

#### M.Sc COURSE STRUCTURE CHOICE – BASED CREDIT SYSTEM DEPARTMENT OF GENETICS, OSMANIA UNIVERSITY (Proposed for academic year 2016 onwards)

Semes	ter – I	(1 roposed to		<i>Jewi</i> 2020 02				
Sl.	Syllabus Ref. No	Subject	Credits	Teaching Hours/ week	Marks			
No.					Internal Assessment	Semester Exam	Total	
	THEORY							
1.	G101 T	Principles of Inheritance	4	4	20	80	100	
2.	G 102 T	Cell Biology & Cytogenetics	4	4	20	80	100	
3.	G 103 T	Fundamentals of Biochemistry	4	4	20	80	100	
4.	G 104 T	Biostatistics and Population Genetics	4	4	20	80	100	
	PRACTICALS #							
1.	G 151 P	Principles of Inheritance	2	4		50	50	
2.	G 152 P	Cytogenetics	2	4		50	50	
3.	G 153 P	Fundamentals of Biochemistry	2	4		50	50	
4.	G 154 P	Biostatistics and Population Genetics	2	4		50	50	
		Total	24	32			600	

#### <u>SEMESTER</u> – II

Sl.	Syllabus	llabus Subject	Credits	Teaching	Marks			
No.	Ref. No			Hours/ week	Internal Assessment	Semester Exam	Total	
		THEORY						
1.	G 201 T	Genome organization and maintenance	4	4	20	80	100	
2.	G 202 T	Gene expression and regulation	4	4	20	80	100	
3.	G 203 T	Plant Genetics & Molecular Breeding	4	4	20	80	100	
4.	G 204 T	Human Genetics	4	4	20	80	100	
	PRACTICALS#							
1.	G 251 P	Genome organization and maintenance	2	4		50	50	
2.	G 252 P	Gene expression and regulation	2	4		50	50	
3.	G 253 P	Plant Genetics & Molecular Breeding	2	4		50	50	
4.	G 254 P	Human Genetics	2	4		50	50	
		Total	24	32			600	

#### <u>SEMESTER - III</u>

Sl. No.	Syllabus Ref. No	Subject	Credits	Teaching Hours/	Marks			
110.	Kei. No			Week	Internal Assessment	Semester Exam	Total	
	THEORY							
1.	G 301 T	Recombinant DNA Technology	4	4	20	80	100	
2.	G 302 T	Immunogenetics	4	4	20	80	100	
3.	G 303 T	ELECTIVE 1: Diagnosis and Therapeutics for Genetic Diseases (A) / Cell & Tissue Engineering (B)	4	4	20	80	100	
4.	G 304 T	ELECTIVE 2: Plant Biotechnology (A)/ Mouse developmental biology and disease models (B)	4	4	20	80	100	
5.	SEC - 3	Personality Development						
		PRACTICALS #						
1.	G 351 P	Recombinant DNA Technology	2	4		50	50	
	G 352 P	Immunogenetics	2	4		50	50	
3.	G 353 P	Diagnosis and Therapeutics for Genetic Diseases (A) / Cell & Tissue Engineering (B)	2	4		50	50	
4.	G 354 P	Plant Biotechnology (A)/ Mouse developmental biology and disease models (B)	2	4		50	50	
		Total	24	32			600	

## SEMESTER - IV

SI.	Syllabus Ref. No	Subject	Credits	Teaching Hours/ Week	Marks			
No.					Internal Assessment	Semester Exam	Total	
	r -							
1.	G 401 T	Bioinformatics	4	4	20	80	100	
2.	G 402 T	Applied Microbial Genetics	4	4	20	80	100	
3.	G 403 T	Elective 3: IPR & Biosafety (A) / Genetic Toxicology (B)	4	4	20	80	100	
3.	G 404 T	Genomics & Society ID	4	4	20	80	100	
	PRACTICALS #							
1.	G 451 P	Bioinformatics	2	4		50	50	
2.	G 452 P	Applied Microbial Genetics	2	4		50	50	
3.	G 453 P	Project Work	4	8		100	100	
		Total	24	32			600	
		GRAND TOTAL	96	128			2400	

## M.Sc (Genetics) Semester – I Paper : I (G101T) PRINCIPLES OF INHERITANCE

## **UNIT I: Eukaryote Model Systems for Genetic Analysis**

- 1.1 Life cycle and importance of Drosophila
- 1.2 Life cycle and importance of Neurospora
- 1.3 Life cycle and importance of Yeast
- 1.4 Life cycle and importance of C. elegans,
- 1.5 Life cycle and importance of Zebra fish,
- 1.6 Life cycle and importance of Arabidopsis
- 1.7 Life cycle and importance of Maize.

## **UNIT II: Mendelian Analysis of Inheritance and Extension to Mendel's Laws**

- 2.1 Mendel's Laws of Inheritance.
- 2.2 Allelic interactions; co-dominance and incomplete dominance; overdominance; pleiotropism; lethals and sub-lethals; penetrance and expressivity.
- 2.3 Position effect Variegation.
- 2.4 Epistasis: Non-allelic interactions and modification of Mendelian ratios.
- 2.5 Multiple alleles-ABO blood groups in humans, Rh blood group incompatibility; self sterility alleles in plants; complex loci in *Drosophila*.
- 2.6 Inborn errors of metabolism, one gene one enzyme concept
- 2.7 Inheritance of polygenic traits with specific examples

## UNIT – III: Linkage and Gene Mapping in Eukaryotes

- 3.1 Chromosomal basis of inheritance and Cytological basis of crossing over- Sterns experiments in *Drosophila*, Creighton and Mc Clintock experiment in maize
- 3.2 Inheritance of linked genes Coupling and Repulsion phase, meiotic recombination, gene mapping in *Drosophila* and maize using two point and three point test crosses with an emphasis on interference and coefficient of coincidence
- 3.3 Evidence for crossing over occurring at four strand stage Tetrad analysis and gene mapping in *Neurospora;* gene mapping using unordered tetrads in yeast.
- 3.4 Mitotic crossing over A. niger

## UNIT – IV: Sex determination and Extra-nuclear inheritance

- 4.1 Genetic basis of sex determination in Drosophila and S.alba
- 4.2 Dosage compensation; Sex -linked, sex-limited and sex-influenced characters
- 4.3 Extra-nuclear inheritance: Maternal effects; mitochondria and chloroplasts inheritance
- 4.4 Male Sterility in plants and their applications.

## Practicals (G 151 P) : Principles of Inheritance

- 1. Life cycle of Drosophila, maintenance of stocks.
- 2. Problems based on Mendelian Laws maize cobs and Drosophila genetics stocks
- 3. Segregation analysis in Drosophila and maize
- Segregation analysis in Diosophila and
  Mitotis in Onion root tips/ Mouse
  Meiosis in Maize/ Grasshopper Testes
- 6. Problems on linkage & sex linkage

#### M.Sc (Genetics) Semester – I Paper : II (G 102 T) CELL BIOLOGY AND CYTOGENETICS

#### Unit I: Cell cycle and Cell division

- 1.1 Structure and function of cellular organelles (Endoplasmic reticulum, Golgi complex, lysosomes, vacuoles, peroxisomes, mitochondria, chloroplast, secretory pathway)
- 1.2.Cytoskeleton and extracellular matrix (Microtubules, intermediate filaments, microfilaments, integrins, focal adhesions, hemidesmosomes, selectins, cadherins, adherin junctions, desmosomes, tight junctions, gap junctions, plasmodesmata and cell wall)
- 1.3. Cell cycle- Phases of cell cycle, restriction points, cell cycle determining genes,  $G_0$  Phase (Quiescence phase, Points of no return), totipotency of stem cells.
- 1.4.Chromosome segregation in mitosis and meiosis mitotic apparatus, distribution of microtubule organizing centres, formation of synaptonemal complex, cytokinesis
- 1.5.Cell death: Apoptosis (Intrinsic and Extrinsic pathways), necrosis and autophagy

#### **Unit II: Chromatin organization**

- 2.1. Components of chromatin Chromosome structure, Euchromatin and Heterochromatin
- 2.2. Chromatin organization Structure and organization of nucleosome in chromatin, solenoids, loops and scaffolds, nucleosome phasing, active and inactive states of chromatin.
- 2.3. Chromatin Modifications Histone modifications and their effect
- 2.4. Dosage compensation, X chromosome inactivation
- 2.5. Evolutionary significance

#### **Unit III: Chromosome Abnormalities**

- 3.1. Structural chromosomal abnormalities- Origin of breaks and gaps, ring chromosomes, Isochromosomes, centric fusion, centric fission- Mechanisms involved. Deletions, duplications, inversions, translocations.
- 3.2. Numerical chromosomal abnormalities- Aneuploidy, Polyploidy. Non-Disjunction and Anaphase lag.
- 3.3. Chromosome instability and associated syndromes
- 3.4. Sister chromatid exchanges and its significance

#### Unit IV: Detection and analysis of chromosomal alterations

- 4.1. Karyotyping and its significance
- 4.2. Banding techniques (G, Q. T, R, etc)
- 4.3. Studies on polytene chromosomes for cytogenetic mapping.
- 4.4.Chromosome break points Mapping( Deletion mapping, translocation mapping, Inversion mapping).
- 4.5. Insitu hybridization, FISH, SKY

# Practicals (G 152 P) : Cell Biology and Cytogenetics

- 1. Barr Body identification

- Dan Dody Identification
  Karyotype analysis
  G banding
  Polytene Chromosome
  Induction of polyploidy

## M.Sc (Genetics) Semester – I Paper : III (G 103 T) Fundamentals of Biochemistry

### **Unit I: Bioenergetics, Enzymology and Biomolecules**

- 1.1 Laws of thermodynamics, Gibbs free energy, Enthalpy, Entropy
- 1.2 Proteins (Primary, secondary & tertiary structures), Ramachandran plot
- 1.3 Catalysis, enzymes and enzyme kinetics, Briggs-Haldane reaction, Michaelis-Menten equation, Coenzymes, Cofactors, enzyme regulation.

## Unit II: Carbohydrate metabolism

- 2.1 Carbohydrates (Classification, monosaccharides, disaccharides, oligosaccharides & polysaccharides).
- 2.2 Glycolysis, TCA cycle, Electron transport chain, Gluconeogenesis, Glycogenesis, Glycogenolysis, Glucuronic acid cycle, Pentose phosphate pathway, Entner-Doudoroff pathway, Cori cycle, Photosynthesis, C3 & C4 cycle

## Unit III: Metabolism of lipids and amino acids

- 3.1 Lipids (Classification, fatty acids, steroids), Hydrolysis of triacyl glycerols, Betaoxidation, Fatty acid biosynthesis, Prostaglandin biosynthesis, Cholesterol metabolism.
- 3.2 Amino acids, Amino acid degradation, Urea cycle, Overview of amino acids Biosynthesis
- 3.3 Nitrogen metabolism: Nitrate and ammonium assimilation
- 3.4 Nucleotide biosynthesis and degradation.

## Unit IV: Cell signalling

- 4.1 Cell communication (autocrine and paracrine), Components of cell signalling (Growth factors, receptors, adaptors and signal transducers)
- 4.2 Calmodulin pathway, GPCR signalling pathways, RTK signaling pathways, Wnt signalling pathways, Toll-like receptor signalling pathways, second messengers.
- 4.3 Overview of signalling.

#### Practicals (G 153 P) : Fundamentals of Biochemistry

- 1. Preparation of buffers.
- 2. Spectroscopy, Centrifugation, X-ray diffraction, NMR
- 3. Carbohydrate Analysis
- 4. Amino acid Analysis
- 5. Isolation and measurement of proteins
- 6. SDS-PAGE
- 7. Column chromatography Gel filtration (size exclusion)

## M.Sc (Genetics) Semester – I Paper – IV (G 104 T) BIOSTATISTICS AND POPULATION GENETICS

## **UNIT I: Biostatistics**

- 1.1.Sampling and Experimental design
- 1.2.Descriptive analysis of data: Types of variables, Data alignment and representation, Measures of central tendency, Measures of dispersion
- 1.3. Concepts of probability: Axioms of probability
- 1.4. Probability distributions : Binomial, Poisson, Normal distribution.
- 1.5.Hypothesis testing: Null and alternate hypothesis, test of significance, Type I and Type II errors, confidence intervals and confidence levels
- 1.6.Estimates and test statistics: Chi-square test (test for goodness of fit, homogeneity test, linkage, test of independence), Z test (for proportions and means), t- test (students t test, paired t test), ANOVA One way and Two-way Anova (F- test).
- 1.7. Correlation and regression (Simple regression, multiple regression, logistic regression)

## **UNIT II: Population Genetics**

- 2.1. Population structure, Gene pool, Estimation of gene and genotype frequencies for biallelic, multiple allelic and X- linked loci.
- 2.2. Hardy-Weinberg principle, Establishment of law for a) autosomal biallelic loci b) multiple allelic loci c) X-linked loci.
- 2.3. Factors affecting HWE: Mutation, Selection, Migration, Genetic drift, Effective population size
- 2.4. Genetic load: Mutational and segregational load
- 2.5. Linkage disequilibrium
- 2.6. Effects of Inbreeding and assortative mating.

## **UNIT III: Quantitative Genetics**

- 3.1. Quantitative traits –features (Population mean, average effect, breeding value, dominance deviation, interaction deviation)
- 3.2. Components of Phenotypic Variance: Reaction Norms, Resemblance between relatives
- 3.3. Genetic architecture of quantitative variance, Genotypic Values: Additivity, dominance and epistasis, genetic covariance (Offspring and one parent, offspring and mid-parent, half sibs, full sibs)
- 3.4. Correlated characters, GXE effects and maternal effects
- 3.5. Heritability (ANOVA and Regression)
- 3.6. Heterosis and Inbreeding depression

## **UNIT IV: Genetic Distance And Phylogentic Analysis**

- 4.1. Genetic diversity
- 4.2. Genetic distance and measures of relatedness, Molecular dating
- 4.3. Cluster Analysis: Construction of cluster diagrams and dendrograms
- 4.4. Principal Component Analysis

- 4.3. Phylogenetic analysis (UPGMA)
- 4.4. Bayesian methods for phylogenetic estimation

## Practicals (G 154 P): Biostatistics And Population Genetics

- 1. Data alignment and Descriptive analysis of data- Manual and Excel
- 2. Problems on probability
- 3. Problems on Chi-Square test
- 4. Problems on Z test
- 5. Problems on t-test
- 6. One way and two-way ANOVA
- 7. Calculation of correlation and regression
- 8. Calculation of gene and genotype frequencies
- 9. Problems on Hardy-Weinberg Equilibrium
- 10. Calculation of inbreeding coefficient
- 11. Estimation of heritability
- 12. NEIs Index

#### M.Sc (Genetics) Semester – II Paper – I (G 201 T) GENOME ORGANIZATION AND MAINTENANCE

## **Unit I: Genome Organization**

1.1.DNA structure

- 1.2. Prokaryotic genome organization
- 1.3. Eukaryotic genome organization
- 1.4.Extrachromosmal genetic elements (plasmids, mitochondrial genome, chloroplast genome)
- 1.5.Horizontal gene transfer (transformation, transduction, conjugation. Genome islands)
- 1.6. Transposable elements and their implication in genome evolution
- 1.7.Bacteriophages (lambda phage)

## **Unit II: Genome Replication and Replication Associated Errors**

- 2.1. DNA replication
- 2.2. Bacterial chromosomal replication
- 2.3. Eukaryotic chromosomal replication
- 2.4. Plasmid Replication
- 2.5. Replication of mitochondrial and chloroplast genomes
- 2.6.Regulation of genome replication
- 2.7. Replication associated errors

## Unit III: DNA Damage and Repair

- 3.1. Internal and external agents causing DNA damages
- 3.2. DNA damages (Oxidative damages, Depurinations, Depyrimidinations, O6methylguanines, Cytosine deamination, single and double strand breaks)
- 3.3. Mechanisms of DNA damage (transition, transversion, frameshift, nonsense mutations)
- 3.4. Repair mechanisms (Photo reactivation, excision repair, mismatch repair, post replication repair, SOS repair)

#### **Unit IV: Genome Rearrangements**

- 4.1. Whole genome duplication
- 4.2. Segmental duplication
- 4.3. Single nucleotide variations
- 5.4. Homologous recombination
- 5.5. Non-homologous end joining
- 5.6. Site-specific recombination
- 5.7 Transposon and repeats mediated rearrangements
- 5.8. Gene conversion

## Practicals (G 251 P): Genome Organization and Maintenance

- 1. Isolation of genomic DNA from plant tissue
- 2. Isolation of genomic DNA from Animal tissue
- 3. Isolation of genomic DNA from human blood
- 4. Induction of mutants using chemical agents
- 5. Checking of DNA Purity and concentration agarose and spectrophotometer
- 6. Problems on DNA Kinetics
- 7. Tm determination of DNA
- 8. Comet Assay

#### M.Sc (Genetics) Semester – II Paper – II (G 202 T) GENE EXPRESSION AND REGULATION

#### **Unit I: Structure of Prokaryotic and Eukaryotic Genes**

- 1.1.Structure of prokaryotic genes
- 1.2. Organization of prokaryotic genes into operons
- 1.3.Structure of eukaryotic genes (introns, exons, UTRs, core and proximal promoters, enhancers)
- 1.4.Number of genes in prokaryotes and eukaryotes
- 1.5.RNA coding genes (rRNA, tRNA)
- 1.6.Regulatory small RNA coding genes (miRNAs)

#### **Unit II: Gene Expression**

- 2.1. Transcription machinery in prokaryotes and eukaryotes
- 2.2. Transcription process (initiation, elongation, termination, processing of transcripts)
- 2.3. Translational machinery in prokaryotes and eukaryotes
- 2.4. Translation process (initiation, elongation, termination, folding, processing)
- 2.5.Co-ordinated regulation of gene expression in prokaryotes and eukaryotes

#### **Unit III: Regulation of Gene Expression**

- 3.1. Regulation of transcription (proximal promoter, specific transcription factors, enhancers, multiple promoters, alternate transcription initiation sites, multiple PolyA sites)
- 3.2. Post transcriptional regulation of gene expression (pre-mRNA splicing, miRNA based regulation)
- 3.3. Alternate transcript formation (Exon skipping, intron inclusion, alternate splice sites, 5' end variations, 3' end variations)
- 3.4. Regulation of translation (codon usage/bias, 5'UTR based signals, upstream ORFs, upstream, start codons, alternate splicing in UTRS, 3'-UTR based regulation)
- 3.5. Post translational regulation of gene expression

#### **Unit IV: Epigenetic Regulation of Gene Expression**

- 4.1. Overview of epigenetic regulation
- 4.2. Chromatin remodelling and gene expression
- 4.3. Histone modifications and gene expression
- 4.4. Small RNA based epigenetic regulation
- 4.5. Propagation of epigenetic regulation (genome imprinting)

# Practicals (G 252 P): Gene Expression and Regulation

- 1. Plasmid DNA isolation
- 2. Isolation of mRNA trizol method
- 3. Understanding Human genome project
- 4. Epigenetic Analysis Insilico
- 5. Serum miRNA analysis.

#### M.Sc (Genetics) Semester – II Paper – III (G 203 T) PLANT GENETICS AND MOLECULAR BREEDING

#### **UNIT I: Principles of Plant Breeding**

- 1.1 Introduction to plant breeding. Domestication of crop plants Centres of origin and diversity; Basic features of plant breeding and Objectives of plant breeding
- 1.2 Plant genetics resources and conservation strategies. Sources of plant genetics resources; Methods of germplasm conservation; Evaluation and utilization of plant genetic resources
- 1.3 Reproductive systems in plants: Sexual reproduction self and cross fertilization Autogamy, Allogamy and often cross pollinated plants; Asexual reproduction and Apomixis
- 1.4 Genetic basis of breeding: Mating systems of plants; Wide hybridization Inter-specific crosses and inter-generic hybridization; Role of wide hybridization in crop improvement.

#### **UNIT II: Plant Breeding Methodologies**

- 2.1 Breeding Methods in self pollinating crops: Pure line selection; Pedigree method; Bulk population methods; Single seed descent method; Back cross method and Multi lines
- 2.2 Breeding methods in cross pollinating crops: Mass selection; Ear-to-row selection; Progeny selection and Recurrent selection methods, Hybrid Breeding – Development and evaluation of inbred lines, A, B and R lines, Development of hybrids., male sterility systems
- 2.3 Mutation breeding: Physical and Chemical mutagens; Mutation breeding in seed crops and vegetative propagation; and TILLING.
- 2.4 Cultivar release and certification, Cultivar release; Seed certification and multiplication; Plant breeders rights

#### **UNIT III: Specific Breeding Methods**

- 3.1 Breeding for disease resistance. Genetics of pathogenecity; Genetics of disease resistance; Methods of breeding for disease resistance
- 3.2 Breeding for insect resistance: Mechanisms of insect resistance; Breeding methods for pest resistance
- 3.3 Breeding for abiotic stress tolerance, Breeding for drought, salinity, temperature and flood tolerance
- 3.4 Breeding for nutritional improvement, Nutritional quality, Improved protein content and Improved oil quality

#### **UNIT IV: Biotechnological Approaches for Crop Improvement**

- 4.1 Plant tissue culture techniques in crop improvement. Introduction to plant cell-tissue culture techniques, Haploids and di-haploids, Somaclonal variation, Protoplast fusion, Micro propagation
- 4.2 Transgenics in crop improvement: Gene transfer methods in plants; Production of

transgenics for biotic and abiotic stress tolerance; Transgenic male-sterility systems and development of hybrids; Cis-genic approaches

- 4.3 Gene silencing: RNAi and its applications for crop improvement
- 4.4 Molecular plant breeding tools, Molecular markers, Marker assisted breeding, Genome mapping QTL mapping

## Practicals (G 253 P): Plant Genetics and Molecular Breeding

- 1. Floral morphology and pollination methods in self-pollinating and cross pollinating crops.
- 2. Callus Initiation and Plantlet Regeneration.
- 3. Agrobacterium/Biolistic mediated gene transfer
- 4. RAPD/SSR analysis
- 5. Linkage analysis
- 6. Heterosis

## M.Sc (Genetics) Semester – II Paper IV (G204 T) HUMAN GENETICS

## **UNIT I: Genes in Families**

- 1.1 Patterns of Inheritance (AD, AR, XD, XR, YL, Maternal inheritance)
- 1.2 Pedigree analysis
- 1.3 Extensions to Mendelian inheritance
  - 1.3.1 Incomplete penetrance and variable expressivity
  - 1.3.2 Epistasis, pleiotropism
  - 1.3.3 Gametic imprinting
  - 1.3.4 Mosaicism
  - 1.3.5 Anticipation
- 1.3 Genetic and phenotypic heterogeneity (Inter and Intra allelic heterogeneity)
- 1.4 Segregation analysis
- 1.5 Analysis of multifactorial condition-polygenic inheritance
  - 1.5.1 Threshold model
  - 1.5.2 Twin studies in genetic analysis

## **UNIT II: Linkage Analysis**

- 2.1 Linkage detection and estimation
  - 2.1.1 Parametric and non-parametric methods: Lod score, y- statistics, sib-pair method, IBD, affected relatives methods
- 2.2 Linkage analysis through family studies-Homozygosity mapping
- 2.3 Extensions of linkage studies for genetic heterogeneity, reduced penetrance and epistasis
- 2.4 Population based Linkage analysis
- 2.5 Whole genome linkage analysis
- 2.6 Genetic models and Allelic effects
- 2.7 Different types of genetic markers
- 2.8 Linkage disequilibrium analysis
- 2.9 Haplotype analysis
- 2.10 Analysis of gene-phenotype interactions

## **Unit III: Genetic Basis of Human Diseases**

- 3.1 Molecular pathology of Chromosome anomalies
  - 3.1.1 Numerical chromosomal disorders
  - 3.1.2 Structural chromosomal disorders
  - 3.1.3 Chromosome instability syndromes
- 3.2 Molecular basis of single gene disorders
  - 3.2.1 Autosomal Dominant and recessive disorders
  - 3.2.2 X-linked dominant and recessive disorders, Y-linked, X-influenced and X-limited disorders

- 3.3 Inherited biochemical diseases
  - 3.3.1 Enzyme defects- amino acid metabolism
  - 3.3.2 Lipid metabolic disorders
  - 3.3.3 Carbohydrate associated disorders
  - 3.3.4 Defects in purine metabolism
  - 3.3.5 Defects in membrane transport
  - 3.3.6 Defects in structural proteins
  - 3.3.7 Collagen disorders
  - 3.3.8 Defects in receptor proteins
- 3.4 Complex genetic diseases Hypertension, Diabetes mellitus
- 3.5 Mitochondrial diseases
- 3.6 Cancer as a genetic disease
- 3.7 Familial and sporadic cancers Oncogenes, tumor suppressor genes, mutator genes.

#### **UNIT IV: Strategies for Disease Gene Identification and Gene Mapping**

- 4.1 Approaches for gene identification
  - 4.1.1 Functional cloning
  - 4.1.2 Positional cloning
  - 4.1.3 Position independent candidate gene approach
  - 4.1.5 Position dependent candidate gene approach
  - 4.1.5 Epigenetic signatures
  - 4.1.6 Transcriptome analysis
- 4.2 Association studies
- 4.3 Case-control studies
  - 4.3.1 Population based studies
  - 4.3.2 GWAS
- 4.4 Mapping:
  - 4.4.1 Low resolution mapping: Sub- chromosomal mapping, Chromosomal break points, FISH, cytogenetic methods, Somatic cell hybrid mapping, Radiation hybrid mapping
  - 4.4.2 High resolution mapping: DNA FIBRE FISH, Restriction mapping, VNTR microsatellite markers for mapping, EST mapping, STS mapping, SNP mapping, Conserved region mapping: IRE, CpG site mapping, Promoter site recognition
  - 4.4.3 Sequencing
- 4.5 Mapping for single gene disorders
- 4.6 Mapping for complex genetic disorders

## Practicals (G 254 P): Human Genetics

- 1. Pedigree analysis
- 2. Sister chromatid exchanges
- 3. Amino acidopathies and carbohydrate metabolic error identification
- 4. Segregation analysis
- 5. Problems on Parametric and non-parametric variables
- 6. Lod score
- 7. Sib pairs
- 8. Haplotype analysis
- 9. LD Maps