit 2)**

**Full Marks: 25**

**Lecture hours**

- |    |   |   |
|----|---|---|
| 1. | Computational Biology:  | 4 |
|    | 1.1. Basics of computers, CPU, input and output devices, operating systems (Windows, LINUX/UNIX), GUI, flowchart and programming concept, server and grid computation, Computer networks and internet, search engine, Boolean Operators |   |
|    | 1.2. Review of Basic statistical software's and computer language: Python, MATLAB, SPSS, R-statistics   |   |
| 2. | Sampling, data and central tendency:  | 5 |
|    | 2.1. Data types, Sampling, Frequency distribution, Quartile and percentile,   |   |
|    | 2.2. Basic probability, venn diagrams, dependent probability, permutations and combinations, making decisions with probability, correlation & causality   |   |
|    | 2.3. Constructing box-plots, expected values with empirical probabilities, binomial distributions, Poisson processes, scatter plots, fitting quadratic and exponential functions to scatter plots                                       |   |
|    | 2.4. Parameters and Statistics, Mean, Median, Mode, different types of distribution, Standard deviation and error, Coefficient of variation, Skewness and Kurtosis  |   |
| 3. | Hypothesis testing  | 5 |
|    | 3.1. Parametric and Non-parametric tests, one sample hypothesis, two sample hypothesis,   |   |
|    | 3.2. Multi-sample hypothesis: The Analysis of Variance, Single factor analysis of variance, confidence limits for population mean; Power and sample size, Homogeneity of variances  |   |
| 4. | Linear regression   | 3 |
|    | 4.1. Regression vs. Correlation, Correlation coefficient, Simple linear regression equation<br>Testing the significance of relation ( $r^2$ )   |   |
| 5. | Testing for goodness of fit   | 3 |
|    | 5.1. Chi-Square goodness of fit, Heterogeneity Chi-Square, Odds ratio   |   |
|    | 5.2. Chi-Square analysis of contingency table   |   |
| 6. | Multiple comparison: Tukey test, Bonferroni and Benjamini Hochberg test, Concept of multivariate analysis   | 3 |
| 7. | Survival analysis: Concept of life tables, censored data, Estimation of survival function, Kaplan – Meier analysis.   | 2 |

**Suggested Readings**

1. Sinha P. K. and Sinha P. (2011). Computer Fundamentals. 6th ed. Bpb Publications.
2. Bailey, N. T. J. (1995). Statistical Methods in Biology. 3rd ELBS ed.
3. Forthofer, N. and Lee, E. S. (2006). Introduction to Biostatistics: A Guide to Design, Analysis and Discovery. Academic Press.
4. Gun A.M., Gupta N.K., Dasgupta B. Fundamentals of Statistics. Volume 1. World Press.
5. Selvin, S. (2004). Biostatistics: How it works? Pearson Education.

6. Sokal, R. R. and Rohlf, F. J. (1995). *Biometry: the principles and practice of statistics in biological research*. 4th ed. W. H. Freeman and Company, New York.
7. Zar J. H. (1999). *Biostatistical Analysis*. 5th ed. Pearson Education (India) Ltd.

## Unit II: Evolutionary Genetics (Credit 2)

**Full Marks: 25**

**Lecture hours**

- |  |   |
|--|---|
| 1. Concept and theories of evolution.  | 1 |
| 2. Historical introduction to evolutionary genetics.   | 1 |
| 3. Population genetics:  | 8 |
| 3.1. Microevolution in Mendelian population  |   |
| 3.1.1. Mendelian Population.   |   |
| 3.1.2. Allele frequencies and genotype frequencies   |   |
| 3.1.3. Hardy-Weinberg equilibrium and conditions for its maintenance   |   |
| 3.1.4. Factors determining the composition and change in allele and genotype frequencies.                    |   |
| 3.2. Molecular population genetics   |   |
| 3.2.1. Mutations: the source of genetic variation.   |   |
| 3.2.2. Chromosomal, DNA and allozyme polymorphism in natural population                                      |   |
| 3.2.2.1. Adaptive genetic polymorphism   |   |
| 3.2.2.2. Balanced polymorphism and heterosis   |   |
| 3.2.2.3. Genetic coadaptation and linkage disequilibrium   |   |
| 3.3. DNA sequence evolution and mechanisms for molecular evolution (neutral theory, punctuated equilibrium). |   |
| 4. Genetic markers and sequencing technologies.  | 1 |
| 5. Mapping of genes: Establishing the link between phenotype and genotype.                                   | 2 |
| 6. Evolution of genetic systems, sex chromosomes and sex determination mechanisms.                           | 2 |
| 7. Speciation processes.   | 1 |
| 8. Selfish genes: conflicts between genetic elements within an individual.                                   | 1 |
| 9. Phylogeny: methods to analyse evolutionary relatedness between populations.                               | 2 |
| 10. Domestication: changes in the genetic composition of wild animals through selective breeding.            | 1 |
| 11. Human Evolution  | 5 |
| 11.1. Hominid evolution: anatomical, Geographical, Cultural  |   |
| 11.2. Molecular phylogenetics of Homo sapiens  |   |
| 11.3. Peopling of continents (Europe, Africa, Asia)  |   |
| 11.4. Ancient DNA and human history  |   |

### Suggested Readings

1. Futuyma, Douglas J.; Kirkpatrick, Mark; (2018) *Evolution*, International fourth edition.: New York: Oxford University Press
2. Hartl & Clark (1997). *Principles of Population Genetics*. Sinaur
3. Hartl and Jones (1998). *Genetics. Principles and Analysis*. Jones and Bartlet
4. Hoelzel (1998). *Molecular Genetic Analysis of Populations*. Oxford Univ

**MSMHG-104: Core Course**  
**Molecular Biology & Bio techniques**  
**(Credit 4)**

**Time: 2h**

**Unit I: Molecular Biology (Credit 2)**

**Full Marks: 50**

**Full Marks: 25**

**Lecture hours**

- |  |   |
|--|---|
| 1. Nucleic Acids   | 5 |
| 1.1. Nature of genetic material, evidences of DNA as the genetic material  |   |
| 1.2. Structure & constitution of nucleic acids (purines, pyrimidines, nucleoside)  |   |
| 1.3. Features of double helix DNA, structure and types and composition of RNA & DNA, unusual structures                                    |   |
| 1.4. Nucleic acid chemistry: non enzymatic transformation and methylation  |   |
| 1.5. Biological role of DNA & RNA, central dogma of molecular biology.   | 7 |
| 2. Eukaryotic chromatin structure and chromosome organization  |   |
| 2.1. Histones, DNA   |   |
| 2.2. Nucleosome and higher-level organization  |   |
| 2.3. Chromosome organization   |   |
| 2.4. Metaphase chromosome: centromere and kinetochore, telomere and its maintenance; Holocentric chromosomes and supernumerary chromosomes |   |
| 2.5. Chromosomal domains (matrix, loop domains) and their functional significance  |   |
| 2.6. Heterochromatin and euchromatin, position effect variegation, boundary elements   |   |
| 2.7. Functional states of chromatin and alterations in chromatin organization  |   |
| 2.8. Structural and functional organization of interphase nucleus  |   |
| 3. Giant chromosomes: Polytene chromosomes, Lampbrush chromosomes  | 1 |
| 4. Review of Replication, Transcription and Translation: Components, General mechanism and modifications                                   | 4 |
| 5. Regulation of gene expression   | 8 |
| 5.1. Regulation of transcription initiation (Operon, Regulon, Enhancer, Promoter, Transcription factors)                                   |   |
| 5.2. Regulation by attenuation and anti-termination  |   |
| 5.3. Post transcriptional regulation   |   |
| 5.4. Mechanism of steroid hormone and stress induced gene expressions  |   |

**Suggested Readings**

1. Watson et al. (2014). Molecular Biology of the Gene. 7th Edition
2. Krebs JE, Goldstein ES and Kilpatrick ST. Lewin's Gene XII, Jones and Bartlett
3. Weaver RF Molecular Biology (2012), 5th Edition, McGraw Hill Higher Education
4. Trun&Trempey (2004). Fundamentals of Bacterial Genetics. Blackwell. 3rd Edition

**Unit I: Biotechniques (Credit 2)****Full Marks: 25****Lecture hours**

- |  |   |
|--|---|
| 1. Basic knowledge of principles and applications of the following microscopy techniques   | 6 |
| 1.1. Light Microscopy  |   |
| 1.2. Dark-field & Phase-contrast Microscopy  |   |
| 1.3. Fluorescence and Confocal Microscopy  |   |
| 1.4. Electron Microscopy   |   |
| 1.5. Photography, Digital imaging and image Processing   |   |
| 2. Biochemical techniques  | 8 |
| 2.1. Centrifugation: types of rotors, clinical, highspeed and ultracentrifuges   |   |
| 2.2. Colorimetry and spectrophotometry: Beer-Lambert law; absorption spectrum, fluorescence spectrum, introduction to mass spectrometry.   |   |
| 2.3. Electrophoresis & blotting: Principle and methods   |   |
| 2.4. Basic of Histology and Histochemistry (Immunohistochemistry & Immunofluorescence)   |   |
| 2.5. Chromatography: Paper, Thin layer chromatography; Columns: ion-exchange, gel-filtration, HPLC and affinity columns, electrophoresis   |   |
| 2.6. Tracer techniques: Properties and units of radioactivity; half-life; measurement of radioactivity by GM counter, liquid scintillation counter; autoradiography; radio-immunoassay; safety rules in handling of radioisotopes and hazardous chemicals. |   |
| 3. Techniques in human gene and chromosome analysis  | 8 |
| 3.1. Human karyotype: banding, nomenclature of banding   |   |
| 3.2. Fluorescence in situ hybridization (FISH)   |   |
| 3.3. Comparative genomic hybridization   |   |
| 3.4. DNA sequencing (Sanger, NGS): Types and application   |   |
| 4. Introduction to nanotechnology  | 1 |
| 5. Introduction to OMICS   | 2 |

**Suggested Readings**

1. Mange and Mange (2005). Basic Human Genetics. Sinauer Assoc
2. Boyer 2007 (2007). Concept of Biochem. 3rd Ed
3. Keith Wilson and John Walker. Principles and Techniques of Biochemistry and Molecular Biology
4. Locquin and Langeron (1983). Handbook of Microscopy. Butterworths

**PRACTICAL PAPERS**

<b>MSMHG105</b>	<b>Lab work based on courses MSMHG101 &amp; MSMHG102</b>	<b>Credits: 2</b>
<b>MSMHG106</b>	<b>Lab work based on courses MSMHG103 &amp; MSMHG104</b>	<b>Credits: 2</b>

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# SEMESTER-II

## MSMHG-201: Core Course Reproductive Biology & Developmental Genetics (Credit 4)

**Time: 2h**

**Unit I: Reproductive Biology (Credit 2)**

**Full Marks: 50**

**Full Marks: 25**

**Lecture hours**

1. Male and female reproductive systems	4
1.1. Gonads and differentiation of sexual characters	
1.2. Hormonal regulation of sexual differentiation	
2. Gametogenesis and Embryonic development	4
2.1. Differentiation of germ cells and gametogenesis	
2.2. Fertilization and implantation	
2.3. Stages of human embryonic development	
3. Reproductive disorders	6
3.1. Disorders of gonads, genital tracts and genitalia	
3.1.1. Pseudohermaphroditism	
3.1.2. True hermaphroditism	
3.1.3. Gonadal dysgenesis	
3.1.4. Anomalies of genital ducts	
3.2. Infertility	
3.2.1. Genetic basis of male infertility	
3.2.2. Genetic basis of female infertility	
3.3. Genetics of Pregnancy loss	
3.4. Congenital malformations and teratogenesis	
4. Technologies in reproductive assistance	5
4.1. Types of ART	
4.2. Cryopreservation of gametes and embryo	
4.3. Ethical, legal and emotional implications	
4.4. Gamete antigens in relation to fertility	
5. Prenatal and pre-implantation diagnosis	6
5.1. Indications for prenatal diagnosis	
5.2. Indications for chromosomal testing	
5.3. Noninvasive methods	
5.4. Invasive methods	
5.5. Legal and ethical considerations	

### Suggested Readings

1. Rimon et al (2002) Principles and Practice of Medical Genetics, Vol I-III.
2. Martin H. Johnson & Barry Everitt. Essential reproduction.
3. Ramon Pinon. Biology of Human Reproduction.
4. Y.W. Loke and Ashley King. Human Implantation: Cell Biology and Immunology.
5. Jonathan S Berek .Berek and Novak's Gynecology.
6. 3. FG Cunningham. Williams Obstetrics

**Unit II: Developmental Genetics (Credit 2)**

**Full Marks: 25**  
**Lecture hours**

- 1. Early development 5
  - 1.1. 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. Genetics of pattern formation 10
  - 2.1. Caenorhabditis: Vulva formation
  - 2.2. Drosophila.
    - 2.2.1. Maternal genes and formation of body axes.
    - 2.2.2. Segmentation genes
    - 2.2.3. Homeotic gene's function
    - 2.2.4. Imaginal disc development.
  - 2.3. Vertebrates
    - 2.3.1. Axes formation and HOX genes
    - 2.3.2. Limb formation in chick
  - 2.4. Programmed rearrangements in genes: Chromatin diminution, Endoreplication cycle, Gene amplification.
- 3. Genetic determination of sex in Caenorhabditis, Drosophila and mammals 3
- 4. Epigenetics in development 3
- 5. Regeneration and Senescence 2
- 6. Embryonic stem cells and their applications 2

**Suggested Readings**

- 1. Alberts et al (2007). Molecular Biology of The Cell. Garland
- 2. Gilbert (2006). Developmental Biology. Sinauer
- 3. Kalthoff (1996). Analysis of Biological Development. McGraw Hill
- 4. Lewin et al. (2009). Lewin's Genes X. Jones & Bartlett Learning, 2009
- 5. Monk (1987). Mammalian Development – A Practical Approach. IRL
- 6. Wolpert (2007). Principles of Development. Oxford

**MSMHG-202: Core Course**  
**Immunology & Immunogenetics**  
**(Credit 4)**

**Time: 2h**

**Unit I: Immunology (Credit 2)**

**Full Marks: 50**  
**Full Marks: 25**  
**Lecture hours**

- 1. General introduction to immune system: Innate and adaptive immunity, Immune responses, specialized immune system. 4
- 2. Components of innate and acquired immunity; phagocytosis; complement and inflammatory responses; pathogen recognition receptors (PRR) and pathogen associated molecular pattern (PAMP); innate immune response; antigens: immunogens, haptens; 4

- |   |        |
|---|--------|
| 3. Generation and regulation of immune responses  | 5      |
| 3.1. Cell mediated immunity   |        |
| 3.1.1. Antigen processing and presentation  |        |
| 3.1.2. MHC-restriction  |        |
| 3.1.3. T Cell Maturation, activation and differentiation  |        |
| 3.1.4. B Cell Generation, Activation and differentiation  |        |
| 3.1.5. Clonal selection and immunological memory  |        |
| 3.2. Humoral Immunity   |        |
| 3.2.1. Antibody: types, structure, function, production   |        |
| 3.2.2. Antigen antibody interactions  |        |
| 3.3. Cytokines  |        |
| 4. Basic and advanced Immunological Techniques: Principles and modalities of Immunodiffusion techniques, Agglutination, ELISPOT, RIA, ELISA, Western blotting, Immunoprecipitation, Immuno-histochemistry, and Flow-cytometry   | 7<br>5 |
| 5. Dynamics of the immune response in health and disease  |        |
| 5.1. Immunotherapeutics and Drug targeting  |        |
| 5.2. Transplantation immunology   |        |
| 5.3. Vaccine: Active and passive immunization; live, killed, attenuated, subunit vaccines; vaccine technology: role and properties of adjuvants, recombinant DNA and protein-based vaccines, plant-based vaccines, reverse vaccinology; peptide vaccines, conjugate vaccines; |        |

### Suggested Readings

1. Janeway & Traver's Immunobiology (2016), Taylor & Francis
2. Abbas et al, Cellular and Molecular Immunology (2017), Saunders
3. Barrett, Text Book of Immunology (1988), Mosloy,
4. Benjamin et al, Immunology – A Short Course (2012), Wiley-Liss
5. Kuby, Immunology (2013), MacMillan,
6. Roitt, Essential Immunology (2017), Blackwell,

### Unit II: Immunogenetics (Credit 2)

**Full Marks: 25**  
**Lecture hours**

- |  |    |
|--|----|
| 1. Immune response proteins: genetic basis of structure and diversity        | 4  |
| 2. Immunoglobulin gene superfamily   | 10 |
| 2.1. B cell receptor genes (Immunoglobulin)                                  |    |
| 2.1.1. Organization of Ig gene loci  |    |
| 2.1.2. Genetic basis of antibody diversity                                   |    |
| 2.1.2.1. Somatic recombination: V(D)J recombination and junctional diversity |    |
| 2.1.2.2. Somatic hypermutation   |    |
| 2.1.2.3. Allelic exclusion   |    |
| 2.1.2.4. Class switching   |    |
| 2.1.3. Regulation of Ig gene transcription                                   |    |
| 2.2. Organization of TCR gene loci and genetic basis of TCR diversity        |    |
| 2.3. Organization of HLA locus: genetic polymorphism and HLA haplotypes      |    |
| 3. Disorders of Human Immune System  | 4  |
| 3.1. Immunological Tolerance and Autoimmune diseases                         |    |
| 3.2. Allergy and hypersensitivity  |    |



- 3.3. Cytokine-related diseases
- 3.4. Immunodeficiencies
- 4. Cancer immunology 3
- 5. Antibody genes and antibody engineering: chimeric, generation of monoclonal antibodies, hybrid monoclonal antibodies; catalytic antibodies and generation of immunoglobulin gene libraries, 4

### Suggested Readings

- 7. Janeway & Traver's Immunobiology (2016), Taylor & Francis
- 8. Abbas et al, Cellular and Molecular Immunology (2017), Saunders
- 9. Barrett, Text Book of Immunology (1988), Mosloy,
- 10. Benjamin et al, Immunology – A Short Course (2012), Wiley-Liss
- 11. Kuby, Immunology (2013), MacMillan,
- 12. Roitt, Essential Immunology (2017), Blackwell,

## **MSMHG-203: Core Course Molecular Human Genetics & Human Genome (Credit 4)**

**Time: 2h**

**Full Marks: 50  
Lecture hours**

- 1. Genetic mapping of Mendelian traits 3
  - 1.1. Identifying recombinants and nonrecombinants in pedigrees
  - 1.2. Genetic and physical map distances
  - 1.3. Genetic markers
  - 1.4. Two-point mapping- LOD score analysis
  - 1.5. Multipoint mapping
  - 1.6. Homozygosity mapping.
- 2. Genetic mapping of complex traits 3
  - 2.1. Difficulties in mapping complex traits
  - 2.2. Allele sharing methods- Affected sib pair analysis
  - 2.3. Allelic association, Linkage disequilibrium mapping, Transmission
  - 2.4. disequilibrium test
- 3. Physical mapping methods 3
  - 3.1. Low resolution mapping- Cell hybrids, mini- and microcells, synteny of genes,
  - 3.2. Radiation hybrid mapping
  - 3.3. Assembly of clone contigs
  - 3.4. Identifying genes in cloned DNA
  - 3.5. Integration of cytogenetic, genetic and physical maps 3
- 4. Identifying human disease genes
  - 4.1. Principles and strategies
  - 4.2. Position-independent and positional cloning, Candidate gene approaches,
  - 4.3. Whole Genome and Exome Sequencing
  - 4.4. Confirming a candidate gene: mutation screening, testing in animal models.
- 5. Molecular pathology 3
  - 5.1. Nomenclature of mutations and their databases
  - 5.2. Loss-of-function and gain-of-function mutations in diseases
  - 5.3. Instability of the human genome: Pathogenicity associated with repeat sequences
- 6. DNA testing 3
  - 6.1. Direct testing.
    - 6.1.1. Screening for unknown mutations

- 6.1.2. Detection of known mutations.
- 6.2. Indirect testing – gene tracking
- 6.3. DNA profiling: establishing identity and relationships
- 6.4. Population screening - ethics, organization and advantages. 6
- 7. Human Genome
- 7.1. The Genome projects
- 7.2. Organization of human genome: Mitochondrial genome, Gross base composition of nuclear genome, Gene density, CpG islands, RNA-encoding genes, functionally identical/similar genes, Diversity in size and organization of genes, Annotation
- 7.3. Gene families: Multigene families, Gene superfamilies, Gene families in clusters, Pseudogenes, Repetitive DNA and transposable elements
- 7.4. Comparative Genomics: Comparative genomics as an aid to gene mapping and study of human disease genes
- 7.5. Disease and genomics

### Suggested Readings

1. Strachan & Read (2011). Human Molecular Genetics. Garland Edition. 4th Edition.
2. Pasternak (2005). An Introduction to Molecular Human Genetics. Fritzgerald. 2nd Edition.
3. Mange and Mange (1999). Basic Human Genetics. Sinauer Assoc. 2nd Edition.
4. Lewis (2007). Human Genetics. WCB & McGraw. 7th Edition.
5. Sudbery (2010). Human Molecular Genetics. Prentice-Hall. 3rd Edition.
6. Brown (2007). Genomes. Bios
7. 4. Coleman and Tsongalis (1997). Molecular Diagnosis. Humana
8. 5. Dale & Scharz (2003). From Genes to Genomes. Wiley
9. 6. Hawley and Mori (1999). The Human Genome. Academic

## **MSMHG-204: Core Course** **Genetic Engineering & Model Genetic Systems (Credit 4)**

**Time: 2h**

**Full Marks: 50**

**Lecture hours**

- |   |   |
|---|---|
| 1. Impact of genetic engineering in modern society;   | 2 |
| 2. General requirements for performing a genetic engineering experiment   | 8 |
| 2.1. Enzymes: Restriction endonucleases and methylases; DNA ligase, Klenow enzyme, T4 DNA polymerase, polynucleotide kinase, alkaline phosphatase; cohesive and blunt end ligation; linkers; adaptors;  |   |
| 2.2. Cloning and expression vectors: Plasmids; Bacteriophages; M13 mp vectors; PUC19 and Bluescript vectors, hagemids; Lambda vectors; Insertion and Replacement vectors; Cosmids; Artificial chromosome vectors (YACs; BACs); Principles for maximizing gene expression expression vectors; pMal; GST; pET-based vectors; Protein purification; His-tag; GST-tag; MBP-tag etc.; Intein-based vectors; Inclusion bodies; methodologies to reduce formation of inclusion bodies; mammalian expression and replicating vectors; Baculovirus and Pichia vectors system, plant based vectors, Ti and Ri as vectors, yeast vectors, shuttle vectors. |   |
| 3. Screening and characterization of clones   | 5 |
| 3.1. Preparation of probes, homopolymeric tailing; labelling of DNA: nick translation, random priming, radioactive and non-radioactive probes,  |   |
| 3.2. Principles of hybridizations and hybridization-based techniques (Colony, plaque, northern, southern, south-western and far-western and colony hybridization, fluorescence in situ hybridization),  |   |

- 3.3. Expression based screening;
- 3.4. Interaction based screening: yeast two-hybrid system
- 4. Basic Principles and Applications of the following techniques 8
  - 4.1. Principles of PCR: primer design; fidelity of thermostable enzymes; DNA polymerases; types of PCR – multiplex, nested; reverse-transcription PCR, real time PCR, touchdown PCR, hot start PCR, colony PCR, asymmetric PCR, cloning of PCR products; T-vectors; proof reading enzymes; PCR based site specific mutagenesis; PCR in molecular diagnostics; viral and bacterial detection;
  - 4.2. Sequencing methods; enzymatic DNA sequencing; chemical sequencing of DNA; automated DNA sequencing; RNA sequencing; chemical synthesis of oligonucleotides; Next generation gene sequencing (NGS)
  - 4.3. Mutation detection: SSCP, DGGE, RFLP.
  - 4.4. Gene transfer techniques: Insertion of foreign DNA into host cells; transformation, electroporation, transfection;
- 5. Cloning strategies: cDNA and genomic libraries, expression and interaction-based cloning, positional cloning 12
  - 5.1. Construction of libraries; isolation of mRNA and total RNA; reverse transcriptase and cDNA synthesis; cDNA and genomic libraries; construction of microarrays – genomic arrays, cDNA arrays and oligo arrays;
  - 5.2. Study of protein-DNA interactions: electrophoretic mobility shift assay; DNase footprinting; methyl interference assay, chromatin immunoprecipitation; protein- protein interactions using yeast two-hybrid system; phage display.
- 6. Gene silencing techniques; introduction to siRNA; siRNA technology; Micro RNA; construction of siRNA vectors; principle and application of gene silencing; gene knockouts and gene therapy; creation of transgenic plants; debate over GM crops; 6
- 7. Introduction to methods of genetic manipulation in different model systems e.g. fruit flies (*Drosophila*), worms (*C. elegans*), frogs (*Xenopus*), fish (zebra fish) and chick; 5
- 8. Transgenics - gene replacement; gene targeting; creation of transgenic and knock-out mice; disease model; introduction to genome editing by CRISPR-CAS with specific emphasis on Chinese and American clinical trials. 4

### Suggested Readings

1. Ausubel et al (2002). Short Protocols in Molecular Biology. Wiley
2. Brown (2000). Essential Molecular Biology VI. AP
3. Brown (2000). Essential Molecular Biology VII. AP
4. Brown (2006). Gene Cloning and DNA Analysis - An Introduction. Blackwell
5. Glick and Pasternak (2003). Molecular Biotechnology. ASM Press
6. Kracher. Molecular Biology - A Practical Approach.
7. Krenzer and Massey (2000). Recombinant DNA and Biotechnology. ASM
8. Micklos and Freyer (1990). DNA Science. CSHL
9. Primrose (2001). Molecular Biotechnology. Panima
10. Robertson et al (1997). Manipulation & Expression of Recombinant DNA. AP
11. Sambrook et al (2001). Molecular Cloning. CSHL
12. Twyman (1999). Advanced Molecular Biology. Viva
13. Watson et al (1992). Recombinant DNA. Freeman
14. Primrose and Twyman (2006). Principles of Gene Manipulation and Genomics. Blackwell

### PRACTICAL PAPERS

<b>MSMHG205</b>	<b>Lab work based on courses MSMHG201 &amp; MSMHG202</b>	<b>Credits: 2</b>
<b>MSMHG206</b>	<b>Lab work based on courses MSMHG203 &amp; MSMHG204</b>	<b>Credits: 2</b>

# SEMESTER-III

## MSMHG-301: Core Course Bioinformatics & Omics (Credit 4)

**Time: 2h**

**Unit I: Bioinformatics (Credit 2)**

**Full Marks: 50**

**Full Marks: 25**

**Lecture hours**

- 1. Introduction to primary Databases:** Types of Biological data- Genomic DNA, cDNA, rDNA, ESTs, GSSs; Primary Databases -Nucleotide sequence databases-GenBank, EMBL, DDBJ, Protein Sequence Databases- UniProtKB, UniProt, TrEMBL, Swiss-Prot, UniProt Archive-UniParc, UniProt Reference Clusters-UniRef, UniProt Metagenomic and Environmental Sequences-UniMES. Literature Databases- PubMed, PLoS, BioMed Central. **3**
- 2. Introduction to Secondary or Derived Databases-** PDB, CSD, MMDB, SCOP, CATH, FSSP, CSA, KEGG ENZYME, BRENDA; Sequence motifs Databases:-Prosite, ProDom, Pfam, InterPro; Composite Databases-NRDB, Genome Databases- Viral genome database (ICTV db), Bacterial Genome database (GOLD, MBGD), Organism specific database (OMIM/OMIA, SGD, WormBase, PlasmODB, FlyBase, TAIR), Genome Browsers (Ensembl, VEGA, NCBI map viewer, UCSC Genome Browse). Bioinformatics Database search engines: -Text-based search engines (Entrez, DBGET/LinkDB) **3**
- 3. File formats, sequence patterns and profiles:** Sequence file formats – GenBank, FASTA, ALN/ClustalW2, PIR; Basic concept and definition of sequence patterns, motifs and profiles, various types of pattern representations viz. consensus, regular expression (Prosite-type) and sequence profiles; Sequence similarity based search engines (BLAST and FASTA); Pattern based search using MeMe and PRATT); Motif-based search using ScanProsite and eMOTIF; Profile-based database searches using PSI-BLAST and HMMer. **3**
- 4. Sequence Analysis and predictions:** Nucleic acid sequence analysis- Reading frames; Codon Usage analysis; Translational and transcriptional signals, Splice site identification, Gene prediction methods and RNA fold analysis; Protein sequence analysis-Compositional analysis, Hydrophobicity profiles, Amphiphilicity detection, Moment analysis, Transmembrane prediction methods, Secondary structure prediction methods. **4**
- 5. Sequence Analysis –** Basic concepts of sequence similarity, identity and homology, definitions of homologues, orthologues, paralogues and xenologues. Scoring matrices: basic concept of a scoring matrix, Matrices for nucleic acid and proteins sequences, PAM and BLOSUM series, matrix derivation methods and principles. Pairwise sequence alignment – Basic concepts of sequence alignment, gap penalties, Needleman and Wunsch, Smith and Waterman algorithms for pairwise alignments and application in Nucleic acid and protein sequences alignments. Multiple sequence alignments (MSA) –The need for MSA, basic concepts of various approaches for MSA (e.g. progressive, hierarchical etc.). Algorithm of CLUSTALW and PileUp and application, concept of dendrogram and its interpretation, Use of HMM-based Algorithm for MSA (e.g. SAM method). **5**
- 6. Protein modelling:** Introduction to protein modelling; force field methods; (Molecular Mechanics), Conformational Space, Molecular Dynamics (MD), Metropolis Monte Carlo (MC); energy minimization methods, Applying Ramachandran Plot constraints. Buried and **4**

exposed residues and water-of-hydration; side chains and neighbours; hydrogen bonds; mapping electrostatic properties onto surfaces; RMS fit of conformers; assigning secondary structures; Protein structure prediction: protein folding and model generation

7. **Computational biology in drug design:** Computational Structural Biology with application to Drug Design; Methods and applications that use computation to model biological systems and human diseases; Signalling and gene-regulatory networks; in silico cell and tissue models; Neural networks & artificial intelligence models; Self-emergent behaviour in physical systems: comparison to biological systems. Liver function alterations during disease – Jaundice, Cirrhosis, Tumors, Reye Syndrome, Drug- and Alcohol-Related Disorders 3

### Suggested Readings

1. Bioinformatics: Sequence and Genome Analysis by Mount D., Cold Spring Harbor Laboratory Press, New York. 2004
2. Bioinformatics- a Practical Guide to the Analysis of Genes and Proteins by Baxevanis, A.D. and Francis Ouellette, B.F., Wiley India Pvt Ltd. 2009
3. Introduction to Bioinformatics by Teresa K. Attwood, David J. Parry-Smith. Pearson

### Unit II: OMICS Study (Credit 2)

**Full Marks: 25**  
**Lecture hours**

- |  |   |
|--|---|
| 1. Introduction to Omics   | 1 |
| 2. Data management   | 2 |
| 2.1. Technology Awareness - Computer clusters (HPC), Super computers, Cloud computing, Storage platforms, Network Attached Storage (NAS); Programming for High Performance Computing - Introduction to Programming languages (Perl and/or Python); Introduction to R;  |   |
| 2.2. Data Awareness -Databases - NCBI, GEO, UCSC Browser;  |   |
| 2.3. High-volume data and its management, Data size, format and type, policies – IPR and ethics, Data Compression and archiving strategies.  |   |
| 3. Introduction to genomics: Structure and organization of prokaryotic and eukaryotic genomes- nuclear, mitochondrial and chloroplast genomes; Computational analysis, Databases, Finding genes and regulatory regions.  | 3 |
| 4. Microarray technology   |   |
| 4.1. Introduction, Basic principles and design,  | 4 |
| 4.2. DNA microarray, cDNA and oligonucleotide arrays   |   |
| 4.3. Instrumentation and structure   |   |
| 4.4. Designing a microarray experiment - The basic steps   |   |
| 4.5. Types of microarrays - expression arrays, protein arrays, Comparative Genomic Hybridization (CGH) arrays, Resequencing arrays; Different platforms (Affymetrix, Agilent etc.);  |   |
| 4.6. Data Processing and Normalization - Algorithms of data processing and Normalization; Tools used to normalize; Microarray databases – NCBI; GEO (Gene Expression Omnibus), Array Expres (EBI); Functional Analysis: Differential gene expression; Gene Ontology functional enrichment tools, Pathway analysis (KEGG Database); |   |
| 4.7. Applications of Microarray technology; case studies   |   |
| 5. Highthroughput Sequencing technologies: Genomic and transcriptome analysis by NGS   |   |
| 5.1. Introduction to NGS, Platform overview and comparison (Illumina, 454 (Roche), SOLiD (Life technology), Specific Biosciences, Ion Torrent, Nanopore, PacBio;   | 7 |
| 5.2. Types of NGS, DNA-sequencing - Whole genome sequencing, exome sequencing, Deep sequencing, ChIP sequencing, RNA-sequencing and the types (small RNA sequencing,   |   |

- non-coding RNA sequencing), Whole transcriptome sequencing
- 5.3. Data Processing and Analysis: Data Quality Check, filtering and Genome assembly and mapping to reference genomes, mapping tools (bowtie, maqetc.),
  - 5.4. Sequence Alignment formats: Sequence Alignment/Map (SAM) format, Binary Alignment/Map (BAM) format,
  - 5.5. Functional Analysis: Pathway analysis, Gene Ontology analysis; Application of different sequencing technique, methylomics, in vivo protein binding, genome wide association studies (GWAS), Histone modification, microbial sequencing, Comparison of Microarray technology and High throughput sequencing technology, case studies
6. Proteomics 6
    - 6.1. Overview of protein structure-primary, secondary, tertiary and quaternary structure
    - 6.2. Relationship between protein structure and function; Outline of a typical proteomics experiment
    - 6.3. Identification and analysis of proteins by 2D analysis, Spot visualization and picking; Tryptic digestion of protein and peptide fingerprinting
    - 6.4. Mass spectrometry: ion source (MALDI, spray sources), analyzer (ToF, quadrupole, quadrupole ion trap) and detector;
    - 6.5. Post translational Modifications: Quantitative proteomics, clinical proteomics and disease biomarkers, mass spectral tissue imaging and profiling;
    - 6.6. Protein-protein interactions: Surfaceomes and Secretomes, Solid phase ELISA, pull-down assays (using GST-tagged protein) tandem affinity purification, far western analysis, by surface plasmon resonance technique; Yeast two hybrid system, Phage display, Protein interaction maps,
    - 6.7. Protein arrays-definition; applications- diagnostics, expression profiling.
  7. Metabolomics: Introduction and overview of metabolites, applications of non-tracer and tracer-based techniques, application of metabolomics 2

### Suggested Readings

1. Sara Goodwin, John D. McPherson, and Richard McCombie. (2016) Coming of Age: Ten Years of Next-generation Sequencing Technologies. Nature Reviews 17: 349.
2. Michael L. Metzker. (2010) Sequencing Technologies – the Next Generation. Nature Reviews 11: 31.
3. James Galagan, Anna Lyubetskaya and Antonio (2013), Gomes. ChIP-seq and the Complexity of Bacterial Transcriptional Regulation. Current topics in microbiology and immunology 363: 43-68
4. A Tutorial Review of Microarray Data Analysis. Sorin Drăghici. (2012) Statistics and Data Analysis for Microarrays Using R and Bioconductor, 2nd Edition. Chapman and Hall/CRC.
5. Huang da W, Sherman BT, Lempicki RA. (2009); Systematic and Integrative Analysis of Large Gene Lists using DAVID Bioinformatics Resources. Nat Protoc. 4(1):44-57.

## **MSMHG-302: Core Course Clinical Genetics & Genetic Counselling (Credit 4)**

**Time: 2h**

**Unit I: Clinical Genetics (Credit 2)**

**Full Marks: 50**

**Full Marks: 25**

**Lecture hours**

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. Genome imprinting Syndromes: Prader-Willi & Angelman syndromes, Beckwith- 1

Wiedeman Syndrome	
4. Neurofibromatosis I	1
5. Disorders of muscle	3
5.1. Dystrophies (Duchenne Muscular dystrophy and Becker Muscular Dystrophy)	
5.2. Myotonias	
5.3. Myopathies	
6. Disorders of Haemopoitic systems	4
6.1. Overview of Blood cell types and haemoglobin	
6.2. Sickle cell anemia	
6.3. Thalassemias	
6.4. Hemophilias	
7. Disorders of eye	3
7.1. Retinitis pigmentosa	
7.2. Cataract	
7.3. Glaucoma	
7.4. Colour blindness	
8. Multifactorial diseases	3
8.1. Hyperlipidemia	
8.2. Atherosclerosis	
8.3. Diabetes mellitus	
9. Mitochondrial syndromes	1
10. Inborn errors of Metabolism and their genetic basis	3
10.1. Phenylketonuria	
10.2. Maple Syrup urine syndrome	
10.3. Mucopolysaccharidosis	
10.4. Galactosemia	
11. Genetic disorders in skeleton and skin	1
12. Management of genetic disorders	1

### Suggested Readings

1. Roderick R. McInnes and Huntington F. Willard (2007). Thompson & Thompson Genetics in Medicine.
2. Cox and Sinclair (1997). Molecular Biology in Medicine. Blackwell
3. DeGrouchy and Turleau (1984). Clinical Atlas on Human Chromosomes. Wiley
4. Jankowski and Polak (1996). Clinical Gene Analysis and Manipulation. Cambridge

### Unit II: Genetic Counselling (Credit 2)

**Full Marks: 25**

**Lecture hours**

1. Historical overview of genetic counseling	3
1.1. Models of Eugenic, Medical/Preventive, Decision making, Psychotherapeutic counseling; current definition and goals	
1.2. Philosophy and ethos of genetic services and counseling	7
2. Components of genetic counseling	
2.1. Indications and purpose	
2.2. Information gathering and construction of pedigrees	
2.3. Medical Genetic evaluation	
2.3.1. Basic components of Medical History	
2.3.2. Past medical history, social & family history	
2.4. Physical examination.	

- 2.4.1. General and dysmorphology examination
- 2.4.2. Documentation
- 3. Patterns of inheritance, risk assessment and counseling in common Mendelian and multifactorial disorders 3
- 4. Biochemical and Molecular genetic tests 6
  - 4.1. In Children
  - 4.2. Pre-symptomatic testing for late onset diseases (predictive medicine)
- 5. Bioethics in research 6
  - 5.1. Bioethics in cloning and stem cell research
  - 5.2. Bioethics in human and animal experimentation, animal rights/welfare

### Suggested Readings

1. Baker et al (1998). A Guide to Genetic Counseling. Wiley
2. Harper (2001). Practical Genetic Counseling. Arnold
3. Rose & Lucassen (1999). Practical genetics of primary care. Oxford
4. Young (1999). Introduction to Risk Calculation in Genetic Counseling. Oxford

## PRACTICAL PAPERS

**MSMHG303    Lab work based on courses MSMHG301 and MSMHG302                      Credits: 2**

### MSMHG-304: General Elective Basic Principles of Genetics (Credit 2)

**Time: 2h**

**Full Marks: 25  
Lecture hours**

1. Scope of Genetics 2
2. Transmission Genetics: genotype, phenotype, chromosomal basis of inheritance (meiosis, segregation, independent assortment, linkage and crossing over) 4
3. Nature and flow of genetic information 5
  - 3.1. DNA and RNA: chemical composition and secondary structure
  - 3.2. Proteins and their roles as structural and catalytic molecules in biological organization
  - 3.3. Replication and transcription
  - 3.4. Genetic code and translation
4. Concept of gene 3
5. Regulation of gene activity 7
  - 5.1. Differential gene activity and cell differentiation
  - 5.2. Positive and negative regulation of gene activity
  - 5.3. General concepts of transcriptional and post-transcriptional of gene activity
6. Genes and diseases (common inherited disorders in man, cancer) 2
7. Human Genome 2

### Books Recommended

1. Griffith et al: An Introduction to Genetic Analysis, Freeman, 2004
2. Hartl & Jones: Essential Genetics: A Genomic Perspective, Jones & Bartlet, 2002
3. Russell: Genetics, Benjamin Cummings, 2002
4. Snustad & Simmons: Principles of Genetics, John Wiley, 2003



**MSMHG-305: Discipline Centric Elective  
(Credit 4)**

**Time: 2h**

**Full Marks: 50  
Lecture hours**

**\*Discipline-centric Elective (Student need to take any one of these Discipline-centric Elective)**

1. **Cancer Genomics** (CORSE CODE: MSMHG 305-DE1)
2. **Infection Biology** (CORSE CODE: MSMHG 305-DE2)
3. **Molecular diagnostics** (CORSE CODE: MSMHG 305-DE3)
4. **Nanobiotechnology** (CORSE CODE: MSMHG 305-DE4)
5. **Stem cell and regenerative medicine** (CORSE CODE: MSMHG 305-DE5)
6. One course of 4 credits from **SWAYAM** may be opted

*\*Based on the Departmental resource, student may be offered all, some or one DE for the particular session. If more than One DE courses are offered students may opt any of these DE course as per given selection criteria. New discipline-centric elective may be included, as per Departmental resource and needs.*

**PRACTICAL PAPERS**

**MSMHG306 Lab work based on courses MSMHG305**

**Credits: 2**

**MSMHG- 307: Community Engagement (CE)  
(Credit 2)**

**Full Marks: 25**

**COMMUNITY OUTREACH:** *Students will undertake any community service or clinical assignments based on their Discipline centric elective*

**SEMESTER IV**  
**MSMHG-101: Core Course**  
**Neurogenetics and Pharmacogenetics**  
**(Credit 4)**

**Time: 2h**

**Unit I: Neurogenetics (Credit 2)**

**Full Marks: 50**

**Full Marks: 25**

**Lecture hours**

1. Nervous system	4
1.1. Major regions of human brain	
1.2. Cellular components of nervous tissue	
1.3. Sub cellular organization of the nervous system	
1.4. Membrane potential and action potential.	
1.5. Neurotransmitters	6
2. Genetic Aspect of Learning and memory	
2.1. Classification of different types of memory	
2.2. Models for understanding learning and memory	
2.3. Molecular basis of short time and longtime sensitization	
3. Circadian rhythms	3
3.1. Invertebrates (Drosophila)	
3.2. Mammals	
3.3. Clinical Implication	
4. Neurogenetic disorders	5
4.1. Syndromes due to triplet nucleotide expansion	
4.2. Alzheimers disease	
4.3. Parkinsons disease	
5. Nature-nurture and behaviour	5
5.1. Genetic experiments to investigate animal behaviour	
5.1.1. Selection studies	
5.1.2. Inbred strain studies	
5.2. Identifying genes for controlling behavior	
5.2.1. Induced mutations	
5.2.2. Quantitative trait loci	
5.2.3. Synteny/orthology	
5.3. Investigating the genetics of human behavior	
5.3.1. Twin and adoption study designs, interpreting heritability	
5.3.2. Linkage and association studies	
5.4. Environmental influence- shared and non-shared environment	
6. Genetics of Psychiatric diseases	2
6.1. Schizophrenia	
6.2. Mood disorders	
6.3. Disorders of childhood.	

**Suggested Readings**

1. Kaplan and Sadock (2014). Synopsis of Psychiatry. Williams & Wilkins. 11th Edition.
2. Plomin et al (2012). Behavioral Genetics. Worth. 6th Edition.
3. Zigmond, Bloom et al., (2012). Fundamentals Neuroscience. Academic Press. 4th Edition.
4. Kandel, Schwartz et al. (2012). Principles of Neuroscience. McGraw Hill. 5th Edition.
5. Anholt and Mackay (2010). Principles of Behavioural Genetics. Elsevier Press.

## Unit II: Pharmacogenomics (Credit 2)

**Full Marks: 25**

**Lecture hours**

- |   |   |
|---|---|
| 1. Introduction to Pharmacogenetics and ecogenetics   | 2 |
| 2. Personalized medicine: future scope : Recent advances in human molecular genetics paving ways towards potential application of personalised therapies/medicines: pharmacogenomics/drug metabolism in relation to individual genetic makeup. biology and medicine | 7 |
| 3. Pharmacogenomics from human genome: Major worldwide, Genome wide association studies (GWAS) and potential use of individual-specific genomic features that impact disease relevance and treatment modalities   | 6 |
| 4. Optogenetics and Pharmacogenetics in neuroscience  | 2 |
| 5. Cancer pharmacogenomics & biomarker discovery and development  | 8 |
| 5.1. Pharmacogenomics; Cancer pharmacogenomics in children; Active ADR surveillance and future directions;  |   |
| 5.2. Uses of biomarkers in cancer research and cancer care; Biomarker discovery and qualification;  |   |
| 5.3. Assay validation, clinical validity, clinical utility; Incorporation of biomarkers into clinical trial design  |   |

### Suggested Readings

1. Lewis (2006). Human Genetics. WCB
2. Maroni (2001). Molecular and Genetic Analysis of Human Traits. Blackwell
3. 8.Nussbaum et al (2004). Genetics in Medicine. Saunders

## MSMHG-402: Core Course Intellectual Property Rights and Biosafety (Credit 4)

**Time: 2h**

**Full Marks: 50**

**Lecture hours**

- |   |    |
|---|----|
| 1. Intellectual Property Rights   | 10 |
| 1.1. Introduction   |    |
| 1.2. Meaning, Relevance, Business Impact, Protection of Intellectual Property   |    |
| 1.3. Copyrights, Trademarks, Patents, Designs, Utility Models, Trade Secrets and Geographical Indications   |    |
| 1.4. Bio-diversity and IPR  |    |
| 1.5. Competing Rationales for Protection of Intellectual Property Rights  |    |
| 1.6. Introduction to the leading International Instruments concerning Intellectual Property Rights: the Berne Convention, Universal Copyright Convention, The Paris Convention, Patent Co-operation Treaty, TRIPS, The World Intellectual Property Organization (WIPO) and the UNESCO |    |
| 2. Patents  | 12 |
| 2.1.1. Concept of Patent  |    |
| 2.1.2. Product / Process Patents & Terminology  |    |
| 2.1.3. Duration of Patents- Law and Policy Consideration Elements of Patentability, - Novelty and Non-Obviousness (Inventive Steps and Industrial Application, Non- Patentable Subject Matter   |    |
| 2.1.4. Procedure for Filing of Patent Application and types of Applications   |    |
| 2.1.5. Procedure for Opposition,  |    |

2.1.6. Revocation of Patents	
2.1.7. Ownership and Maintenance of Patents	
2.1.8. Assignment and licensing of Patents	
2.1.9. Working of Patents- Compulsory Licensing	
2.1.10. Patent Agent- Qualification and Registration Procedure	
3. Patent Databases & Patent Information System	3
3.1.1. Patent Offices in India	
3.1.2. Importance of Patent Information in Business Development	
3.1.3. Patent search through Internet, Patent Databases	10
4. Biosafety	
4.1. Introduction of bio-safety,	
4.2. Biotechnology and bio-safety concerns at the level of individuals, institutions, society, region, country and world with special emphasis on Indian concerns.	
4.3. Primary Containment for Biohazards	
4.4. Biosafety practices in laboratory: laboratory associated infections and other hazards,	
4.5. Assessment of biological hazards and level of biosafety.	
5. Bio safety regulation: Special procedures for recombinant DNA based product production & handling of rDNA products and process in industry and in institutions (Indian context).	8
6. Roles of Institutional Biosafety Committee	4
7. Good laboratory practice	3

### Suggested Readings

1. IPR Biosafety & Bioethics by DeepaGoel and ShominiParashar, 2013
2. Essentials of Licensing Intellectual Property by Alexander I. Poltorak and Paul J. Lerner, wiley publisher, 2002
3. How to Invent and Protect Your Invention by Joseph P. Kennedy Sr. and Wayne H. Watkins, Wiley publisher, 2012
4. Patents for Chemicals, Pharmaceuticals and Biotechnology, by Phillip W Grubb, 2016
5. Laboratory Biorisk Management: Biosafety and Biosecurity - CRC Press, Reynolds M. Salerno, Jennifer Gaudio, 2015
6. Biological Safety: Principles and Practices (Biological Safety: Principles & Practices) 4th Edition, by Diane O. Fleming (Editor), Debra L. Hunt (Editor), 2006

### **MSMHG403 (Core Course) Project Proposal Preparation (Credit 4)**

**Full Marks: 50**

**Each student will formulate a research proposal and present the proposal before the panel of examiners.**

### **MSMHG404 (Discipline Centric Elective) Journal Club/ Communication Skills and Seminar Presentation (Credit 4)**

**Full Marks: 50**

**The student will deliver a comprehensive seminar on a current topic of his/her choice in the field of Molecular Biology & Human Genetics. The topic of Seminar must be different from the topics**

covered in any of the courses.

**MSMHG405 (Discipline Centric Elective)  
Visit of a National Lab or Biotech Industry  
(Credit 4)**

**Full Marks: 50**

National laboratory or Industrial Visit for the students of different courses for their exposures.

**MSMHG406  
Dissertation and Comprehensive Viva  
(Credit 4)**

**Full Marks: 50**

Each student will undertake an experimental project under supervision of one of the teachers (in or outside of the department) during Semester-IV and submit two copies of the dissertation which will include: a) Review of the relevant literature, b) Objectives of the study, c) Materials and Methods, d) Results/Observations (supported by figures/tables etc. as required), e) Discussion of the Results/Observations, f) Summary and g) References.

The understanding of the whole course content from semester-I through semester-IV will be assessed through a comprehensive viva-voce examination.