# 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**<br>**VIII-A-1 :** STEM CELL TECHNOLOGY<br>. | **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**<br>**VIII-A-2 : Practical**<br>**VIII-A-3: Practical** | **2**<br>**2**<br>**2** | **50**<br>**50**<br>**50** | **02**<br>**02**<br>**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

**Practicals**

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

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### 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**

2.1Historical 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

**Practicals:**

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,

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### 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 PCRfor different clinical applications

**Practicals:**

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.

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**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 nuclear genomes

1.2Genomes, transcriptomes and proteomes

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.

**Practicals:**

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