ANDHRA UNIVERSITY

DEPARTMENT OF HUMAN GENETICS

PROGRAM : M.SC HUMAN GENETICS

REGULATION AND SYLLABUS

EFFECTIVE FROM 2020-2021 BATCH

Program Outcomes:

PO1. The M.Sc Human Genetics course in Andhra University is to stimulate the students to the fact that as we go down the scale of magnitude from cells to organelles to molecules for the better understanding of various genetic disorders or diseases.

PO2. To train the students in basic and advanced areas of Human Genetics, focusing on the recent trends of the discipline with particular emphasis on the practical aspects.

PO3. Students laboratory training will empower them to register in research institutions for their career.

PO4. To stimulate scholarly progression and intellectual development of the program educating the students constantly with excellence.

PO5. To promote talent and personality development, to encourage self-confidence and self-reliance in the student to achieve their goals.

Program Specific Outcomes:

PSO1. To pervade the latest advances in human genetics by organizing conferences, symposia, workshops and webinars.

PSO2. To furnish student accessibility to online journals.

PSO3. To update the curriculum that suits the needs of competitive examinations

PSO4. To deliver skilled manpower suitable for different government and non-governmental organisations.

PSO5. Students can compete in National level competitive exams such as UGC-CSIR, NET-JRF, GATE and APSET.

PSO6. Students gain familiarity in various practical methods and analytical techniques enabling them to pursue higher education.

PSO7. The Master's degree in human genetics will address the increasing need for skilled scientific manpower with an understanding of global research applications in the field of basic and applied human genetics.

SEMESTER I

PAPER 1.1- BASIC HUMAN GENETICS

Course Outcomes:

CO1. To introduce the students the main concepts of basics of Human Genetics.

CO2. To enhance knowledge towards the Mendels laws of inheritance and extension of Mendelism.

CO3. To study the gene interactions, modifying genes, polygenic inheritance and multifactorial inheritance.

CO4. To know the construction of human pedigree, modes of inheritance and mitochondrial inheritance.

CO5. To have better understanding about the linkage and crossing over, genetic and physical mapping.

CO6. To get information about twin studies, Eugenics, Euthenics and Euphenics.

Learning outcomes:

LO1. Acquired knowledge on the importance of classical Mendelian genetics theories and extension of Mendelism.

LO2. Enhanced knowledge on gene interactions, modifying genes, polygenic inheritance and multifactorial inheritance.

LO3. Gain knowledge on ancestral history and construction of human pedigrees related to genetic disorders.

LO4. Obtained better understanding in modes of inheritance and mitochondrial inheritance.

LO5. Get a hold on the linkage and crossing over, genetic and physical mapping.

LO6. To improve knowledge about twin studies, Eugenics, Euthenics and Euphenics.

Course Specific Outcomes:

CSO1. Basic Human Genetics subject generates scientific knowledge up to gene level and enhances the knowledge in the fundamental level of Human Genetics field.

CSO2. This subject gives huge research opportunities in multidisciplinary of life sciences.

CSO3. The scope of research underlying genetic disorders, behavioral and medical genetics will become enhanced with clinical trials of novel drug design, drug toxicity and efficacy.

Unit -I

Introduction to Genetics; Mendelism- Mendel and his experiments, Law of segregation Law of independent assortment; Chromosomal basis of segregation and independent assortment. Extensions of Mendelism-Allelic variation and gene function, Dominance relationships, basis of dominant and recessive mutations; Multiple allelism, allelic series.

Unit -II

Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity, phenocopy; lethal and sub lethal mutations; Gene interactions and modifying genes; Pleiotropy; Polygenic inheritance; Multifactorial inheritance

Unit -III

History of Human Genetics; Pedigrees- gathering family history, pedigree symbols, construction of pedigrees; Monogenic traits - Autosomal inheritance-dominant and recessive; Sex-linked inheritance- dominant and recessive; Sex-limited and Sex-influenced traits; Y-linked ; Mitochondrial inheritance

Unit -IV

Linkage- Linkage mapping , Linkage analysis; crossing over – Types of crossing over, Mechanism of crossing over, Factors affecting crossing over ;Genetic mapping ; Physical mapping.

Unit -V

Heredity and environment (twin studies), Eugenics-history, Modern Eugenics, types of Eugenics; Euthenics and Euphenics.

Books suggested:

1. Principles of Human Genetics - Curt Stern
2. Human Genetics - Mckusick V. A
3. Basic Human Genetics –Mange E.J and Mange A.P
4. Principles of Genetics - Gardner et al.
5. Principles of Genetics - Snustad et al.
6. Genetics in Medicine-Thompson and Thompson
7. Genetics, Molecular Biology, Evolution and Ecology-Verma P.S and Aggarwal V.K

SEMESTER I

PAPER-1.2: POPULATION GENETICS AND BIOSTATISTICS

Course Outcomes:

CO1. To introduce the students the main concepts of population genetics and bio-statistics

CO2. To get knowledge the Hardy-Weinberg principle and its importance in population genetics

CO3. To understand the concept of mating patterns, inbreeding coefficient and genetic polymorphism.

CO4. To understand the main concepts of genetic demography and sampling techniques.

CO5. To have an insight in measures of central tendency and measures of dispersion

CO6. To develop an understanding on probability, correlation, regression and significance of chi-square test and "t"test.

Learning Outcome:

LO1. Acquire knowledge on the Hardy-Weinberg principle and the skill of calculation of gene frequencies and genotype frequencies.

LO2. Get a hold on the concept of mating patterns, inbreeding coefficient.

LO3. Obtain knowledge on the concept of genetic polymorphism and types.

LO4. Learn about various sampling techniques, classification of data and tabulation.

LO5. Acquire the skill of calculation of measures of central tendency and measures of dispersion.

LO6. Get deep insight on probability, correlation, regression and significance of chi-square test and "t"test.

Course Specific Outcomes:

CSO1. Acquire knowledge on the main concepts of Population Genetics and Bio-statistics.

CSO2. Able to analyze the Hardy-Weinberg principle, calculation of gene frequencies and genotype frequencies.

CSO3. Able to identify the types of consanguineous marriages and learn calculation of inbreeding coefficient.

CSO4. Familiar with the importance and significance of tests like chi-square test and "t"test.

Unit – I

Mendelian Population and scope of population genetics. Gene and genotype frequencies, Mating patterns, Random and Non-random mating, Hardy-Weinberg principle, Extension of H-W principle to multiple alleles, sex-linked alleles. Factors that change Hardy Weinberg Equilibrium. Inbreeding and Inbreeding coefficient.

Unit – II

Genetic polymorphism, Types of Genetic polymorphisms, Examples for Genetic Polymorphism. Application of population genetics and role of population genetics in Human Genetics. Genetics of origin and evolution of Human races. Genetic Demography, Birth rate, Death rate, Reproductive rate. Index of opportunity for natural selection.

Unit – III

Bio-Statistics: Definition, basic concepts of biostatistics, Collection of data .sampling techniques, classification of data and tabulation. Frequency distribution, Graphic presentation of data and Diagrammatic presentation of data.

Unit-IV

Measures of central tendency- mean median and mode. Measures of Dispersion - range, mean deviation, standard deviation and variance

Unit – V

Probability – laws of probability for independent events – permutations and combinations– conditional probability. Binomial distribution. Correlation and regression, Tests of significance – chi square test and‘t’ test.

BOOKS SUGGESTED:

1. The Genetics of Human Populations by LL Cavalli-Sforza and WF Bodmer
2. Population Genetics Theory by James F. Crow and W. Kimura Harper and Row,
3. Introduction to Biostatistics by P.S.S. Sundara Rao and J. Rich,
4. Introduction to Biostatistics by Robert R. Sokel and James F. Kohlf

SEMESTER I

PAPER 1.3 - HUMAN CYTOGENETICS

Course Outcomes:

CO1. To introduce the students the main concepts of human cytogenetics.

CO2. To study about the history and the standardization of human cytogenetics.

CO3. To understand the importance of karyotyping and banding techniques.

CO4. To get comprehensive knowledge on numerical and structural abnormalities of human chromosomes with examples.

CO5. To get detailed information about gene mapping techniques.

CO6. To know about different reproductive disorders, infertility and recurrent pregnancy loss.

Learning Outcomes:

LO1. Get deep insight about the history of human cytogenetics.

LO2. Obtained knowledge about the standardization of human cytogenetics.

LO3. Gain knowledge about the chromosomes, Karyotyping and banding techniques.

LO4. Get a hold on numerical and structural abnormalities of human chromosomes and their clinical importance.

LO5. Acquire knowledge about different mapping techniques.

LO6. Obtain knowledge on reproductive disorders, infertility and recurrent pregnancy loss, which are very challenging problems of the present situation.

Course Specific Outcomes:

CSO1. Acquire working knowledge about the preparation of karyotyping.

CSO2. Gain practical knowledge about the human leucocytes blood culture technique.

CSO3. Able to analyze and interpret the results of banding techniques.

CSO4. Able to identify the hetero chromatin from buccal smear test.

Unit – I

History and growth of Human Cytogenetics; Morphological variability of the human chromosomes; Karyotyping; Banded chromosomes and individual characterization of the human chromosomes; Standardization in Human Cytogenetics - Old classification and New classification.

Unit – II

The origin and transmission of chromosomal abnormalities- Non disjunction; Types of numerical chromosomal abnormalities; Examples – Down syndrome, Edward syndrome, Patau syndrome, Turner syndrome, Klinefelter syndrome, XXX- syndrome and XYY- syndrome.

Unit – III

Structural chromosomal abnormalities; Examples - Cri-du-chat syndrome, Wolf- Hirschhorn syndrome, Fragile X – syndrome; Chromosomal instability syndromes; Heterochromatin and Lyon's Hypothesis, Uniparental disomy and Genomic imprinting

Unit – IV

Gene mapping- Somatic Cell Hybridization (SCH); Molecular Cytogenetics - Fluorescence In Situ Hybridization (FISH); Comparative Genomic Hybridization (CGH); Array Comparative Genomic Hybridization (aCGH); Marker chromosomes; Human artificial chromosomes.

Unit – V

Disorders of sex development – Primary Amenorrhea, Gonadal dysgenesis, Testicular feminization, Pseudohermaphroditism, True hermaphroditism; Infertility - Genetic basis of male infertility, Genetic basis of female infertility; Chromosomal abnormalities in recurrent pregnancy loss.

Books suggested:

1. Human Cytogenetics (vol. I & II) – J.L. Hamerton

2. Human Chromosomes - E.H. Ford

3. Human Genetics – F. Vogel and A.G. Motulsky.

4. Basic Human Genetics – E.J. Mange and A.P. Mange.

6. New Chromosomal Syndromes - J.J. Yunis

SEMESTER II

PAPER 2.1- CELL BIOLOGY AND SYSTEM PHYSIOLOGY

Course Outcomes:

CO1. To introduce the students the main concepts of cell biology and system physiology.

CO2. To study about the cell structure and its organelles.

CO3. To get detailed information of cytoskeleton, plasma membrane and cytoplasmic matrix.

CO4. To impart knowledge on ultra structure of nucleus and cellular interactions.

CO5. To get comprehensive knowledge on mitosis and meiosis cell division, cell signaling and communication.

CO6. To know about the physiology of different systems working in the Human body.

Learning Outcomes:

LO1. Acquire knowledge about the cell structure and its organelles

LO2. Enhanced knowledge on detailed information of cytoskeleton, plasma membrane and cytoplasmic matrix.

LO3. Gain knowledge on ultra structure of nucleus and cellular interactions.

LO4. Obtained better understanding on comprehensive knowledge on mitosis and meiosis cell division.

LO5. Get deep insight into the cell signaling and communication.

LO6. Get a hold on the physiology of different systems working in the Human body.

Course Specific Outcomes:

CSO1. Acquire practical knowledge about the different stages of mitosis cell division.

CSO2. Able to analyze and interpret the different organelles of the cell.

CSO3. By gaining practical knowledge able to differentiate the stages of meiosis cell division.

CSO4. Get knowledge on cell signaling and communication.

Unit- I

Cell structure; Organelles of the cell- Golgi apparatus, Mitochondria, Ribosomes, Nucleus, Lysosomes, Peroxisomes, Endoplasmic reticulum, Vesicles, Cytoskeleton, Plasma membrane, Cytoplasmic matrix; Chemical constituents of cell- Carbohydrates, Llipids, Proteins, Nucleic acids, Vitamins, Minerals.

Unit- II

Ultra structure of nucleus-Nuclear membrane, Nucleolus, Chromatin composition and organization;Cellular interactions- Differentiations of the Cell membrane, Intercellular communication and gap junctions, cell coat and cell recognition, The cell surface of cancer Cells.

Unit- III

Cell division- Mitosis and Meiosis; Cell cycle and its regulation; Cell death (apoptosis, cell aging); Cell signaling and communication- Signal transduction: Cell signaling hormones and receptors, Cell surface receptors, Signaling through G-protein coupled receptors, Signal transduction pathways.

Unit-IV

Circulatory system – Blood, Anatomy of the heart, Cardiac cycle, Blood pressure, Neural and chemical regulation; Respiratory system – Structure and function of lungs; Neural and chemical regulation; Nervous system – Brain, Spinal cord and Neurons, Central and Peripheral nervous system, Action potential and regulation; Skeletal and Muscular system.

Unit- V

Digestive system – Digestion, Absorption; Excretory system – Structure and function of kidney, Regulation.; Endocrine system – Endocrine glands, Mechanism of hormone action, Neuroendocrine regulation; Reproductive system – Female and male reproductive systems, Reproductive processes.

Books suggested:

1. The Cell, A Molecular Approach- G. M. Cooper and R. E. Hausman

2. Molecular Biology of the Cell - Bruce Alberts et al.

3. Cell and Molecular Biology- E.D.P. De Robertis and E.M.F. De Robertis, JR.

4. Molecular Cell Biology-Harvey Lodish et al.

5. Fundamentals of Anatomy and Physiology-F.H. Martini et al.

6. Gray’s Anatomy for Students –Raveendranath Veeramani

7. Guyton and Hall Textbook of Medical Physiology –J.E. Hall et al.

SEMESTER II

PAPER-2.2: HUMAN BIOCEMICAL AND IMMUNOGENETICS

Course Outcomes:

CO1. To introduce the student to the main concepts of Human Biochemical Genetics and Immunogenetics.

CO2. To study the biochemical polymorphisms such as enzyme and protein polymorphisms.

CO3. To understand the genetic causes of inborn errors of metabolism, inheritance patterns and heterogeneity.

CO4. To get the information about the basic concepts of Immunity system.

CO5. To study the structure and diversity of immunoglobulin molecules and the genetic basis of antibody diversity.

CO6. To learn the Major Histocompatability (MHC), HLA system and Immunodeficiency diseases.

Learning Outcomes:

LO1. Gain knowledge learn the enzyme polymorphisms and protein polymorphisms.

LO2. Acquire knowledge on the inborn errors of metabolism.

LO3. Obtain knowledge on the structure and functions of normal hemoglobin and hemoglobin disorders.

LO4. Acquire knowledge on the basic concepts of Immunity system.

LO5. Get deep insight into the structure and diversity of immunoglobulin molecules and the genetic basis of antibody diversity.

LO6. Get a hold on the Major Histocompatability (MHC), HLA system and Immunodeficiency diseases

Course Specific Outcomes:

CSO1. Acquire knowledge about the main concepts of human biochemical genetics and immunogenetics

CSO2. Learn and understand the structure and functions normal hemoglobin and hemoglobin disorders.

CSO3. Familiar with the importance of pharmacogenetics and ecogenetics.

CSO4. Gain knowledge of the basic concepts of immunity system and acquires the knowledge about the vaccines.

Unit – I

The concept of Biochemical polymorphism, Enzyme and protein polymorphisms –ACP, ESD, HP and GC systems; Hemoglobinopathies; Pharmacogenetics – Glucose- 6-Phosphate dehydrogenase deficiency ; Ecogenetics – Alpha -1- Antitrypsin.

Unit – II

Inborn errors of metabolism–Disorders of carbohydrate metabolism–Galactosaemia; Disorders of amino acid metabolism – Alkaptonuria, Phenylketonuria, Albinism and Homocystinuria,Disorders of Lysosomal enzymes–Tay- Sachsdisease and Mucopolysaccharidoses; Disorders of Lipoprotein and lipid metabolism – Hyper Lipoproteinemia; Disorders of Purine metabolism- Lesch Nyhan syndrome; Disorders of Pyrmidine metabolism – Orotic Aciduria

Unit – III:

The immune response – Basic concepts; the innate immune system–Phagocytes, the complement system, natural killer cells; The adaptive immune system – Cellular immune system, humoral immune system; Organization and structure of lymphoid organs – bone marrow, thymus, spleen and lymph nodes, Cells of the immune system – B Lymphocytes, T-Lymphocytes. T-cell receptor – structure and function

Unit-IV

Genetic basis of structure and diversity; Immunoglobulin molecules and the genetic basis of antibody diversity. Immunological memory. Immuno regulation. Adjuvants and immunological tolerance.

Nature of antigens and antibodies. Structure and function of antibodies. Isotypes, Allotypes and Idiotypes. Antigen – antibody interactions.

Unit-V

The Major Histocompatability complex-Class I (HLA-A,B,C,E,F & G),Class II (HLA-DP,DR,DQ) and Class III (Complement genes); Immunodeficiency diseases- Agamma-globulinemia, Severe combined immuno-deficiency, Ataxia telangiectasia, Wiskott- Aldrich syndrome.

Autoimmunity – Altering immune function (vaccines and transplants); immuno-therapy (monoclonal antibodies and cytokines), immunity breakdown (AIDS)

SUGGESTED READINGS:

1. Principles of Human Biochemical Genetics by H. Harris
2. Human Genetics by A.G. Motulsky and F. Vogel
3. The metabolic basis of inherited diseases by Scriver et al.
4. Medical Genetics by Lynn B. Jorde et al
5. Basic Immunogenetics - Fudenberg et al
6. Text book of Immunology - S.T, Barrot

SEMESTER II

PAPER 2.3 - MEDICAL GENETICS

Course Outcomes:

CO1. To introduce the students the concept of Medical Genetics.

CO2. To know about Medical Genetics and scope of Medical Genetics.

CO3. To understand the different types of the diseases associated with different parts of Human body like skin, skeletal system, muscles, eyes, jaws and ears.

CO4. To get detailed insight into the disease associated with different systems of Human body like digestive system, respiratory system, central nervous system, kidney and urogenital system, reproductive system and endocrine system.

CO5. To get comprehensive knowledge on life style disorders.

CO6. To get knowledge about neurodegenerative disorders.

Learning Outcomes:

LO1. Acquired knowledge about the medical genetics, its scope and different diseases associated not only with different pats but also with different systems of the human body.

LO2. Obtained knowledge on the different types of the diseases associated with different parts of human body.

LO3. Get deep insight into the disease associated with different systems of human body

LO4. Get a hold on the endocrine system.

LO5. Gain knowledge on different life style disorders which causes risk to the present population.

LO6. Obtain knowledge on different neurogenetic disorders which are challenging of the current situations.

Course Specific Outcomes:

CSO1. Acquire knowledge about the mode of inheritance of diseases related to different organs of the human body.

CSO2. Gain knowledge about the diseases associated with different systems of the human body.

CSO3. Able to analyze and interpret the different life style diseases.

CSO4. Able to identify the different life style disorders.

Unit – I

Scope of Medical Genetics; Skin- Ichthyosis, Psoriasis, Multiple neurofibromatosis, Porphyrias, Blooms syndrome; Skeletal system – Ankylosing spondylitis, Osteogenesis imperfecta, Rheumatoid arthritis; Osteoporosis; Marfans syndrome.

Unit – II

Muscle – Muscular dystrophies, Myotonia; Eye – Glaucoma, Cataract, Retinoblastoma ; Jaws – Hare lip and Cleft palate; Ears - Deafness.

Unit – III

Digestive system – Hypertrophic pyloric stenosis, , Cirrhosis of liver ; Respiratory system – Cystic fibrosis ; Cardiovascular system – Congenital heart disease; Central nervous system – Spina bifida, Anencephaly; Kidney and urogenital system – Cystinosis, Polycystic kidney ; Reproductive system- Polycystic ovary disease, Endocrine system – Congenital hypothyroidism or Cretinism, Goiter.

Unit – IV

Life style disorders- Diabetes, Hypertension, Hyperlipidemia, Coronary heart disease, Stroke, Obesity, Anxiety, Chronic obstructive pulmonary disease.

Unit – V

Neurodegenerative disorders- Parkinson's disease, Alzheimer's disease, Spinocerebellar ataxia, Prion disease; Neuromuscular disorders- Multiple sclerosis, Amyotrophic lateral sclerosis, Myasthenia gravis; Psychiatric disorders- Schizophrenia, Bipolar disorder.

Books Suggested:

1. Medical Genetics – Jorde et al.

2. Genetics and Medicine – M.W. Thompson et al.

3. Clinical Genetics – A. Sorsby

4. Genetic Disorders of Man – R. M. Goodman

5. Emery's Elements of Medical Genetics – R. F. Mueller and I.D Young

6. Human Genetics – F. Vogel and A.G. Motulsky.

SEMESTER III

PAPER 3.1 - GENETIC TOXICOLOGY AND CANCER GENETICS

Course Outcomes:

CO1. To introduce the students the main concepts of genetic toxicology and cancer genetics

CO2. To understand the fundamentals of genetic toxicology.

CO3. To study the Routes and sites of toxic exposure, different techniques used to detect gene toxicity and treatment.

CO4. To have an elaborative perception on cancer and its relation with epigenetic, complex factors involvement in inherited versus sporadic cancer formation.

CO5. To get information about the molecular modifications of proto oncogenes, tumor suppressor genes and carcinogens involved in formation of different types of cancers, their prevention, diagnosis and treatment.

CO6. To know about stem cell study in cancer therapy, angiogenesis, stochastic versus cancer stem cell model to explain the formation of cancer.

Learning Outcomes:

LO1. Able to develop the knowledge on chromosomal alterations and sister chromatid exchanges.

LO2. Get a hold on the basic sites of toxic exposure, their distribution, metabolism and excretion.

LO3. Get deep insight into the characteristics of cancer cells, chromosomes in neoplasias and relation of oncogenes to chromosomal defects.

LO4. Gain knowledge on Knudsons hypothesis and types of cancers.

LO5. Acquired knowledge on cancer prevention, diagnosis and treatment.

LO6. Obtain knowledge on cancer stem cells in cancer initiation and progression

Course Specific Outcomes:

CSO1. The subject illuminates the knowledge on the effects of Mutagens on the chromosomal alterations and DNA synthesis.

CSO2. The course enhances the knowledge on changes in alteration in proto-onco and tumour suppressor genes which play an important role in the development of cancer.

CSO3. Helps in understanding how somatic and germ line mutations can cause a cell to make proteins that effect the growth of the cells and cellular differentiation carried to cancer formation.

Unit – I

Origin of genetic toxicology; Historical prospective of genetic toxicology; Fundamentals of genetic toxicity; Mechanism of induction of chromosomal alterations and sister chromatid exchanges; Mutagens- chemical, physical, biological, environmental and food; Antimutagens.

Unit – II

Routes and sites of exposure; Absorption; Distribution and excretion of toxicants; Xenobiotic metabolism; Consequences of genotoxic effects in humans; Techniques to detect genotoxicity; Treatment.

Unit – III

Introduction to cancer; Characteristics of cancer cells; Chromosomes in neoplasias and relation of oncogenes to chromosomal defects; Chromosomal instability syndromes; Cancer as a genetic disorder and cancer in families; Inherited versus sporadic cancers; The role of Epigenetics in cancer.

Unit – IV

Molecular changes in proto- oncogenes; Tumor suppressor genes; Knudsons Hypothesis; Types of cancers – Retinoblastoma, Skin cancer, Lung cancer, Esophageal and Colon cancer, Brain cancer, Breast cancer, Cervical cancer, Prostate cancer ; Cancer and environment - Physical, Chemical and Biological carcinogens; Cancer prevention ; Diagnosis; Treatment.

Unit – V

Cancer stem cells: Introduction to stem cells, Angiogenesis, Stochastic Vs Cancer stem cell model for cancer formation, Cancer stem cells in cancer initiation and progression, Cancer stem cell pathways, Regulation by microRNAs , Therapeutic strategies.

Books suggested:

1. Molecular Biology in Medicine - Cox and Sinclair
2. Principles of Genetic Toxicology- David Brusick.
3. Genetic Toxicology- Albert P, Li, R.H, Heflich.
4. Principles and Practice of Medical Genetics- Rimoin et al.
5. Clinical Genetics- Robinson and Linden
6. Clinical Genetics: A Short Course – Wilson
7. Cancer Genetics E-book-Cubocube (Wasserman Laboratory, University of British Columbia (UBC)

SEMESTER III

PAPER- 3.3: Genetic Screening, Counseling and Gene Therapy

Course Outcomes:

CO1. To introduce the students the main objectives of genetic screening and counseling.

CO2. To study the scope of genetic screening, testing methods of genetic screening and also population screening for genetic diseases.

CO3. To get information on prenatal and postnatal screening methods.

CO4. To know about the scope and methods of genetic counseling.

CO5. To understand the importance of gene therapy and its classification.

CO6. To know about inheritance and their risk assessment.

Learning Outcomes:

LO1. Gain knowledge on the scope of genetic screening.

LO2. Get deep insight into testing methods of genetic screening and also population screening for genetic diseases.

LO3. Enhanced knowledge on the Prenatal screening methods and postnatal screening.

LO4. Acquire knowledge on scope of genetic counseling and methods of counseling, patterns of inheritance and risk assessment.

LO5. Obtain knowledge on gene therapy, its classification and its clinical applications.

LO6. Get a hold on the detection of autosomal recessive carriers.

Course Specific Outcomes:

CSO1. Acquire knowledge on prenatal and post natal screening tests like amniocentesis, ultra sonography, chorionic villous sampling etc., which are used for genetic testing before birth to identify whether the new born offspring is more or less likely to have certain birth defects.

CSO2. The course also helps to identify the genetic risk factors based on an expert review of personal and family health histories and moulds the students to be genetic counselors.

CSO3. The course has huge benefits on how to manage the health care and future perspectives of gene mapping and gene therapy, so that accurate treatment maybe available at gene level.

Unit - I

Scope of genetic screening-Invasive and Noninvasive testing methods - Prenatal and Postnatal screening. Population screening for genetic diseases, Newborn screening, carrier screening family screening and their medical importance

Unit - II

Prenatal screening methods- Amniocentesis- Chronic Villous sampling, Ultrasonography, fetoscopy, maternal blood sampling.

Postnatal screening - chromosomal abnormalities both structural and numerical cytogenetic disorders and molecular methods.

Unit - III

Scope of genetic counseling - methods of genetic counseling, educating the counselee, presenting the risks and options and guiding. Social, ethical and legal issues. Patterns of inheritance and risk assessment, chromosomal disorders, autosomal dominant and recessive disorders, X-linked disorders, multifactorial-polygenic disorders. Reproductive failures, consanguinity.

Unit - IV

History of Gene Therapy - classification of gene therapy- class-I, Class II and Class III. Types of gene therapy- germ line gene therapy and somatic gene therapy.

Unit - V

Detection of autosomal recessive carriers - Huntington's Disease - Restriction length polymorphisms (RFLPs) and DNA probes-Cystic fibrosis - Sickle cell Disease; Types of stem cells; Cord blood banking; stem cells therapy.

SUGGESTED READINGS:

1. Human Molecular Genetics by T. Strachan and AP Read
2. Human Genetics by F. Vogel and A.G. Motulsky
3. Genetic Engineering by Sandhya A. Mitra
4. Medical Genetics by Jordee et al
5. Genetic Counseling by W. Fuhramann and F. Vogel

SEMESTER III

PAPER 3.4 - GENOMICS, PROTEOMICS AND BIOINFORMATICS

Course Outcomes:

CO1. To introduce the students on the concepts of genomics, proteomics and bio-informatics.

CO2. To understand the genetic mapping and physical mapping techniques.

CO3. To know about the DNA sequencing techniques, bio -chips, DNA micro arrays, gene annotation, and gene structure predictions.

CO4. To get information about protein structure and its determination methods.

CO5. To study the protein chip arrays and functional proteomics.

CO6. To know about bioinformatics and biological databases.

Learning Outcomes:

LO1. Gain knowledge on concepts of genetic mapping and physical mapping techniques.

LO2. Acquire knowledge on the DNA Markers for genetic mapping and human genome project.

LO3. Get a hold on DNA sequencing techniques, structural and functional genomics.

LO4. Obtained knowledge on the Protein structure and its determination methods, structural hierarchy, domains, folds, and motifs.

LO5. Get deep insight into docking, drug design methods and pharmacogenomics.

LO6. Get knowledge on types of databases and database searching.

Course Specific Outcome:

CSO1. Acquire fundamental knowledge in Genomics, Proteomics and Bio- informatics.

CSO2. Gain working knowledge of these Biological databases tools and methods;

CSO3. Appreciate their relevance for investigating specific contemporary biological questions;

CSO4. Critically analyse and interpret results of their study

Unit – I

Introduction to genomics; Genetic mapping and Physical mapping techniques-Restriction mapping (fingerprint mapping and optical mapping); Fluorescent in situ hybridisation (FISH) mapping: DNA Markers (STS,RFLP,CAPS RAPD's,SNP's,AFLP) for genetic mapping; Human genome project.

Unit – II

DNA sequencing – Maxam gilbert and Dideoxy methods; Shot gun method; Cycle sequencing; Automated DNA sequencing ; NGS (Next generation sequencing); Bio chips; Bio-Sensors; DNA micro arrays; Gene annotation; Gene structure predictions; Gene ontology consortium recommendations; Structural and Functional genomics.

Unit – III

Protein structure and its determination -Structural hierarchy; Domains; Folds; Motifs. Secondary structure prediction methods; Fold recognition and abinitio structure prediction; Homology-comparative modelling of proteins.

Unit – IV

Protein chip arrays; Functional proteomics; Docking; Drug design- Computer aided drug design; Ligand-based drug design; and Structure-based drug design; Pharmacogenomics.

Unit V

Introduction to Bioinformatics –Bioinformatics – Online tools and offline tools. Biological databases. Types of data bases – Gen bank, Swiss port, EMBL, NCBL, and PDB. Database searching using BLAST and FASTA.

Books suggested:

1 Malcolm Campbell and Laurie J. Heyer Genomics, proteomics and Bioinformatics Benjamin Cummings 2002

2 Lynn B. Jorde et al Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics Wiley 2006

M.Sc. HUMAN GENETICS

(Course Structure - (w.e.f. 2011-2012)

(Little Modifications in BoS held on 31-07-2014)

Semester – I

Paper	Title of the paper	Internal	Semester End	Total	Credits
1.1	Basic Human Genetics	20	80	100	4
1.2	Population Genetics and Biostatistics	20	80	100	4
1.3	Human Cytogenetics and Reproductive Genetics	20	80	100	4
1.4	Molecular Genetics and Molecular mechanisms of Human Pathogens	20	80	100	4
1.5	Practical - I	--	100	100	2
1.6	Practical - II	--	100	100	2

Semester – II

Paper	Title of the paper	Internal	Semester End	Total	Credits
2.1	Developmental and Behavioral Genetics	20	80	100	4
2.2	Human Biochemical and Immunogenetics	20	80	100	4
2.3	Medical and Cancer Genetics	20	80	100	4
2.4	Molecular Genetics and Human Genome Project	20	80	100	4
2.5	Practical - III	--	100	100	2
2.6	Practical - IV	--	100	100	2
2.7	Fundamentals of Human Genetics (Non-Core/Optional Paper)	20	80	100	4

Semester – III

Paper	Title of the paper	Internal	Semester End	Total	Credits
3.1	Clinical Genetics and Genetic Toxicology	20	80	100	4
3.2	DNA Technology and Genetic Engineering	20	80	100	4
3.3	Genetic Screening, Counseling and Gene Therapy	20	80	100	4
3.4	Genomics and Proteomics	20	80	100	4
3.5	Practical -V	--	100	100	2
3.6	Practical -VI	--	100	100	2
3.7	Advanced Human Genetics (Non-Core/Optional Paper)	20	80	100	4

Semester – IV

4.1	Dissertation based on Project work	200	8
4.2	Comprehensive Vivo – Voce	100	4

Grand Total I – IV Semesters 2300 - 80

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SEMESTER – I

PAPER–1.1: BASIC HUMAN GENETICS

Unit -I

Introduction to Genetics; Mendelism- Mendel and his experiments, Law of segregation, Law of independent assortment; Chromosomal basis of segregation and independent assortment. Extensions of Mendelism-Allelic variation and gene function- Dominance relationships, basis of dominant and recessive mutations; Multiple allelism, allelic series.

Unit -II

Genotype to phenotype: effect of the environment on phenotype development- Penetrance and expressivity, phenocopy; lethal and sub lethal mutations; Gene interactions and modifying genes; Pleiotropy; Polygenic inheritance; Multifactorial inheritance

Unit -III

History of Human Genetics; Pedigrees- gathering family history, pedigree symbols, construction of pedigrees; Monogenic traits - Autosomal inheritance-dominant and recessive; Sex-linked inheritance- dominant and recessive; Sex-limited and sex-influenced traits; Y-linked ; Mitochondrial inheritance

Unit -IV

Linkage and crossing over – types of crossing over ; Genetic and Physical mapping; heredity and environment (twin studies).

Recommended Books				
1.	Atherly *et al*	The Science of Genetics	Saunders	1999
2.	EJ Mongia and AP Mongia	Basic Human Genetics		
3.	Fairbanks *et al*	Genetics	Wadsworth	1999
4.	Gardner *et al*	Principles of Genetics	John Wiley	1991
5.	Snustad *et al*	Principles of Genetics	Wiley and sons	1998
6.	Griffiths *et al*	An Introduction to Genetic Analysis	Freeman	2004
7.	Curt stern	Principles of Genetics		
8.	Snustad *et al*	Principles of Genetics	Wiley and sons	1998
9.	Strickberger	Genetics	Mcmillan	1985
10.	Thomson and Thomson	Genetics in Medicine		

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PAPER-1.2: POPULATION GENETICS AND BIOSTATISTICS

Unit – I

Mendelian Population and scope of population genetics. Gene and genotype frequencies, mating patterns, Hardy-Weinberg principle, heterozygotes, extention of H-W principle to multiple alleles, sex-linked alleles. Non-random matings, inbreeding and assortative matings, inbreeding coefficient. Factors that change allelic frequencies.

Unit – II

Genetic polymorphism, transient and stable and factors responsible for stable polymorphism. DNA markers and population differences. Application of population genetics. Role of population genetics in genetic counseling. Genetics of origin and evolution of human races. Genetic Demography, age and gender specific death and birth rates, intrinsic rate of natural increase. Index of opportunity for natural selection.

Unit – III

Importance of population studies, sampling techniques, classification of data and tabulation. Measures of central tendency- mean, median and mode. Measures of dispersion - variance and standard deviation.

Unit – IV

Probability – laws of probability for independent events – permutations and combinations – conditional probability. Binomial distribution. Tests of significance – chi square test and 't' test.

BOOKS SUGGESTED:

1. The Genetics of Human Populations by LL Cavalli-Sforza and WF Bodmer Freeman and Company, 1971.
2. Population Genetics Theory by James F. Crow and W. Kimura Harper and Row, 1970.
3. Introduction to Biostatistics by P.S.S. Sundara Rao and J. Rich New Delhi
4. Introduction to Biostatistics by Robert R. Sokel and James F. Kohlf

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PAPER-1.3: HUMAN CYTOGENETICS AND REPRODUCTIVE GENETICS

Unit – I

History and development of Human cytogenetics–;Morphological variability of the human chromosome and karyotyping; Banded chromosomes and individual characterization of the human chromosomes; Standardization in Human Cytogenetics;

Unit – II

The origin and transmission of chromosomal abnormalities; numerical chromosomal abnormalities(five classical syndromes);structural chromosomal abnormalities (5p-); Fragile X – chromosome, heterochromatin and Lyon's hypothesis; somatic cell hybridization and use of somatic cell hybrids in gene mapping; Fluorescence in situ hybridization

Unit – III

Male and female reproductive systems; gonads and differentiation of sexual characters; hormonal regulation of sexual differentiation; Reproductive disorders- pseudohermaphroditism; true hermaphroditism; gonadal disgenesis, testicular feminization;

Unit – IV

Infertility - genetic basis of male infertility, genetic basis of female infertility; recurrent pregnancy loss;

SUGGESTED BOOKS

1. Human Cytogenetics (vol. I & II) – J.L. Hamerton
2. Human chromosomes : E.H. FORD
3. Human Genetics – F. Vogel and A.G. Motulsky.
4. genetics and Medicine – M.W Thompson, R.R. Meinees and H.F Willard
5. Basic human genetics – E.J. Mange and A.P. Mange.
6. Medical Genetics – Jorde et al
7. New chromosomal syndromes : J.J. Yunis
8. Comprehensive clinical endocrinology : Besser and Thorner
9. Principles and practice of Medical genetics : Rimoin etal

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PAPER- 1.4: MOLECULAR GENETICS AND MOLECULAR MECHANISMS OF HUMAN PATHOGENS

Unit-I

Structure and types of nucleic acids(DNA and RNA); mitochondrial genome; transposons; pseudogenes; mechanisms of replication in prokaryotes and eukaryotes; regulation of DNA synthesis; DNA repair systems.

Unit-II

Transcription mechanisms in prokaryotes and eukaryotes; post transcriptional processing-5' capping, 3' polyadenylation, splicing; translation mechanisms in prokaryotes and eukaryotes; post translation modifications; regulation of transcription- operon and regulon ; regulation of translation.

Unit-III

Host-pathogen interaction, evolution of pathogenecity and regulation of virulence; Mechanism of drug resistance in pathogens; Molecular mechanisms for origin of new pathogens

Unit-IV

Molecular biology of pathogens: HIV, Hepatitis viruses, Mycobacterium tuberculosis, Vibrio cholerae, Plasmodium, Leishmania, Trypanosoma, Entamoeba

SUGGESTED BOOKS

1. Molecular Biology: Weaver, RF
2. Genes and Genomes: Singer,M and Berg, P
3. Molecular Cell Biology: Lodish et al.
4. Molecular Biology: Bruce Alberts et al.
5. Microbiology: Principles & Explorations : Black
6. Introductory Microbiology : Heritage et al
7. Biology of Microorganisms : Madigen et al
8. Microbiology-A Human Perspective : Nesler et al

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PAPER- 1.5: PRACTICAL – I

Part – A

1. **Genetics of Blood Groups**
   a) ABO –typing
   b) Rh (D) typing
   c) ABH Secretor status

2. **Genetic Traits**
   a) Colour Blindness
   b) Phenyl Thio Carbamide (PTC)

3. **Dermatoglyphics**
   a) Finger ball patterns
   b) Palmar patterns

Part – B

(Assignment)

1. Measures of Central Tendency
2. Measures of Dispersion
3. Correlation
4. Probability
5. Binomial Distribution
6. Tests of Significance
7. Gene frequencies-Hardy Weinberg Equilibrium
8. Genetic Polymorphism
9. Mutational Rates

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PAPER- 1.6: PRACTICAL – II

Part – A

1. Chromosome Nomenclature
2. Karyotyping
3. Metaphase drawing
4. Drumstick and Barr body identification
5. Demonstration of Human blood lymphocyte culture
   a. Washing and sterilization of glassware
   b. Medium preparation
   c. Setting up of lymphocyte culture
   d. Harvesting and slide preparations
   e. Identification of individual chromosomes
   f. Preparation of G-banded chromosomes

Part – B

1. Isolation of DNA from peripheral Blood
2. Isolation of DNA from Tissue
3. Isolation of RNA from Lymphocytes
4. Quantification of Genomic DNA
5. Quality check for Genomic DNA

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SEMESTER – II

PAPER – 2.1: DEVELOPMENTAL AND BEHAVIORAL GENETICS

Unit - I

Genetics of embryonic development; basic concepts in development , animal models in human development. Major processes in embryonic development- pattern formation, axis specification and organogenesis.

Unit – II

Genomic imprinting; transgenic animals; later phases of embryonic development; phenocopies and malformations; the development of structure and birth defects in humans; sex differentiation and its errors

Unit – III

Scope and importance of behavioral genetics; animal models; behavioral genetic experiments in the mouse

Unit – IV

Genetics of human behaviour; chromosomes and mental retardation; chromosomes and antisocial behaviour and aggressiveness; genetics and alcoholism; genetics of psychiatric disorders- Schizophrenia; ethnic differences in IQ tests; twins in behaviour genetic research; behavioural genetics and affective disorders

SUGGESTED BOOKS

1. Human Genetics: Vogel and Motulsky
2. Basic Human Genetics : Mange and Mange
3. Genetic studies in Affective Disorders: DP Papolos and HM Lachman
4. Medical Genetics : Jorde et al

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PAPER-2.2: HUMAN BIOCEMICAL AND IMMUNOGENETICS

Unit – I

The concept of Biochemical polymorphism, enzyme, protein and DNA polymorphisms – ACP, ESD, HP and GC systems; Hemoglobinopathies; Pharmacogenetics – Glucose- 6-Phosphate dehydrogenase deficiency ; Ecogenetics – Alpha -1- Antitrypsin.

Unit – II

Inborn errors of metabolism–Disorders of carbohydrate metabolism–Galactosaemia; Disorders of amino acid metabolism – Alkaptonuria, Phenylketonuria, Albinism and Homocystinuria; Disorders of Lysosomal enzymes–Tay- Sachsdisease and Mucopolysaccharidoses; Disorders of Lipoprotein and lipid metabolism – Hyper Lipoproteinemia; Disorders of Purine metabolism-Lesch Nyhan syndrome; Disorders of Pyrmidine metabolism – Orotic Aciduria

Unit – III:

The immune response – Basic concepts; the innate immune system–Phagocytes, the complement system, natural killer cells; The adaptive immune system – Cellular immune system, humoral immune system; Genetic basis of structure and diversity; Immunoglobulin molecules and The genetic basis of antibody diversity.

Unit – IV

The Major Histocompatability complex-Class I (HLA-A,B,C,E,F & G),Class II (HLA-DP,DR,DQ) and Class III (Complement genes); Immunodeficiency diseases- Agamma- globulinemia, Severe combined immuno-deficiency, Ataxia telangiectasia, Wiskott- Aldrich syndrome.
Autoimmunity – Altering immune function (vaccines and transplants); immuno-therapy (monoclonal antibodies and cytokines), immunity breakdown (AIDS)

SUGGESTED READINGS:

1. Principles of Human Biochemical Genetics by H. Harris
2. Human Genetics by A.G. Motulsky and F. Vogel
3. The metabolic basis of inherited diseases by Scriver et al.
4. Medical Genetics by Lynn B. Jorde et al
5. Basic immunogenetics - Fudenberg et al

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PAPER - 2.3: MEDICAL AND CANCER GENETICS

Unit – I

Scope ofMedical Genetics. Skin- Ichthyosis, psoriasis, multiple neurofibromatosis, porphyrias, blooms syndrome. The skeletalsystem –spina bifida and anencephaly -Ankylosing spondylitis, Rheumatoid arthritis, Osteogenesis imperfecta. Muscle – muscular dystrophies, Myotonia.

Unit – II

Eye – Glaucoma, cataract. Jaws – Hare lip and palate. Ears -Deafness. Alimentary system - Hypertrophic pyloric stenosis, cirrhosis of liver. Respiratory system – Cystic fibrosis, Cardio vascular system – congenital heart disease, coronary heart diseases and Hypertension, Central nervous system – The ataxias and familial spastic paraplegia- Kidney and urinogenital tract – Cystinosis, polycystic kidney .Endocrine system – Cretinism, goiter, diabetes.

Unit – III

Cancer Genetics: What is cancer? Characteristics of cancer cells, Chromosomes in neoplasias, cancer as a genetic disorder, cancer in families, loss of cell cycle control; Inherited versus sporadic cancers.

Unit – IV

Molecular changes in proto-oncogenes, tumor suppressor genes-Knudson's Hypothesis, Retinoblastoma, Lung cancer, Colon cancer, Brain cancer, Breast cancer, Prostrate cancer, cervical and esophageal cancers; Cancer and environment: physical, chemical and biological carcinogens;. Cancer prevention, diagnosis and treatment

SUGGESTED BOOKS

1. Medical Genetics – Jorde et al
2. genetics and Medicine – M.W Thompson et al
3. Clinical Genetics – A. Sorsby
4. Genetic Disorders of Man – R. M. Goodman
5. Emery's Elements of Medical Genetics – R. F. Mueller and I.D Yound
6. Human Genetics – F. Vogel and A.G. Motulsky.

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PAPER- 2.4: MOLECULAR GENETICS AND HUMAN GENOME PROJECT

Unit – I

Cloning of disease genes: functional cloning-Hemophilia, positional cloning- Cysticfibrosis, Duchenmuscular dystrophy, Huntington disease; molecular pathology- gene deletions, codon deletions, duplications and insertions; mutations- point mutations, RNA splice mutations, transcriptional mutations; Mitochondrial mutations and diseases

Unit – II

Molecular diagnosis- infectious diseases; genetic diseases and direct testing-Sickle cell anemia, Beta thalassemia, Cystic fibrosis, Duchenmuscular dystrophy, Huntington disease; gene tracking- Cystic fibrosis and Huntington disease

Unit – III

The genome project- history, organization and goals of human genome project; mapping strategies, current status of various maps; human genome diversity; Organization of human genome- Mitochondrial genome, gross base composition of nuclear genome,gene density.

Unit – IV

Gene families-Multigene families, Classical gene families, families with large conserved domains, families with small conserved domains,Gene superfamilies,Gene families in clusters,origin of gene families

SUGGESTED BOOKS

1. Human Molecular Genetics: Strachan and Read
2. Principles of Genetics : Snustad and Simmons
3. Human genetics-The Molecular Revolution: Mc Kokey
4. Molecular Diagnosis: Jeffery et al.
5. The Human Genome : Hawley and Mori
6. From Genes to Genomes :Dale & Schartz
7. Genomes :Brown

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PAPER- 2.5: PRACTICAL – III

Part – A
(Assignment)

1. Structure of Egg
2. Structure of Sperm
3. Cell cycle and Mitosis
4. Meiosis
5. Spermatogenesis
6. Oogenesis
7. Cleavage and Gastrulation in Mammals
8. Non-disjunction
9. Human Embryo Development
10. Transgenic Animals
11. Klinefelters syndrome
12. Downs syndrome
13. Fragile X syndrome

Part – B

1. Sickling Test
2. Separation of abnormal heamoglobins
3. Estimation of Hb
4. Estimation of Hb A2
5. Red Cell Enzymes – ACP, ESD
6. Plasma proteins – HP, CP
7. ELISA Test
8. Radial Immunodiffusion (RID)
9. Quantitative Precipitin Assay

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PAPER- 2.6: PRACTICAL – IV

Part – A

(Assignment)

1. Introduction to medical genetics
2. Skin disorders- Ichthyosis, Multipile neurofibromatosis, Blooms syndrome
3. Skeletal disorders- Rheumatoid Arthritis, Osteogenesis imperfeta
4. Muscle disorders- Muscular distrophys
5. Eye disorders- Glacoma, Retinoblastoma
6. Cardiovascular disorders- Congenital heart disease, coronary heart disease, hypertension
7. Endocrinal disorders- Critinism, Goiter

Part – B

1. Primer designing
2. Insertion deletion polymorphism
3. DNA Finger printing – RFLPs and VNTRs
4. Amplification and purification of DNA fragments
5. ARMS-PCR
6. Multiplex PCR
7. Nested PCR
8. DNA sequencing methods

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Paper: 2.7: FUNDAMENTALS OF HUMAN GENETICS

(Optional Paper /Non Core Subject)

UNIT- I:

Mendals laws of inheritance; Simple single factor inheritance (autosomal dominant, autosomal recessive, x-linked dominant, x-linked recessive and Y-linked characters); Multifactorial inheritance; Sex influenced and sex limited characters ; Polygenic inheritance.

UNIT- II:

Multiple alleles; Lethal and sub lethal genes; Penetrance and expressivity; Mutations; Linkage and Crossing over; Heredity and environment (Twin study).

UNIT- III:

Population studies and their importance; Family studies and pedigree analysis; Estimation of gene, genotype and phenotype frequencies; Randam mating and its consequences; Hardy- Weinberg law; Genetic polymorphism (balanced and transient); Inbreeding – types of consanguineous marriages.

UNIT- IV:

Scope of genetic councelling- Methods of genetic councelling
Scope of genetic screening - Prenatal and postnatal genetic screening methods.

Suggested Books:

1, Principles of Human Genetics – Curt Stern
2. Human Genetics – F. Vogel and A. G. Motulsky
3. Basic Human Genetics – Mange and Mange
4. Genetic Counselling - W. Fuhramann and F. Vogel
5. Genetics of Human Populations – Cavallis S. forza and Bodmer

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PAPER-3.1: CLINICAL GENETICS AND GENETIC TOXICOLOGY

Unit – I

Scope of clinical genetics; Monogenic diseases- Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome; Polygenic diseases- Hyperlipidemia, Diabetes mellitus, Atherosclerosis; Inborn errors of metabolism and their genetic bases- Phenylketonuria, Maple syrup urine syndrome, Mucopolysaccharidosis, Galactosemia.

Unit – II

Neurogenetic disorders- Major regions of human brain and nerve conduction, Charcot-Marie-Tooth syndrome, spinal muscular atrophy; Syndromes due to triplet nucleotide expansion -Alzheimer's disease; Genetic disorders of Haemopoitic systems- Sickle cell anemia, Thalassemias, Hemophilias.

Unit – III

Origin of genetic toxicology; historical prospective of genetic toxicology; fundamentals of genetic toxicity; mechanism of induction of chromosomal alterations and sister chromatid exchanges; mutagens-chemical, physical, biological, environmental and food; antimutagens.

Unit – IV

Mechanisms of gene mutations; germinal mutations and human genetic diseases; mutations and cancers; genetic toxicology and congenital malformations; consequences of genotoxic effects in humans.

Recommended Books

S.No	Author(s)	Title	Pub	Year
1	Cox & Sinclair	Molecular Biology in Medicine	Blackwell	1997
2	DeGrouchy & Turleau	Clinical Atlas on Human Chromosomes	Wiley	1984
3	Jankowski & Polak	Clinical Gene Analysis and Manipulation	Cambridge	1996
4	David Brusick	Principles of Genetic Toxicology		
5	Pasternak	An Introduction to Molecular Human Genetics	Fritzgarald	2000
6	Albert P, Li, RH,Heflich	Genetic Toxicology		
7	Rimoin *et al*	Principles & Practice of Medical Genetics, vol. I-III	Churchill	2002
8	Robinson & Linden	Clinical Genetics Handbook	Blackwell	1994
9	Strachan & Read	Human Molecular Genetics	Wiley	1999
10	Wilson	Clinical Genetics: A Short Course	Wiley-Liss	2000

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PAPER- 3.2: DNA TECHNOLOGY AND GENETIC ENGINEERING

Unit – I

Enzymes used in DNA technology, Restriction and modification enzymes, Other nucleases, Polymerases, Ligase, kinases and phosphatases. Nucleic acids, Isolation and purification of DNA (genomic and plasmid) and RNA, Gel electrophoresis of nucleic acids (denaturing and native), Pulse-field gel electrophoresis of DNA.

Unit – II

Cloning vectors, Plasmids, Phages, Cosmids, Artificial chromosomes, Shuttle vectors, Expression vectorsCloning techniques, Construction of genomic and cDNA libraries, Positional cloning: RFLP mapping, chromosome walking and jumping, Screening and characterization of clones, Preparation of probes, Restriction mapping, Principles of hybridizations and hybridization based techniques (colony, plaque, Southern, Northern and in situ hybridizations) .

Unit – III

DNA sequencing, Oligonucleotide synthesis, Polymerase Chain Reaction and its applications, Microarray technology, ELISA, western and south-western blotting, Promoter characterization: promoter analysis through reporter genes, electrophoretic mobility shift assay, DNA foot-printing, DNA fingerprinting, Mutagenesis, Site directed mutagenesis, Transposon mutagenesis, Construction of knock-out mutants.

Unit – IV

Gene transfer techniques, Electroporation and microinjection, Transfection of cells: Principles and methods,Germ line transformation in *Drosophila* and transgenic mice: Strategies and methods. Applications of Recombinant DNA Technology, Crop and live-stock improvement, Molecular genetic analysis of human diseases, DNA drugs and vaccines, Biosafety and ethical considerations

Recommended Books				
1	Ausubel *et al*	Short Protocols in Molecular Biology	Wiley	2002
2	Brown	Essential Molecular Biology vol. I	AP	2000
3	Brown	Essential Molecular Biology vol. II	AP	2000
4	Brown	Gene Cloning - An Introduction	Stanley Thornas	1995
5	Glick & Pasternak	Molecular Biotechnology	ASM Press	1998
6	Kracher	Molecular Biology - A Practical Approach		
7	Primrose	Molecular Biotechnology	Panima	2001
8	Reischel	Molecular Diagnosis of Infectious Diseases	Humana	1998
9	Robertson *et al*	Manipulation & Expression of Recombinant DNA	AP	1997
10	Twyman	Advanced Molecular Biology	Viva	1999
11	Watson *et al*	Recombinant DNA	Freeman	1992
12	Sandhya Mitra	Genetic Engineering Principles and Practice	Macmillan	1996

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PAPER- 3.3: GENETIC SCREENING, COUNSELING AND GENE THERAPY

Unit – I

Scope of genetic screening- Prenatal and Post natal screening. Population screening for genetic diseases, family screening.

Unit – II

Prenatal screening methods- Amniocentesis- Chronic Villous sampling, Ultrasonography, fetoscopy, maternal blood sampling.

Post-natal screening- chromosomal abnormalities, cytogenetic disorders and molecular methods.

Unit – III

Scope of genetic counseling- methods of genetic counseling, educating the counselee, presenting the risks and options and guiding. Social, ethical and legal issues. Patterns of inheritance and risk assessment, chromosomal disorders, autosomal dominant and recessive disorders, X-linked disorders, multifactorial-polygenic disorders. Reproductive failures, consanguinity.

Unit – IV

Gene Therapy- classification of gene therapy- class I, II, and III. Types of gene therapy- germ line gene therapy and somatic gene therapy.

SUGGESTED READINGS:

1. Human Molecular Genetics by T. Strachan and AP Read
2. Human Genetics by F. Vogel and A.G. Motulsky
3. Genetic Engineering by Sandhya A. Mitra
4. Medical Genetics by Jordee et al
5. Genetic Counseling by W. Fuhramann and F. Vogel

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PAPER- 3.4: GENOMICS AND PROTEOMICS

Unit – I

Introduction to genomics, genetic mapping of human chromosomes, mapping of genetic disease locus to chromosome location, multilocus mapping of human chromosome, physical mapping of human genome, cloning human disease genes, human genome project.

Unit – II

DNA sequencing, bio chips, DNA micro arrays, gene annotation, gene structure predictions, gene ontology consortium recommendations, structural and functional genomics.

Unit – III

Protein structure and its determination, structural hierarchy, domains, folds, motifs. Secondary structure prediction methods, fold recognition and abinitio structure prediction, homology- comparative modeling of proteins.

Unit – IV

Protein chip arrays, functional proteomics, docking, rational drug design.

Suggested Readings:

1	MalcolmCampbell and Laurie J.Heyer	Genomics,proteomics and Bioinformatics	Benjamin Cummings	2002
2	Lynn B. Jorde et al	Encyclopedia of genetics, Genomics, Proteomics and Bioinformatics	Wiley	2006

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PAPER- 3.5: PRACTICAL – V

Part – A
(Assignment)

1. Introduction to clinical genetics
2. Monogenic diseases- Cystic fibrosis, Tay-Sachs syndrome
3. Polygenic diseases- Hyperlipidemia, Diabetes mellitus
4. Inborn errors of metabolism and their genetic basis- Phenylketonuria, Galactosemia.
5. Neurogenetic disorders- Major regions of human brain and nerve conduction- Alzheimer's disease, Charcot-Marie-Tooth syndrome
6. Genetic disorders of Haemopoitic systems- Sickle cell anemia, Thalassemias,

Part – B

1. Preparation of Plasmids
2. Restriction digestion
3. Ligation of DNA fragments
4. Extraction of DNA fragments from agarose gel
5. Restriction Mapping
6. Transformation
7. Blotting Techniques:

   a) Southern
   b) Western
   c) Northern

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PAPER 3.6: PRACTICAL – VI

Part – A

(Assignment)

1. Genetic Counselling methods
2. Prenatal screening methods
3. Postnatal screening methods
4. Patterns of inheritance
5. Counselling for single gene disorders (Case studies)
6. Counselling for multifactorial disorders (Case studies)

Part – B

[Genomics and Proteomics]

1. Introduction of Genomics.
2. Genetic mapping techniques.
3. Literature search from NCBI database.
4. Genome map viewer from NCBI.
5. Detecting open reading frame.
6. Working Human Genome from NCBI.
7. Ensemble – A brief visit.
8. SDS PAGE
9. Protein folding.
10. Protein estimation.
11. Protein Agarose gel electrophoresis.
12. Analysis of protein structure.
13. Study of physical properties of protein.

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Paper 3.7: ADVANCED HUMAN GENETICS

(Optional Paper /Non Core Subject)

UNIT- I

History and development of human cytogenetics; Standardization in human cytogenetics- different conferences; Morphological variability of human chromosomes; Karyotyping.

UNIT- II

Origin and transmission of numerical chromosomal abnormalities (classical syndromes); Structural chromosomal abnormalities (4p- and 5p-); Human leucocyte blood culture; Human chromosome banding techniques.

UNIT- III :

The concept of bio chemical polymorphism- enzyme and protein polymorphisms- ACP, ESD, HP and GC; Pharmacogenetics- G6PD; Hemoglobinopathies; In born errors of metabolism- Carbohydrate and aminoacid metabolisms.

UNIT- IV :

The immune response – Basic concept; Innate immune system (phagocytes, complement system, natural killer cells); Adaptive immune system (cellular and humoral); Genetic basis of structure and diversity; Antibody diversity ; Major histocompatability complex (class I, II and III)..

Suggested Books:

1. Human Cytogentics - J. L. Hamerton
2. New Chromosomal Syndromes – J.J. Yunis
3. Molecular Structure of Human Chromosome – J.J. Yunis
4. Principles of Human Biochemical Genetics- H. Harris
5. Basic Immunogenetics – Fuden Berg et al.,

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Model Question Paper

M. Sc Degree Examination
Human Genetics
First Semester

Paper 1.1 – BASIC HUMAN GENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. What are Mendel Laws and explain the laws with suitable examples?

Or

2. Give an account on Multiple alleles?

3. Write an essay on the effect of environment on phenotype development?

Or

4. Explain polygenic inheritance with suitable examples?.

5. Discuss the sex-linked and sex-influenced types of inheritance with suitable examples ?.

Or

6. Explain the autosomal recessive inheritance with suitable examples?.

7. Discuss the concept of linkage and. crossing over ?

Or

8. Write an essay on heredity and environment by using twin studies?.

9. Write short notes on any FOUR of the following.

   a) Mutations
   b) Pleiotropy
   c) Y-linked inheritance
   d) Pedigree
   e) Phenocopy
   f) Physical mapping
   g) Mitochondrial inheritance
   h) Penetrance

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Model Question Paper

M. Sc Degree Examination
Human Genetics
First Semester

Paper 1.2 – POPULATION GENETICS AND BIOSTATISTICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks: 80

Answer ALL questions
All questions carry equal marks

1. Define Hardy-Weinberg Law and describe the factors which disturb the equilibrium ?

Or

2. What is inbreeding and calculate Inbreeding coefficient for a) uncle niece marriage, b) first cousin marriage, c) second cousin marriage

3. Write an essay on genetic polymorphism? .

Or

4. Write an Index of opportunity for natural selection ?.

5. Give an account on various sampling techniques ?.

Or

6. Find out the mean and standard deviation for the following data?

Weight (lbs)	-	No. of persons
100-110	-	04
110-120	-	20
130-140	-	32
140-150	-	33
150-160	-	17
160-170	-	08
170-180	-	02

7. Explain the laws of probability ? Discuss its significance special reference to Human population genetics ?

Or

8. Explain the binomial distribution in detail ?

9. Write short notes on any FOUR of the following.
   a. DNA markers
   b. Genetic demography
   c. variance
   d. Chi-square test
   e. Sex linked alleles
   f. Mating patterns
   g. Birth rates
   h. Genetic drift

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Model Question Paper

M. Sc Degree Examination
Human Genetics
First Semester

Paper 1.3 – HUMAN CYTOGENETICS AND REPRODUCTIVE GENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Give an account on History and development of human Cytogenetics?

Or

2. Describe the morphological variability of human chromosomes?

3. Discuss heterochromatin and Lyon hypothesis?

Or

4. Write about the origin and transmission of chromosomal abnormalities?

5. Describe the hormonal regulation of sexual differential in females ?

Or

6. Describe various reproductive disorders in males?

7. Discuss the genetic basis of female infertility ?

Or

8. Write an essay recurrent pregnancy loss ?

9. Write short notes on any FOUR of the following.

   a. Banding techniques
   b. Chicago conference
   c. FISH
   d. Fragile X syndrome
   e. Gonadal dysgensis
   f. Female reproductive system
   g. Down syndrome
   h. Somatic cell hybridization

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Model Question Paper

M. Sc Degree Examination
Human Genetics
First Semester

Paper 1.4 – MOLECULAR GENETICS AND MOLECUAL MECHANISM OF HUMAN PATHOGENS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Give a detailed account on mechanism of DNA replication in eukaryotes ?

Or

2. Describe the DNA repair system ?

3. Explain the translational mechanism in prokaryotes and eukaryotes ?

Or

4. Discuss about the regulation of transcription?.

5. Discuss the mechanism of drug resistance in pathogens ?

Or

6. Explain the post pathogen interaction ?

7. Discuss the molecular structure of HIV ?

Or

8. Explain the molecular biology of hepatitis virus ?.

9. Write short notes on any FOUR of the following.

   a. Transposons
   b. Psudogenes
   c. Post transcriptional process
   d. RNA splicing
   e. Plasmodium
   f. Operon
   g. Replication
   h. Okazaki fragments

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Second Semester

Paper 2.1 – DEVELOPMENTAL AND BEHAVIOURAL GENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Discuss the basic concepts in Developmental genetics?

OR

2. Write an essay on axis specification and organogenesis?

3. Write an essay on Genomic imprinting? Illustrate your answer with suitable examples?

OR

4. Highlight on sex differentiation and its errors with suitable examples?

5. Give an account of the significance of behavioural genetic experiments in the mouse?

OR

6. Write an essay on Genetic aspects of alcoholism?

7. Discuss the twin studies in behavior genetic research?

OR

8. Is mental retardation genetically controlled? Explain?

9. Write short notes on any four of the following.
   a) Pattern formation
   b) Gastrulation
   c) Transgenic animals
   d) Birth defects
   e) Schizophrenia
   f) Klinifelters syndrome
   g) Manic depressive illness
   h) Chromosomes and antisocial behaviour

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Second Semester

Paper 2.2 – HUMAN BIOCHEMICAL AND IMMUNOGENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks: 80

Answer ALL questions
All questions carry equal marks

1. Explain the concept of biochemical polymorphism with suitable examples?

OR

2. Describe the Genetics and clinical symptoms of hemoglobinopathies?

3. Discuss the disorders of aminoacid metabolism?

OR

4. Give an account on Hyperlipoprotenemia?

5. Write about the types of Immune systems? Give a detailed account of adaptive immune system?

OR

6. Give an account on different types of immunoglobins?

7. Write notes on major histo compatibility complex in man?

OR

8. Discuss the immunodeficiency diseases with suitable examples?

9. Write short notes on any FOUR of the following.

   a) Acid Phosphatase
   b) Alpha-1-anti trypsin
   c) Galactosaemia
   d) DNA Polymorphism
   e) Innate Immunity
   f) T-lympocytes
   g) Phagocytosis
   h) AIDS

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Second Semester

Paper 2.3 – MEDICAL AND CANCER GENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Describe the following genetic disorders?
   a) Marfan's Syndrome
   b) Anencephaly and Spina bifida
   c) Rheumatoid Arthritis

OR

2. Write an essay on scope of Medical Genetics ?

3. Give brief account on
   a) Retinoblastoma
   b) Cystic Fibrosis
   c) Coronary Heart Disease (CHD)

OR

4. Explain the major metabolic changes associated with diabetes?

5. Write about chromosomal aberrations in neoplasias?

OR

6. Write an essay on genetic basis on Cancer?

7. Give an account on molecular changes in proto-oncogenes?

8. Write an essay on the breast cancer?

9. Write short notes on any **FOUR** of the following.
   a) Psoriais
   b) Myotonia
   c) Pyloric sterosis
   d) Polycystic kidney disease
   e) Characteristics of cancer cells
   f) Loss of cell cycle control
   g) Cancer and environment
   h) Knudson's Hypothesis

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Second Semester

Paper 2.4 – MOLECULAR GENETICS AND HUMAN GENOME PROJECT
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Give an account of the cloning of human diseases genes and mention its significance?

OR

2. Describe the Mitochondrial diseases?

3. Explain the molecular diagnosis of Duchene Muscular Dystrophy (DMD)?

OR

4. Discuss the Gene tracking in cystic fibrosis and Huntington's disease?

5. Discuss the Genetic mapping strategies?

OR

6. Describe the physical mapping of human genome and state its importance?

7. Describe the origin and clusters in gene families?

OR

8. Explain the concept of Gene families?

9. Write short notes on any FOUR of the following.

a) Beta thalassemia
b) Multi gene families
c) Gene Density
d) Mitochondrial Genome
e) Cystic fibrosis
f) Positional cloning
g) Transcriptional mutations
h) FISH

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Non-Core / Optional Paper

Model Question Paper

M. Sc Degree Examination
Human Genetics
Second Semester

Paper 205 – FUNDAMENTALS OF HUMAN GENETICS
(Effective from the Admitted Batch of 2008-2009, Modified in 2014)

Time: 3 hours

Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Describe Mendel's laws of inheritance with suitable examples?

OR

2. Discuss the multifactorial and polygenic inheritance?

3. Explain the penetrance and expressivity with suitable examples?

OR

4. How do you evaluate the relative roles of heredity and environment through twin studies?

5. Discuss the importance of population studies and family studies?

OR

6. Describe the genetic polymorphism with suitable examples?

7. Explain the various types of Genetic counseling?

OR

8. Describe the prenatal diagnosis methods and their importance in medicine?

9. Write short notes on any FOUR of the following.
   a) Autosomal recessive
   b) Sex limited characters
   c) Multiple allelism
   d) Crossing over
   e) Fetoscopy
   f) Inbreeding
   g) Amniocentesis
   h) Natural selection

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Third Semester

Paper 3.1 – CLINICAL GENETICS AND GENETIC TOXICOLOGY
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours

Max. Marks: 80

Answer ALL questions
All questions carry equal marks

1. Describe the modes of inheritance with suitable examples

Or

2. Discuss about polygenic diseases with examples.

3. Give an account on Neurogenetic disorders.

Or

4 Write about Genetic disorders of Haemopoitic systems.

5 Discuss about genetic toxicology and its applications.

Or

6. Explain mechanisms of induction of chromosomal alterations.

7. Give an account on mechanisms of gene mutations.

Or

8. Write about consequences of genotoxic effects in humans.

9. write short note on any FOUR of the following:

   a. Diabetes mellitus
   b. Sickle cell disease
   c. In born errors of metabolism
   d. Antimutagens
   e. Cancers
   f. Germinalmutations
   g. Galactosemia
   h. Sister chromatid exchange

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Model Question Paper

M. Sc Degree Examination
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Third Semester

Paper 3.2 – DNA TECHNOLOGY AND GENETIC ENGINEERING
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours

Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. What are the enzymes used in DNA technology explain.

Or

2. Explain in brief genomic and plasmid DNA isolation and purification.

3. Give an account of cDNA synthesis and cDNA libraries.

Or

4 Explain the hybridization based techniques in brief.

5 Give an account of the PCR in detailed and its applications. Describe the PCR modifications.

Or

6. Discuss about the direct and insertional mutagenesis.

7. Describe the hazards of genetic engineering and its impact on society.

Or

8. Discuss about the gene transfer techniques.

9. write short note on any FOUR of the following:

   a. Polmerases.
   b. Pulse-field gel electrophoresis
   c. Southern blotting
   d. Chromosome walking
   e. Vaccines
   f. Electroporation
   g. Probes
   h. Transgenic animals

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Third Semester

Paper 3.3 – GENETIC SCREENING, COUNSELING AND GENE THERAPY
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Write an essay on postnatal screening and explain different type of postnatal Screening methods?

Or

2. What is prenatal diagnosis? Explain various methods used in prenatal diagnosis?

3. Write an essay on Genetic Counseling explain various social ethical and legal issues involved?

Or

4. Write an essay on autosomal dominant and autosomal recessive Disorders?

5. What are various prenatal Screening methods and explain the amniocentesis method with suitable diagram?

Or

6. Write an essay on Chronic Villus Biopsy (CVB)? How it is useful in medical diagnosis?

7. What is the Gene therapy? Explain the Classification of Gene therapies?

Or

8. Write an essay on Gene therapy and Types of Gene Therapy?

9. Answer any FOUR of the following:

a) Multifactorial Inheritance

b) Pedigree analysis

c) DNA finger printing

d) Educating the Counselee

e) Ultrasonography

f) Zygote gene therapy

g) Numerical Chromosomal abnormalities.

h) Fetoscopy

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Model Question Paper

M. Sc Degree Examination
Human Genetics
Third Semester

Paper 3.4 – GENOMICS AND PROTEOMICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours

Max. Marks : 80

Answer ALL questions
All questions carry equal marks

I. Give an account of salient features of Human genome project.

OR

2. Describe the cloning of human disease genes and mention its significance

3. .Enumerate and explain the recommendations of gene ontology consortium.

OR

4. Write an illustrated account of DNA micro array technology.

5. Describe the protein structure and its determination

OR

6. What is proteome? Write an account on proteomics.

7. Write an account of protein chip arrays and docking

OR

8. What is rational drug design? How is it useful in discovering new medicines?

9. Write short notes on any FOUR of the following

a) Biochips

b) Fold recognition

c) Relationship between gene and protein

d) FISH

e) DNA marker

f) Open reading frame (ORF)

g) Motif

h) Drug design.

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Non-Core / Optional Paper

Model Question Paper

M. Sc Degree Examination
Human Genetics
Third Semester

Paper 305 – ADVANCED HUMAN GENETICS
(Effective from the Admitted Batch of 2011-2012, Modified in 2014)

Time: 3 hours Max. Marks : 80

Answer ALL questions
All questions carry equal marks

1. Describe the growth of human cytogenetics?

OR

2. Explain the structure and morphological variability of human chromosomes?

3. Write an essay on the different banding techniques used to identify chromosomes?

OR

4. Describe any two numerical chromosomal abnormalities and explain how they are transmitted to the next generation.

5. What is polymorphism? Give an account of red cell enzyme polymorphisms in Man?

OR

6. Describe the clinical symptoms and genetics of alkaptonuria and galactosemia?

7. Discuss how antibody diversity is achieved in man?

OR

8. Along with the concept of immune response, write down the different immune systems found in human body?

9. Write short notes on any FOUR of the following:

   a. Major Histocompatibility Complex
   b. Pharmacogenetics
   c. Cat-cry syndrome
   d. Karyotyping
   e. Denver Conference
   f. Human leucocyte blood culture
   g. Plasma protein polymorphisms
   h. Immunoglobulins

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(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 fibrila – 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.

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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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PAPER – II – SPECIAL PAPER INCLUDING RESEARCH METHODOLOGY

(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. Inbron errors of metabolism : Disorder of carbohydrate metabolism; disorders of aminoacid metabolism; hypo several enzyme deficiencies, miscellaneous disorders.
6. Immunogenetics : the immune respects baric concepts ; the innate immune system : the adaptive immune system. Immunoglobulins ; (cellular and adoptive systems). The major histocompatability 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.

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7. Pharmacogenetics : G6PD; Pseudocholinesterase; Issomiazid inactivation.

8. Ecogenetics : carcinogenesis ; a-1-antitripsin deficiency – foods.

9. Gene mutations and inherited diseases :
   The molecular pathology of inherted disease : dominance and recessivity ; hetro genity of inherited disease: heredity and environment.

10. Heterozygote detection.
    Treatment of inherited metabolic disease.

*

11. Blood groping 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, lon – 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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PAPER – II – SPECIAL PAPER INCLUDING RESEARCH METHODOLOGY

(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 infections diseases.

Consanguinity and its genetic consequences

Genetic load

Genetic distance

Gene diversity

Gene flow

Genetic drift.

*

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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PAPER – II – SPECIAL PAPER INCLUDING RESEARCH METHODOLOGY

(D). MOLECULAR GENETICS

(Optional)

Recombinant DNA technology :

Different enzymes and vectors used in genetic engineering.

Constructing gene libraries.

Digesting genomic DNA; Ligting 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 cakaryotic cells applications of Recombinant DNA technology.

Vaccines ; Monoclonal antibodies: Protein engineering : Transgenics.

Structural analysis of gene

Analysis and annotation Databnaks: 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 genetics diseases.

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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

*

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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PAPER – II – SPECIAL PAPER INCLUDING RESEARCH METHODOLOGY

(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.

*

Blood groping 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

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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.

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Model Question Paper

M. Phil.,/ Pre. Ph.D., Examinations
(Effective from the Admitted Batch after June, 2009)

Human Genetics

Paper-I – ADVANCES IN HUMAN GENETICS
(Compulsory)

Marks : 100

Time : 3 Hours

Answer any FIVE Questions
All questions carry equal marks

1. Explain different Chromosome mapping techniques?
2. Illustrate the Mendels mode of inheritance with human examples?
3. Describe the genetic polymorphism with examples?
4. Describe the nucleosome structure?
5. What are prenatal diagnostic methods? Explain any two of them?
6. Describe the genetic counseling methods and genetic counseling for X-linked recessives disorders?
7. Write an essay on genetics of Schizophrenia?
8. Explain the application of Hardy-Weinberg law with suitable examples?
9. Describe the applications of recombinant DNA technology?
10. Answer any FOUR of the following
    a) C-banding
    b) Repetitive DNA
    c) Secondary construction
    d) Mutagenesis
    e) Natural Selection
    f) Protein evolution
    g) First Cousin marriage
    h) Genetic variability of neurotransmitter

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Model Question Paper

M.Phil./ Pre-Ph.D. Examinations
(Effective from the Admitted Batch after June, 2009)

Human Genetics
(Optional Paper)

Paper-II (A): Human Cytogenetics
(Special Paper including Research Methodology)

Time : 3hours

Max. Marks: 100

Answer any FIVE questions
All questions carry equal marks

1. Write about the position of the chromosomes within the cell?
2. Describe new approaches to human gene mapping by somatic cell genetics?
3. Describe different types of banding techniques?
4. Give an account on molecular organization and function of human genome?
5. Write about the chromosome polymorphism and primate evolution?
6. Describe the new chromosome techniques and their medical applications?
7. Write about the genetic counselling?
8. Describe about the in situ hybridization techniques?
9. Describe the role of prenatal diagnostic methods in genetic counseling?
10. Write short note on any FOUR of the following:
    a. Amniocentesis
    b. 4p- syndrome
    c. Physical Mapping
    d. Translocations
    e. Trisomy 9
    f. Q banding
    g. FISH
    h. Chorionic Villi sample

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M.Phil./ Pre-Ph.D. Examinations
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Human Genetics
(Optional Paper)

Paper-II (B) : Human Biochemical Genetics
(Special Paper including Research Methodology)

Time : 3hours

Answer any FIVE questions
All questions carry equal marks

Max. Marks: 100

1. Describe the biochemical polymorphisms with suitable examples?
2. Elaborate on DNA polymorphism and its role in molecular diagnosis?
3. Write an essay on haemoglobinopathies?
4. Discuss about the major histocompatability complex?
5. What is pharmacogenetics? Discuss about G6PD role in pharmacogenetics?
6. Discuss about Alpha 1 antitripsins role in relation to ecogenetics?
7. Describe the gene mutations and inherited diseases?
8. Describe in detailed HPLC technique?
9. Describe the disorders of Amino acid metabolism?
10. Write short notes on any FOUR of the following:
    a) RFLP's
    b) Acid phosphatase
    c) Compliment system
    d) Pseudocholinesterase
    e) Innate system
    f) SNPs
    g) Galactosemia
    h) Thin layer chromatography

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Model Question Paper

M.Phil./ Pre-Ph.D. Examinations
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Human Genetics
(Optional Paper)

Paper-II (C): Human Population Genetics
(Special Paper including Research Methodology)

Max. Marks: 100

Time : 3hours

Answer any FIVE questions
All questions carry equal marks

1. Describe the natural selection with suitable examples?
2. Distinguish the genetic equilibrium from unstable equilibrium?
3. Briefly discuss about frequency dependent and density dependent selection?
4. Write about the consanguinity and its genetic consequences?
5. Describe the genetic load? With suitable examples?
6. What is the genetic distance and explain the influence of genetic distance on natural selection?
7. How the infectuas diseases effects the natural selection?
8. Two random samples of size 900 and 400 have means 21 and 21.3 with SD 3 and 3.1 respectively. These two samples are drawn from form the same population or different population? Explain with statistical evidences?
9. Describe the Gene Diversity and Gene flow with suitable examples?
10. Write any FOUR of the following:
    a) Genetic equilibrium
    b) Gene diversity
    c) LOD Scores
    d) Selection intensity
    e) Density dependent selection
    f) Founder effect
    g) t-test
    h) Sickle cell disease

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Model Question Paper

M. Phil. / Pre. Ph.D., Examinations
(Effective from the Admitted Batch after June, 2009)

Human Genetics
(Optional Paper)

Paper- III (D) - Molecular Genetics
(Special Paper including Research Methodology)

Time : 3 Hours

Marks : 100

Answer any FIVE Questions
All questions carry equal marks

1. Give an account of applications of recombinant DNA technology?
2. Write about different kinds of DNA libraries and ways of screening them?
3. What are the different strategies used to clone human disease genes?
4. Explain how genetic diseases can be diagnosed using molecular methods?
5. Write an essay on genetic mapping of human chromosomes?
6. Discuss the strategies used to deliver a therapeutic gene to cells of target tissue?
7. Write about different expression systems used for the production of proteins?
8. What is the advantage of using mammalian expression system?
9. Discuss about different methods used for identification of mutations?
10. Write short notes on any **Four** of the following:
    a. BACs
    b. Restriction enzymes
    c. Antisense RNA
    d. Physical mapping
    e. Northern blot
    f. Invitro mutagenesis
    g. Automated sequencing
    h. Protein engineering

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Model Question Paper

M.Phil./ Pre-Ph.D. Examinations
(Effective from the Admitted Batch after June, 2009)

Human Genetics
(Optional Paper)

Paper-III (E): Genetic Screening and Counseling
(Special Paper including Research Methodology)

Time : 3hours

Max. Marks: 100

Answer any FIVE questions
All questions carry equal marks

1. What is genetic counseling? Write about the scope of genetic counseling?
2. Describe the counseling for dominant defective phenotypes?
3. Write about the post-natal genetic screening methods?
4. Describe the counseling for multi factorial inheritance?
5. Discuss about the counseling for X-linked defective disorders?
6. What is the scope of genetic screening and how can we identify the disputed Paternity?
7. Write about the prenatal genetics screening methods?
8. Discuss about the risk calculation methods for different genetic disorders?
9. Describe the various types of banding techniques?
10. Write any FOUR of the following:
    a) Amniocentesis
    b) Multi factorial inheritance
    c) Genetic counseling
    d) Ultrasonography
    e) RFLP
    f) Alpha-fetoprotein (AFP)
    g) Genetic Polymorphism
    h) Gene therapy

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