Human Genetics syllabus w.e.f. 2016- 17 AB.pdf Humangenetics\_IIsem\_2016-17AB.pdf BSc Human Genetics CBCS\_IIIsem.doc Humangenetics\_IVsem\_2015-16AB.pdf Human Genetics\_VSem\_2015-16AB.pdf HUMAN GENETICS\_VI sem\_2015-16AB\_WEB.pdf

# ADIKAVI NANNAYA UNIVERSITY RAJAMAHENDRAVARAM CBCS / Semester System (W.e.f. 2016-17 Admitted Batch) I Semester Syllabus HUMAN GENETICS GENETICS & HUMAN HEREDITY

## Unit - I : Mendelian Genetics and Extensions

- 1.1 Mendelian Principles of inheritance Law of segregation, Law of independent assortment –animal examples; Mendelian inheritance of human traits; Chromosome theory of inheritance.
- 1.2 Incomplete dominance and codominance; Multiple alleles, Lethal alleles, Pleiotropy, Penetrance and Expressivity general animal and human examples
- 1.3 Two gene interactions Epistatic, nonepistatic interactions; Polygenic inheritance in man and other animal organisms.
- 1.4 Genes and environment -norm of reaction, phenocopies, developmental noise

## Unit - II: Sex Linked Inheritance and Sex Determination

- 2.1 Sex Linked Inheritance Sex linked inheritance in Drosophila and human; Sex limited and Sex influenced inheritance
- 2.2 Sex Determination Sex determination in Drosophila Genic balance theory
- 2.3 Sexdetermination in eukaryotes -heterogametic, homogametic, haplodiploidy, role of environmental factors, mosaics
- 2.4 Sex determination in mammals- human Y chromosome

#### **Unit – III: Extrachromosomal Inheritance**

- 3.1 Mitochondrial inheritance (petite mutations); Mitochondrial inheritance in man
- 3.2Maternal inheritance-shell coiling in snail, Ephestia pigmentation
- 3.3Infective heredity- symbionts in Drosophila, Kappa particles in Paramecium.

## Unit – IV: Linkage, crossing over and chromosome mapping

- 4.1 Linkage and chromosome mapping in eukaryotes cytological basis of crossing over; recombination frequency, two factor and three factor crosses.
- 4.2 Linkage and chromosome mapping in prokaryotes bacteria and bacteriophages transformation, transduction, conjugation; gene mapping in bacteria.
- 4.3 Genetic definition of gene Complementation test, intragenic complementation,rII locus of phage T4

## Unit -V: Variation in Chromosome number and structure

- 5.1 Specialized chromosomes -Lampbrush chromosomes. Polytene chromosomes: Supernumerary chromosomes.
- 5.2 Variation in chromosome structure Deletion, Duplication, Inversion, Translocation, Position effect
- 5.3Variation in chromosome number Euploidy and Aneuploidy

- 1. Mendel's laws through seed ratios& Drosophila mutants.
- 2. Statistical tests in genetic analysis application of laws of probability (product rule, sum rule, binomial probability); chi square test and its application in the analysis of genetic data.
- 3. Study of linkage, recombination, chromosome mapping using test cross data.
- 4. Pedigree analysis for dominant and recessive autosomal and sex linked traits.
- 5. Study of human genetic traits: Sickle cell anaemia, Xeroderma, Pigmentosum, Albinism, red-green Colour blindness, Widow's peak, Rolling of tongue, Hitchhiker's thumb and Attached ear lobe.
- 6. Incomplete dominance and gene interaction through seed ratios
- 7. Blood Typing: ABO groups & Rh factor.
- 8. Study of aneuploidy: Down's, Klinefelter's and Turner's syndromes.
- 9. Permanent Slides showing Translocation Ring, Laggards and Inversion Bridge.
- 10. Mitosis & Meiosis through temporary squash preparation.
- 11. Squash preparation of salivary glands to observe polytene chromosomes.
- 12.Smear technique to demonstrate sex chromatinin buccal epithelialcells.

## **Suggested Readings**

- 1. Gardner, E.J., Simmons, M.J., Snustad, D.P. (1991). Principles of Genetics, John Wiley & sons, India.8th edition.
- 2. Snustad, D.P. and Simmons, M.J. (2010). Principles of Genetics, John Wiley & Sons Inc., India.5th edition.
- 3. Klug, W.S., Cummings, M.R., Spencer, C.A. (2012). Concepts of Genetics. Benjamin Cummings, U.S.A. 10th edition.
- 4. Griffiths, A.J.F., Wessler, S.R., Carroll, S.B., Doebley, J. (2010). Introduction to Genetic Analysis. W. H. Freeman and Co., U.S.A. 10th edition.

## ADIKAVI NANNAYA UNIVERSITY: RAJAMAHENDRAVARAM CBCS/ SEMESTER SYSTEM

**II SEMESTER: B.Sc. HUMAN GENETICS** 

(FOR 2016-17 ADMITTED BATCH)

### Human Genetics& Cytogenetics

#### Unit 1 Basic Human Genetics - Monogenic traits

- 1.1 History of Human Genetics.
- 1.2 Pedigrees family history, symbols, construction of pedigree
- 1.3 Monogenic traits autosomal inheritance, sex-linked inheritance, sex-limited and sex influenced inheritance, mitochondrial inheritance
- 1.4 Complications in pedigree patterns non-penetrance, expressivity, pleiotropy, genetic heterogeneity, genomic imprinting, uniparental disomy, male lethality, X inactivation, consanguinity

#### **Unit 2 Basic Human Genetics - Complex traits**

- 2.1 Approaches to analysis of complex traits Nature vs nurture, monozygotic and dizygotic twins
- 2.2 Polygenic inheritance of continuous traits normal growth charts, dysmorphology
- 2.3 Polygenic inheritance of discontinuous traits threshold model, liability and recurrence risk
- 2.4 Genetic susceptibility in multifactorial disorders diabetes

## Unit 3 Genetic Mapping of Mendelian and Complex characters

- 3.1 Identifying recombinants and non-recombinants in pedigrees
- 3.2 Genetic and physical map distances genetic markers, mapping of genetic traits
- 3.3 Two point mapping LODscore analysis, multipoint mapping, homozygositymapping

3.4 Genetic mapping of complex traits - difficulties in mapping, allele sharing methods, sib pair analysis, allelic association, linkage disequilibrium mapping

## **Unit 4 Human Chromosomes**

- 4.1 History of human cytogenetics
- 4.2Cell division cycle mitotic process, meiotic process
- 4.3 Human karyotype banding, nomenclature of banding
- 4.4 Nomenclature of aberrant karyotypes

#### **Unit 5 Chromosome anomalies**

- 5.1 Common syndromes due to numerical chromosome changes
- 5.2 Common syndromes due to structural alterations (translocations, duplications, deletions, microdeletions, fragile sites)
- 5.3Common chromosome abnormalities in cancer

- 1. Preparation of pedigree charts for blood group, tongue rolling, ear lobes and colorblindness
- 2. Preparation of metaphase chromosome spread using peripheral blood sample.

- 3. Polygenic inheritance finger print ridge count
- 4. Study of various abnormal karyotypes observed in humans.
- 5. Genetics of codominant genes blood groups.
- 6. Sterilization techniques for leukocyte culture
- 7. Inoculation and Culture of human leucocytes
- 8. Preparation of metaphase plates and their staining and analysis
- 9. G- banding of metaphase plates and their analysis
- 10. Human karyotyping numericals on chromosome number.
- 11. Barr Body analysis.
- 12. Micrometric analysis of chromosomes.
- 13. Camera-lucida drawing of chromosomes.
- 14. Dermatoglyphics
- 15. Sister Chromatid exchange analysis from peripheral blood

## **Suggested Readings**

- 1. Human Genetics: Concept and Application by Ricki Lewis 10<sup>th</sup> Edition
- 2. Vogel and Motulsky's Human Genetics: Problems and Approaches
- 3. The Principles of Clinical Cytogenetics by Steven L. Gersen, Martha B. Keagle 3 edition.
- 4. Human Cytogenetics: Constitutional Analysis: a Practical Approach by Denise E. Rooney.

## Adikavi Nannaya University B.Sc Human Genetics

## Semester - III Paper- III: Human Molecular Genetics

## Unit 1 DNA, RNA and Protein Structure

- 1.1Building blocks and chemical bonds in DNA, structure of DNA, A-B-Z and triplex DNA,
- 1.2Building blocks and chemical bonds in RNA Structure of RNA
- 1.3 Building blocks and chemical bonds in peptides- primary, secondary, tertiary and quaternary structure of proteins

#### Unit 2 Gene expression

- 2.1 Central dogma of molecular biology
- 2.2 RNA transcription
- 2.3 RNA processing
- 2.4 Translation, post-translation processing

## Unit 3 DNA replication, recombination, Mutagenesis and DNA repair

- 3.1 DNA replication semiconservative, semi-discontinuous, DNA replication machinery
- 3.2 DNA recombination
- 3.3DNA mutagenesis
- 3.4 DNA repair

#### **Unit 4 Human Chromosome Organization**

- 4.1 Packaging of DNA multiple hierarchies of DNA folding
- 4.2Chromosomes as functional organelles –origins of replication, telomeres, centromeres
- 4.3. Heterochromatin and euchromatin

#### Unit 5 Human Genome organization

- 5.1 Mitochondrial genome replication, genes, genetic code
- 5.2 Nuclear genome protein coding genes, RNA genes
- 5.3 Nuclear genome highly repetitive DNA, heterochromatin and transposon repeats

- 1. Extraction of DNA from human lymphocytes
- 2. Quantitation of DNA by UV spectrophotometry
- 3. Paper chromatography of amino acids
- 4. Electrophoresis: agarose gel electrophoresis, PAGE
- 5. Study of isozymes by PAGE
- 6. Comet assay to measure DNA damage
- 7. Problem based on homologous and site-specific recombination
- 8. Effects of mutagens on wt and repair deficient E.coli strains.
- 9. Preparation of Human chromosome spread and banding

## **Suggested Readings**

- 1. Human Molecular Genetics by T. Strachan
- 2. Human Molecular Genetics by Gerard Meurant
- 3. Human Molecular Genetics by Christopher G Mathew.
- 4. Human Molecular Genetics by Sudbery

5. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, Johannes Zschocke.

## **ADIKAVI NANNAYA UNIVERSITY**

## CBCS/SEMESTER SYSTEM IV SEMESTER: B.SC HUMAN GENETICS W.E.F. 2015-16 ADMITTED BATCH

### DSC 1D (Paper- IV): Recombinant DNA Technology

### **Unit 1 Cell Based Cloning**

1.1Restriction endonucleases and other enzymes used in manipulating DNA molecules
1.2Cloning vectors – plasmid vectors, lambda and cosmid vectors, P1 phage vectors,
YAC, BAC, M13 or phagemid vectors, expression vectors
1.3Introducing recombinant DNA into recipient cells
1.4 DNA libraries -generation of genomic and cDNA libraries; chromosomal DNA libraries

#### **Unit 2 Screening constructs**

- 2.1 Nucleic acid hybridization-
- 2.2 Sequencing DNA- Sanger's method
- 2.3 PCR- fundamentals, designing of primers, real time PCR
- 2.4 Human- rodent somatic cell hybrids

## Unit 3 Genetic and Physical Mapping of Human genome

- 3.1 Genetic Mapping genetic polymorphism, RFLP,STR polymorphism,
- 3.2 Homozygosity mapping, linkage dis-equilibrium mapping, radiation hybrid mapping
- 3.3Genotyping SNPs
- 3.4 Physical mapping assembling contigs from BAC libraries

## Unit 4 Cloning Human disease genes

4.1 Cloning human disease genes- functional candidate gene cloning, positional candidate gene cloning

4.2Detection of mutations in human genes –SSCP analysis, DGGE, chemical mismatch cleavage

4.3Detection of mutation in human gene – DNA sequencing, heteroduplex analysis, protein truncation

## Unit 5 Applications of rDNA technology

5.1 DNA fingerprinting – use of mini-satellites for DNA fingerprinting, single locus probes, STRs

5.2 Genetic testing – prenatal testing, neonatal screening, diagnosis of genetic disease in children after birth, pre-symptomatic testing.

5.3 In vivo, in vitro gene therapy; vehicles for gene therapy; gene therapy for heritable and non- heritable genetic diseases.

- 1. Isolation of plasmid DNA from *E. coli* cells.
- 2. Digestion of plasmid DNA with restriction enzymes.
- 3. Estimation of size of a DNA fragment after electrophoresis using DNA markers
- 4. Construction of restriction digestion maps from data provided
- 5. Recovery of DNA from low-melting temperature agarose gel
- 6. Preparation of competent cells of E.coli
- 7. Transformation of competent E.coli cells with plasmid DNA
- 8. Amplification of a DNA fragment by PCR.
- 7. Complementation of beta-galactosidase for Blue and White selection.
- 8. Southern blotting
- 9.Western blotting.

## **Suggested Readings**

1. Gene Cloning and DNA Analysis (2010) 6th ed., Brown, T.A., Wiley-Blackwell publishing(Oxford, UK), ISBN: 978-1-4051-8173-0.

 Principles of Gene Manipulation and Genomics (2006) 7th ed., Primrose, S.B., and Twyman, R. M., Blackwell publishing (Oxford, UK) ISBN:13: 978-1-4051-3544-3.
 Molecular Biotechnology: Principles and Applications of Recombinant DNA (2010) 4th ed., Glick B.R., Pasternak, J.J. and Patten, C.L., ASM Press (Washington DC), ISBN: 978-1-55581-498-4 (HC).

4. Human Molecular Genetics by Sudbery.

## Adikavi Nannaya University CBCS Semester System B.Sc. Human Genetics Semester - V PAPER-V : STATISTICS AND INFORMATICS IN HUMAN GENETICS

#### **Unit 1 Descriptive Statistics**

- 1.1 Methods of presentation and interpretation of data frequency distribution, graphical representation of data, histogram, frequency polygon, frequency curve.
- 1.2 Measures of Central tendency mean, median, mode
- 1.3 Measures of Dispersion standard deviation, variance.

#### **Unit 2 Elementary Probability**

2.1 Mathematical definition of probability of an event, Use of permutations and combinations in calculations of Probability

2.2 Conditional probability, Additive and Multiplication law of Probability.

2.3 Probability Distributions: Binomial, Poisson and normal distributions.

#### Unit 3 Correlation analysis, test of significance and ANOVA

3.1Correlation and regressionanalysis— Relationship between variables 3.2Test of significance – statistical and scientific hypothesis, null and alternative hypothesis, procedure of hypothesis testing,

3.3Test of significance – student's t test, chi-square test.

3.4.ANOVA – general idea of one way and two way analysis

#### Unit 4 Computers, operating systems and Internet

4.1 Principles of computer operations –basic computer architecture, hardware architecture

4.2Principles of computer operations – software architecture, operating systems, Programming languages –traditional and scripting languages, Java, markup languages, application programs

4.3 Internet Services, email, WWW search engines

#### **Unit 5 Bioinformatics**

5.1 History of Bioinformatics

5.2 Databases and search tools – NCBI, EBI, GenomeNet; Databasemining tools – BLAST 5.3 Database archives – nucleic acid sequence databases, genome databases and genome browsers, protein sequence databases, databases of protein families,.

Frequency distribution
 Various types of graphs
 Mean, Median, Mode
 Standard deviation, variance and coefficient of variation
 Testing of hypotheses regarding population mean
 Testing of hypotheses about the difference between population means
 Chi-square test
 Testing of Correlation Coefficient
 Fitting of simple linear regression
 One-way ANOVA&Two-way ANOVA
 Internet basics
 Sequence retrieval (protein and gene) from NCBI, Structure download (protein and DNA) from PDB
 Molecular file formats - FASTA, GenBank, Genpept, GCG, CLUSTAL, Swiss-Prot, FIR.

## Suggested Readings

- 1. Fowler, J., Cohen, L. and Jarvis, P. (1998). Practical Statistics for Field Biology. John Wiley and Sons, 2nd ed. .
- 2. Bland, M. (2006). An Introduction to Medical Statistics. Oxford University Press, 3rd ed.
- 3. Finney, D.J. (1980). Statistics for Biologists. Chapman and Hall Ltd.
- 4. Wayne, W, Daniel (1999). Biostatistics: A Foundation for Analysis in Health Sciences. John Wiley and Sons, 7th ed.

## Adikavi Nannaya University CBCS Semester System B.Sc. Human Genetics Semester - V Paper - VI: Human Biochemical and Cytogenetics

#### UNIT-1

The concept of Genetic Polymorphism

Blood Groups: ABO, MN and Rh systems, ABH saliva secretion

#### UNIT-2

Hemoglobin – Structural Diversity & Hemoglobinopathies

Serum Proteins Haptoglobin

Heterochromatin and genetic inactivation : Lyon's hypothesis

#### UNIT-3

Inborn errors of metabolism : Albinism, Phenylketoneuria and Alkaptonuria.

Pharmacogenetics: glucose-6-phosphatedehydrogenase deficiency, Pseudocholinesterase deficiency

#### UNIT-4

Human somatic chromosomes - Nomenclature : Standardization in Human Cytogenetics - Denver Report (1960), London Report (1953), Chicago Report (1966), Paris Report (1971)

Morphological variability of Human chromosomes.

#### UNIT-5

Chromosomal abnormalities in Man - Numerical aberrations –Classical syndromes (Down syndrome, Edward syndrome, Patau syndrome, Turner syndrome, Klienfelter syndrome)

Structural aberrations- Cri-du- chat syndrome, Wolf-Hirsch horn syndrome

#### III Year Practical VI: Practicals in Human Biochemical and Cytogenetics.

Slide test for sickle cell Haemoglobin, Hb typing on Paper electrophoresis. BCB dye test for G6PD enzyme deficiency, Hb estimation by Haemoglobinometer. Starch - agarose gel electrophoresis for Hb and G6pD systems, single chemical screening tests and paper chromatography for screening amino acidurias.

Analysis of Interphase Nuclei - Buccal smear and blood smear Preparation of Karyotypes of normal male and female Metaphase drawing

# ADIKAVI NANNAYA UNIVERSITY Structure of HUMAN GENETICS under CBCS w.e.f. 2015-16 ADMITTED BATCH

Year	Semester	Paper	Title	Hours	Marks	Credits
III	VI	VII (A)	HUMAN POPULATION GENETICS & GENETIC COUNSELLING	3	100	03
			Practical	2	50	02
		VIII-A	Cluster Elective-A VIII-A-1: STEM CELL TECHNOLOGY	3	100	03
			VIII-A-2 : MOLECULAR PATHOLOGY IN HUMAN DISEASES	3	100	03
			VIII-A-3: HUMAN GENOME PROJECT AND GENOMES	3	100	03
			VIII-A-1 : Practical VIII-A-2 : Practical VIII-A-3: Practical	2 2 2	50 50 50	02 02 02

# ADIKAVI NANNA UNIVERSITY CBCS PATTERN HUMAN GENETICS

## With effect from 2015-16 AB

## Semester – VI

## ELECTIVE : A : (Paper VII): HUMAN POPULATION GENETICS & GENETIC COUNSELLING

## Unit – I

- 1. Fundamental factors of population genetics matting pattern. Mendelian population, Mutation, Selection, Migration and genetic drift.
- 2. Consequences of inbreeding and Estimation of Inbreeding Coefficients.

## Unit – II

- 1. Consequences of Random mating Hardy- were berg considering 2 alleles at a locus and multiple alleles.
- 2. Estimation of gene, genotype and phenotype frequencies with examples of autosomal diallelic and Multiple allele characters.
  - Gene count method
  - Square root method

## Unit – III

- 1. Introduction to genetic counselling
- 2. Pedigree analysis and modes of inheritance
- 3. Importance of mutation, genetic heterogeneity, pleiotropim, phenocopy, penetrance & Expressivity

## Unit – IV

- 1. Identification of genetic defects Pre implantation diagnosis
  - Pre natal diagnosis
  - Invasive amniocentesis
    - Chorionic villi sampling
    - $\alpha$  fetoprotein assay
    - Non Invasive Ultrasonography
  - Post- natal diagnosis
    - Clinical observation, Biochemical studies, Cytogenetic studies Serological studies, Molecular studies

## Unit – V

 Process of genetic counseling Educating the counselee Presenting the Risks, options and guidance Gene therapy

- 1. Estimation of gene and genotype frequencies by using gene count method
- 2. Estimation of gene and genotype frequencies by using square root method
- 3. Pedigree analysis of single gene characters
- 4. Preparation of ideogram for
- Normal Male
- Normal Female
- Abnormal Kayotype

# ADIKAVI NANNA UNIVERSITY CBCS PATTERN HUMAN GENETICS

## With effect from 2015-16 AB

### Semester – VI

## CLUSTER ELECTIVE : (Paper VIII) A1: : STEM CELL TECHNOLOGY

#### **Unit I Embryogenesis**

- 1.1 Embryogenesis gametogenesis oogenesis and spermatogenesis; fertilization and development- molecular events of fertilization, activation of sperm motility, gamete fusion; early embryogenesis
- 1.2 Model organisms for developmental studies genetics of axis specification
- 1.3Mammalian development
- 1.4 Organogenesis and developmental plasticity

## Unit 2 Biology of stem cells

- 2.1 Historical perspectives, concept of stem cells
- 2.2 Cellular and molecular features of stem cells
- 2.3 Regulation of stem cells, self-renewal and molecular markers
- 2.4 Derivation, differentiation and propagation of stem cells

### Unit 3 Types of Stem Cells

- 3.1Embryonic stem cells and germ stem cells
- 3.2 Fetal adult stem cells and cancer stem cells
- 3.3 New generation stem cells
- 3.4 Induced pluripotent stem cells and patient specific stem cells

#### Unit 4Lineage specific differentiation of stem cells

- 4.1Stem cells, progenitors and their differentiation
- 4.2 Stem cell differentiation specific culture systems
- 4.3Molecular phenotyping and cell propagation enrichment strategies

## **Unit 5Applications**

- 5.1Medical need for stem cells and preservation of stem cells
- 5.2.Genetically engineered stem cells for gene therapy
- 5.3 Stem cell therapy neurodegenerative disorders, cardiovascular disorders, metabolic disorders, hematopoietic disorders, organ disorders, autoimmune disorders, reproductive failures

- 1. Study of early development of chick.
- 2. Mounting of chick embryos during development.
- 3. Morphogenetic movements in chick in vivo experiment.
- 4. Study of imaginal discs in Drosophila
- 5. Culturing cells aseptic techniques, media
- 6. Subculturing and cell lines
- 7. Cryopreservation
- 8. Quantitation –cell count,
- 9. Quantitation cytotoxicity & cell viability,

# ADIKAVI NANNA UNIVERSITY CBCS PATTERN HUMAN GENETICS With effect from 2015-16 AB

## Semester – VI CLUSTER ELECTIVE : (Paper VIII): A 2 –MOLECULAR PATHOLOGY IN HUMAN DISEASES

#### Unit 1Human diseases I

- 1.1 Etiology, pathology and symptoms of genetically inherited diseases PKU, alkaptonuria, galactosemia, Von Gierke disease, LeschNyhan syndrome, Gout, sickle cell anaemia, beta thalassemia, diabetes
- 1.2 Mode of infection, symptoms and epidemiology of disease causes by viruses (HIV, Hepatitis B, Rabies, HSV-1)
- 1.3Mode of infection, symptoms and epidemiology of disease caused by bacteria typhoid, syphilis, TB

## Unit 2.Human diseases II

- 2.1 Mode of infection, symptoms and epidemiology of disease caused by fungi aspergillosis, histoplasmosis.
- 2.2 Mode of infection, symptoms and epidemiology caused by protozoa –malaria, amoebiasis.
- 2.3 Cancer genetics tumor suppressor genes, oncogenes, Molecular basis of oncogenesis

## Unit 3Basic Instrumentation principles and techniques

- 3.1Principles of electrophoresis and immunoblotting
- 3.2 Principles of DNA sequencing and methods of genotyping and mutation analysis
- 3.3Principles and applications of PCR
- 3.4In situ hybridization techniques ISH, FISH

## Unit 4Genetic testing for hereditary disorders

- 4.1 Genetic testing for thalassemia
- 4.2 Genetic testing for familial colorectal cancer
- 4.3 Genetic testing for familial breast and ovarian cancer
- 4.4EGFR mutation in lung cancer, HER2 amplification in breast cancer, FISH test for early bladder cancer detection,KRAS mutation detection for colorectal cancer

## Unit 5Molecular diagnosis of infectious diseases

- 5.1 Principles of HPV testing and methods of genotyping
- 5.2 Hepatitis B virus infection testing for viral load and HBV DNA mutants detection
- 5.3 Molecular techniques -NestedPCR, Real Time PCR for different clinical applications

- 1. Sterile techniques, Pipetting, Preventing contamination.
- 2. Extract and assess the purity of DNA.
- 3. Agarose gel electrophoresis
- 4. Set up PCR.
- 5. Evaluate Southern blot data
- 6. Analyze PCR product using agarose gel electrophoresis and interpret results
- 7. Demonstration of karyotyping
- 8. Isolate cellular RNA, purify mRNA
- 9. Set up RT-PCR using commercial kit
- 10. Analyze RT-PCR results by agarose gel.

# ADIKAVI NANNA UNIVERSITY CBCS PATTERN HUMAN GENETICS With effect from 2015-16 AB

#### Semester – VI

#### **CLUSTER ELECTIVE : (Paper VIII): A3 : HUMAN GENOME PROJECT AND GENOMES**

#### **Unit 1Genome Organization and Studying Genomes**

1.1Genome – general features, features of eukaryotic nucleargenomes1.2Genomes, transcriptomes and proteomesgenomes

1.3Genome diversity – significance of genomes – bacteria, yeast, Caenorhabditis, Homo sapiens, Arabidopsis.

#### **Unit 2 Mapping Genomes**

2.1Genetic mapping – pedigree analysis, DNA markers – RFLPs, SSLPs, SNPs
2.2 Physical mapping – restriction mapping, FISH, radiation hybrid mapping, STS mapping

2.3 Sequencing genome- assembly of contiguous DNA sequence, shotgun method, clonecontig method, whole-genome shotgun sequencing

#### **Unit 3Genome Projects**

3.1Human genome project, HapMap Project, 1000 genome project, ENCODE project 3.2 Other genome projects.

3.3Applications and proposed benefits of HGP -ELSI.

#### **Unit 4Understanding Genome sequence**

4.1 Locating the genes in a genome sequence

4.2Determining the functions of individual genes

4.3. Transcriptome - microarrays

4.4 Proteome – protein profiling

#### **Unit 5 Molecularphylogenetics**

5.1Phenetics and cladistics

5.2 Reconstruction of DNA based phylogenetic tree

5.3Applications of molecular phylogenetics – evolutionary relationship between humans and primates; origin of AIDS; human prehistory.

- 1. Isolation and purification of genomic DNA.
- 2. Detection of SNPs using SNP specific primers and PCR.
- 3. Study of VNTR's in human genome as the polymorphic loci.
- 4. Design primers for PCR based detection of the gene and mapping primers on the genome
- 5. Introduction to NCBI websites
- 6. Introduction to database: protein data bank, nucleic acid database, Genbank .
- 7. Web based analysis to retrieve a nucleotide sequence from NCBI,
- 8. Sequence alignment using BLASTn, BLASTp, CLUSTALW.
- 9. Gene finding tools GenScan, GLIMMER
- 10.Introduction to proteomics Protparam, GOR, unPredict, SWISSMODEL.
- 11. Visualization software Rasmol
- 12. Generating phylogenetic tree using PHYLIP