

Andhra University  
Department of Human Genetics: College of Science & Technology.

**M.Phil – Pre Ph.D. Syllabus**  
(Effective from the Admitted Batch after June, 2009)

**PAPER – I – ADVANCES IN HUMAN GENETICS**  
**(Compulsory)**

The Human Chromosomes :

Simple staining methods – Special staining methods – Individual variations.

Structural differences along the chromosomes : Introduction, Repetitive DNA – Cytological localization of repetitive DNA Differences in base composition of DNA – differences in the protein components – packing differences – DNA replication patterns. Genetic mapping in human chromosomes.

Fine structure of chromosomes : Introduction – Structure and arrangement of fibrils – Single standard and multi stranded chromatids – major Coits – Giemsa bands, inter band zones and secondary constructions – bridges between chromatids and chromosomes – Centromeric region.

Formal Genetic of Man :

Mendel's modes of inheritance and their application to humans; hardy – Weinberg law and its applications.

Linkage analysis in humans : Pedigree method and cell hybridization.

Genetic polymorphism and diseases; Mutagenesis and carcinogenesis.

Natural Selection and genetic polymorphisms

Human Evolution : Chromosome Evolution.

Protein Evolution.

Behaviour Genetics :

Genetic variability of neurotransmitters

Hormone action

Genetics of EEG

Genetics of effective disorders and Schizophrenia.

### Genetic Counseling :

- a). Detection of genetic disorders; Prenatal diagnosis; and Amniocentesis.
- b). Case illustrations of common problems.
  1. Family history with Down's Syndrome
  2. Family history with X-linked recessive disease.
  3. Achandroplasia
  4. Huntington Disease
  5. Previous child with undiagnosed mental retardation
  6. Previous child with undiagnosed multiple malformation syndrome.
  7. Previous child with congenital heart disease.
  8. First cousin marriage.
  9. Erythroblastosis fetalis
  10. Family history of breast cancer.

### Genetic Screening :

Genetic manipulation / Genetic Engineering  
Biologic future of Mankind.

### **Text Books :**

1. Chromosomes in Mitosis and Interphase: H.G. Schwarzacher.
2. Molecular structure of Human Chromosomes. Ed. By J.J. Yunis.
3. Human Genetics : F. Vogel and A.G. Motulsky.
4. Human Biology : J.M. Tanner, N.A. Barnicot, G.A. Harrison.
5. Clinical Genetics – A Sorsby.
6. Genetics and Medicine – Thompson & Thompson.
7. The Principles of human Biochemical Genetics – H. Harris.
8. The Biochemical genetics of man – D.J.H. Brock & O. Mayo.
9. The Metalbolic basis of inherited disease – J.B. Stanbury, J.B. Wyngarden, D.S. Fredrickson, J.L. Goldstein and M.S. Brown.
10. The Genetics of Human Populations – Cavalli – Sforja and Bodmer.

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**PAPER – II – SPECIAL PAPER INCLUDING RESEARCH METHODOLOGY**

**(A). HUMAN CYTOGENETICS**

**(Optional)**

The position of chromosomes within the cell : Introduction – peripheral position – Association of Nucleolar organizer chromosomes – constancy of chromosome position.

Molecular organization and function of human genome.

New approaches to Human Gene mapping by somatic cell genetics – somatic cell hybridization Human gene map – New mapping methods.

New chromosome techniques – Methods of banding fixed chromosomes – origin of chromosomal bands.

Banding patterns, chromosome polymorphism, and primate evolution.

New chromosome techniques and their medical application – Significance and application of banding techniques – synchronization techniques.

Chromosomal abnormalities other than classical chromosome disorders. Syndromes involving chromosome 4, 8, 9, 11, 12, 20 and 22 – Abnormal chromosomes 14 and 15 in Abortion syndromes and Malignancy.

Prenatal genetic diagnosis and genetics counseling : Prenatal diagnosis of cytogenetics disorders – Amniocentesis – Chorionic Villi sample (CVS) – Genetic counseling.

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Centrifugation techniques (Basic Principles of sedimentation, centrifuges and their use; Design and care of preparation of Motors; separation methods in ultracentrifuges, Density gradient separation).

Molecular cytogenetic techniques (FISH, gene mapping (Physical), In situ hybridization,

**Text Books :**

1. Chromosomes in Mitosis and Interphase : H.G. Schwarzachr.
2. Molecular structure of Human Chromosomes : Ed. By. J.J. Yunis.
3. New chromosome syndromes : E.D. By J.J. Yunis.
4. Clinical Atlas of Human chromosomes Jean De Grouchy and C. Turlean.
5. Human Genetics : F. Vogel and A.G. Motulsky.

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**(B). HUMAN BIOCHEMICAL GENETICS**

**(Optional)**

1. The concept of Biochemical Polymorphism.
  - a). Red cell blood group Polymorphism – ABO, Rh, MNS
  - b). Red cell enzyme Polymorphism - ACP1, ESD, GLO1
  - c) Protein Polymorphism : HP, GC, Alb, TF.
  - d) DNA Polymorphism.
2. Enzyme and Protein diversity in human population ; common and rare alleles :  
the  
extent of allelic variation; the cause of allelic diversity.
3. The blood group substances : Biochemical path ways of ABH substances.
4. The Hemoglobin : Structure, Synthesis and functions HB; structural variants and  
variants of synthesis of globin chairs.
5. Inborn errors of metabolism : Disorder of carbohydrate metabolism; disorders of  
aminoacid metabolism; hypo several enzyme deficiencies, miscellaneous  
disorders.
6. Immunogenetics : the immune respects basic concepts ; the innate immune  
system : the adaptive immune system. Immunoglobulins ; (cellular and adoptive  
systems). The major histocompatibility complex (Class – I, II and III),  
complement system; immuno deficiencies diseases; Anti immunity ; acquired  
immuno deficiencies ; molecular immuno genetics : DNA level studies in HLA  
reptain.

7. Pharmacogenetics : G6PD; Pseudocholinesterase; Issomiaid inactivation.
  8. Ecogenetics : carcinogenesis ; a-1-antitripsin deficiency – foods.
  9. Gene mutations and inherited diseases :  
The molecular pathology of inherited disease : dominance and recessivity ; hetrogenity of inherited disease: heredity and environment.
  10. Heterozygote detection.  
Treatment of inherited metabolic disease.
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11. Blood grouping and serological techniques; Electrophoretic methods for the detection of protein polymorphisms. DNA and RNA isolation methods. PCR methods, Restriction fragment length polymorphisms
  12. Chromatographic Techniques (General principles low pressure column chromatography, HPLC, Adsorption chromatography, Ion – exchange chromatography, Affinity chromatography, Gas liquid chromatography (GLC), Thin layer chromatography (TLC) paper chromatography.

**Recommended Books :**

1. Principles of Human Biochemical Genetics – harries
2. The Biochemical genetic of Man – D.H.J. Brock & O. Mayo.
3. Human Genetics – F. Vogal and A. G. Motuloky
4. Blood groups in Man - Race and Sanger.
5. Genetics in Redicius – Thousand Thousenson.
6. Immunoben – P.M. Lydyard, A. Whelan and M.W. Fanger.

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**(C). HUMAN POPULATION GENETICS**

**(Optional)**

Genetic equilibrium

Natural selection leading to

- i) Changes of gene frequencies in one direction.
- ii) Genetic equilibrium
- iii) Unstable equilibrium

Frequency dependant and density dependant selection. Selection due to infectious diseases.

Consanguinity and its genetic consequences

Genetic load

Genetic distance

Gene diversity

Gene flow

Genetic drift.

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Sampling Techniques :

Analysis of Variance and co-variance : One way and two ways classification.

Multivariate methods : Simple partial and Multiple correlations, Multiple regression, path analysis and factor analysis; Multiple classification analysis.

LOD Scores, 'Z' Scores,

Chi-square test, Critical ratio, 't' test.

**Recommended Books :**

1. The genetics of human populations – by L.L Cavalli – Sforza and W.F. Bodmer.
2. Human Genetics – by F. Vogel and A.G. Motulsky
3. An Introduction to population Genetics theory – by J.F. Crow and Kumura, M.
4. Population Genetics – by C. C. Li.

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**(D). MOLECULAR GENETICS**

**(Optional)**

Recombinant DNA technology :

Different enzymes and vectors used in genetic engineering.

Constructing gene libraries.

Digesting genomic DNA; Ligating DNA Molecules; Genomic DNA libraries cDNA libraries.

Screening gene libraries :

Immunochemical methods; Nucleic acid hybridization.

Expression of cloned genes :

Expression in bacteria, Expression in yeast; Expression in eukaryotic cells  
applications of Recombinant DNA technology.

Vaccines ; Monoclonal antibodies: Protein engineering : Transgenics.

Structural analysis of gene

Analysis and annotation Databases: Sequence comparison Functional analysis of  
gene

Allelic replacement and gene knock-outs; Complementation Studying gene function  
through protein interactions; Antisense RNA

Whole genome analysis :

Genetic mapping of human chromosomes

Physical mapping of the human genome

Molecular diagnosis of infectious diseases

Molecular diagnosis of genetic diseases.



Cloning human disease genes :

Functional gene cloning : Positional gene cloning ; Candidate gene cloning  
Positional candidate gene cloning .

Human gene therapy :

Ex vivo gene therapy; In vivo gene therapy ; Viral gene delivery system

Nonviral gene delivery system ; pro drug activation therapy

Nucleic acid therapeutic agents; Lligonucleotide correction of genetic conditions

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Isolation of Nucleic acids (DNA & RNA) Techniques of DNA – RNA analysis (PCR <  
Multiplex genomic analysis, reverse RTPCR, Primer extension pre-amplification. Analysis  
of extracted and amplified DNA (Southern blot, Allele specific oligonucleotide probes, Dot  
blot, In situ hybridization, RFLP, Northern blot).

Techniques to screen for mutations in genomic DNA (SSCP, SNP's Denaturing  
gradient gel electrophoresis, Automated sequencing).

### **Reference Books :**

1. Human Molecular Genetics – T. Strachan and AP Read.
2. Principles of Genetics – DP. Snustad & M.J. Simmons
3. Recombinant DNA – JD Watson, M. Gilman, J. Witkowski & M. Zoller
4. Molecular Biotechnology – Principles and Applications – B.R. Glick & U.  
Pastermak
5. Introduction to Molecular Medicine – D.W. Ross
6. From Genes to genomics – J.W. Dale & M.V. Schantz
7. Genetic Engineering – Sandhya Mitra.

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**(E). GENETIC COUNSELING AND SCREENING**

**(Optional)**

Genetic Counseling

Scope of Genetic Counseling – General approach to Genetic Counseling

Counseling for different Genetic Disorders.

(Counseling for dominant defective phenotypes

Counseling for recessive defective phenotypes

Counseling for X – linked defective phenotypes

Counseling for Multi-factorial defective phenotypes

Counseling for chromosomal related defects).

Disputed Paternity

Genetic Screening

Scope of Genetic Screening

Pre natal and post natal Genetic Screening. Methods and Diagnosis

(Amniocentesis – Chorionic Villus Sampling – Ultrasonography-fetoscopy – maternal blood sampling)

Risk calculations for different genetic disorders

(Autosomal Dominant, Autosomal recessive, Sex linked recessive and Multi-factorial disorders)

Gene Therapy-classification -different types of gene therapy.

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Blood grouping and serological techniques; Electrophoretic methods for the detection of protein polymorphisms.

Banding techniques (G, C and Q banding), FISH, In situ hybridization techniques

DNA and RNA isolation methods, PCR methods, RFLP's, SNP's

**Text Books :**

1. Human Molecular Genetics : Strachan T and A.P. Read
2. Human Genetics : F. Vogel and A.G. Motulsky
3. Genetic Engineering : Sandhya Mitra.
4. Medical Genetics : Jorde etal
5. Genetic Counseling : W. Fuhman and F. Vogel
6. Genetic in Medicine : Thompson and Thompson.