ANDHRA PRADESH STATE COUNCIL OF HIGHER EDUCATION

MINOR

Subject: Human Genetics

w.e.f. AY 2023-24

COURSE STRUCTURE

Year	Semester	Course	Title of the Course	No. of Hrs /Week	No. of Credits
I	II	1	Principles of Genetics	3	3
			Principles of Genetics Practical Course	2	1
II	III	2	Human Molecular Genetics	3	3
			Human Molecular Genetics Practical Course	2	1
	IV	3	Clinical genetics and Genetic Counselling	3	3
			Clinical genetics and Genetic Counselling Practical Course	2	1
		4	Developmental and behavioural genetics	3	3
			Developmental and behavioural genetics Practical Course	2	1
III	V	5	Human Genome Project and Genome	3	3
			Human Genome Project andGenome Practical Course	2	1
		6	Cellular and Molecular Immunology	3	3
			Cellular and Molecular Immunology Practical Course	2	1

SEMESTER-II

COURSE 1: PRINCIPLES OF GENETICS

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:
1. Historical overview and laws of Inheritance
2. Understand Mendel's principles and deviations
3. Gene interactions and their outcome through gene mapping
4. Understand the mitochondrial inheritance in different organisms
5. Understand the variance and heritability of traits

II. Syllabus (Total Teaching Hours: 45)

UNIT-1 HISTORY OF GENETICS (9hr)

1. Pre-mendelian Genetic concepts, Heredity, and environment, the concept of phenotype andgenotype, pure lines and inbred lines
2. Biography of Mendel and his experiments on pea plants. Mendel laws
3. Deviations of Mendelism (Incomplete dominance and codominance; Multiple alleles, Lethalalleles, Pleiotropy, Penetrance, and Expressivity, Epistasis, and non-epistasis)

UNIT- 2 Sex Linked Inheritance and Sex Determination (9hr)

1. Chromosome theory of Sex determination: XX- XY, XX-XO, ZZ-ZW, Genic balance theoryof Bridges, Intersexes and Super sexes in Drosophila,
2. Sex differentiation in Drosophila and Man, Sex limited and Sex influenced inheritance
3. Sex determination in mammals- and role of human Y chromosome

UNIT-3 LINKAGE, CROSSING OVER, AND GENE MAPPING (9hr)

1. Linkage - Definition, Linkage group- Drosophila and man; Types of linkage-complete linkageand incomplete linkage, Significance of linkage.
2. Crossing over - definition; recombination and recombination frequency, Mechanism of crossing over: Chiasma Interference and coincidence; Coupling and Repulsion hypothesis.
3. Gene Mapping – physical mapping and genetic mapping, mapping in eukaryotes and prokaryotes

UNIT – 4 EXTRACHROMOSOMAL INHERITANCE (9hr)

1. Characteristic features of Cytoplasmic Inheritance; Inheritance of- Mitochondrial DNA, Chloroplast DNA, Kappa particles in Paramecium, Shell coiling in snail.
2. Infective heredity -Drosophila, petite mutations and mitochondrial inheritance in man
3. Epigenetics and genome imprinting in humans

UNIT -5 INHERITANCE OF QUANTITATIVE TRAITS (9hr)

1. Continuous and Discontinuous variation
2. Polygenic Inheritance and Multifactorial Inheritance
3. Genetic Variance, Heritability (narrow sense and Broad sense)

III . Skills Outcome

On Successful Completion of this Course, Student shall be able to

1. Study the Mendel Laws and their Deviations
2. Study the Chromosomal Recombination's
3. Study the Genetic Disorders
4. Identification of the Blood Groups

SEMESTER-II

COURSE 1: PRINCIPLES OF GENETICS

Practical Credits: 3 3 hrs/week

IV .

1. Mendel's laws through seed ratios& Drosophila mutants
2. Study of linkage, recombination, and chromosome mapping using test cross data.
4. Pedigree analysis for dominant and recessive autosomal and sex-linked traits.
5. Study of human genetic traits: Sickle cell anemia, Xeroderma Pigmentosum, Albinism.
6. Tests for red-green Colour blindness, Widow's peak, Rolling of the tongue, Hitchhiker's thumb, and Attached ear lobe.
7. Incomplete dominance and gene interaction through seed ratios
8. Blood Typing: ABO groups & Rh factor.
9. Study of aneuploidy: Down's, Klinefelter's and Turner's syndromes.
10. Mitosis &Meiosis through temporary squash preparation.
11. Smear technique to demonstrate sex chromatin buccal epithelial cells

V. REFFERENCES

1. Gardner, E.J., Simmons, M.J., Snustad, D.P. (1991). Principles of Genetics, JohnWiley & sons, India.8th edition.

2. Snustad, D.P. and Simmons, M.J. (2010). Principles of Genetics, John Wiley & SonsInc., India.5th edition.

3. Klug, W.S., Cummings, M.R., Spencer, C.A. (2012). Concepts of Genetics. BenjaminCummings, U.S.A. 10th edition.

4. Griffiths, A.J.F., Wessler, S.R., Carroll, S.B., Doebley, J. (2010). Introduction toGenetic

Analysis. W. H. Freeman and Co., U.S.A. 10th edition

VI. Co-Curricular Activities

a) Suggested C0-Curricular Activities

1. Assignments
2. Seminars, Group Discussions on related topics
3. Preparation of mitosis and meiosis slides
4. Pedigree preparations based on community5.Colur blindness study in a community
6. Blood group Studies

SEMESTER-III

COURSE 2: HUMAN MOLECULAR GENETICS

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:

1. On Nucleic Acids and Proteins
2. On Gene Expression
3. On DNA Replication and Their Mechanism
4. On Chromosomal Organization Of DNA
5. On Mitochondrial Genome and Nuclear Genome

II. Syllabus (Total Teaching Hours: 45hr)

Unit 1 DNA, RNA and Protein Structure (9hr)

1.1 Building blocks and chemical bonds in DNA, – structure of DNA, A-B-Z and triplex DNA,

1.2 Building blocks and chemical bonds in RNA – Structure of RNA

1.3 Building blocks and chemical bonds in peptides- primary, secondary, tertiary and quaternary structure of proteins

Unit 2 Gene expression (9hr)

2.1 Central dogma of molecular biology

2.2 RNA transcription, and RNA Processing

2.3 Translation, post-translation processing

Unit 3 DNA replication, Mutagenesis and DNA repair (9hr)

3.1 DNA replication – modes of Replication, DNA replication machinery and mechanism

3.2 DNA mutagenesis

3.3 DNA repair

Unit 4 Human Chromosome Organization (9hr)

4.1 Packaging of DNA – multiple hierarchies of DNA folding

4.2 Chromosomes as functional organelles –origins of replication, telomeres, centromeres

4.3 Heterochromatin and euchromatin

Unit 5 Human Genome organization (9hr)

5.1 Mitochondrial genome – replication, genes, genetic code

5.2 Nuclear genome – protein coding genes RNA genes

5.3 Nuclear genome – highly repetitive DNA, heterochromatin and transposon repetitive

III. SKILL OUTCOMES

On successful completion of practical course students shall be able to

1. Learn The Extractions Of DNA From Various Sources
2. Learn The Chromatographic Techniques
3. Learn The Electrophoresis Techniques
4. Learn DNA Damage By Various Assays

SEMESTER-III

COURSE 2: HUMAN MOLECULAR GENETICS

Practical Credits: 1 2 hrs/week

IV.PRACTICALS Hours 2 hours per week = 30 hours

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

V .Suggested Readings :

1. Human Molecular Genetics by T. Strachan
2. Human Molecular Genetics by Gerard Meurant
3. Human Molecular Genetics by Christopher G Mathew.
4. Human Molecular Genetics by Sudbury
5. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, JohannesZschocke.

VI. Co-curricular Activities

a) Suggested Co-curricular Activities

1. Assignments

2. Group Discussions and Seminar On Related Topics

3. Genomic Isolation Techniques

4. Molecule Separation Techniques

SEMESTER-IV

COURSE 3: CLINICAL GENETICS & GENETIC COUNSELLING

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:
1. On Single Gene Disorders
2. On Metabolic Disorders
3. On Genome Imprinting and Neurodegenerative Disorders
4. On Blood Disorders and Polygenic Syndromes
5. On Genetic Counselling and Their Risk Factors

II. Syllabus (Total Teaching Hours: 45)

Unit-1 Genetic Disorders I (9hr)

1. Monogenic diseases – Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome
2. Inborn errors of metabolism – Phenylketonuria, Maple syrup urine syndrome, galactosemia
3. Genome imprinting syndromes –Prader Willi and Angelman syndrome

Unit-2 Genetic Disorders II (9hr)

1. Genomic syndromes – Neurofibromatosis I
2. Neurogenetic disorders – Charcot Marie Tooth syndrome, spinal muscular atrophy, Alzheimer's diseases, syndromes due to triplet nucleotide expansion
3. Muscle genetic disorders – dystrophies, myotonias, myopathies

Unit-3 Genetic Disorders III (9hr)

1. Genetic Disorders of Haemopoietic systems- sickle cell anaemia, thalassemia's,haemophilia
2. Genetic disorders of eye – colour-blindness, retinitis pigmentosa, glaucoma
3. Complex polygenic syndromes – atherosclerosis, diabetes mellitus
4. Mitochondrial syndromes

Unit-4 Genetic Counselling (9hr)

1. Role of genetic counselling
2. Causes and factors for seeking counselling
3. Dysmorphology
4. Prenatal and preimplantation diagnosis

Unit-5 Practical Genetic Counselling (9hr)

1. Process of genetic counselling - Constructing a family tree, diagnostic information, risksand odds, estimation of risks
2. Genetic counselling in Mendelian disorders
3. Genetic counselling in non-Mendelian disorders
4. Ethical and legal issues in genetic counselling

III. Skill Outcomes

On successful completion of practical course students shall be able to

1. Learn Metaphase Chromosome Preparations
2. Learn Banding Techniques
3. Sex Chromatin Analysis from Different Sources
4. Learn Different Biochemical Tests

SEMESTER-IV

COURSE 3: CLINICAL GENETICS & GENETIC COUNSELLING

Practical Credits: 1 2 hrs/week

IV. Practical Syllabus Hours 2 hours per week = 30 hours

1. Metaphase chromosome preparations from bone marrow of mouse, rat, human
2. Chromosome preparation from lymphocyte culture
3. G-banding, C-banding, R-banding
4. Karyotyping
5. Meiosis in mouse testis
6. Sex chromatin (buccal mucosa, hair bud)
7. Micronuclei assay
8. Chromosome preparation from chorionic villi, stem cells, cell line
9. Sister Chromatid Exchange (SCE)
10. Molecular markers for tumour detection
11. Geneticcounseling (pedigree analysis in disease conditions, risk calculation)
12. Y-chromosome microdeletion
13. Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

V. SUGGESTED READINGS

1. Chen, Harold Atlas of Genetic Diagnosis and Counseling Springer 2012.
2. Thompson and Thompson & Thompson Genetics in Medicine, Robert L. Nussbaum, Roderick R. McInnes, Huntington F. Willard (eds)

VI. Co-curricular Activities

a) Suggested Co-curricular Activities

1. Assignments
2. Group Discussions and Seminar On Related Topics
3. Debate on Different Disorders
4. Visit to Near Genetic Counselling Center
5. Visit to Cytogenetic Labs

SEMESTER-IV

COURSE 4: DEVELOPMENTAL AND BEHAVIORAL GENETICS

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:
1. On Germ Cells And Fertilization
2. On Different Development Genes
3. On Basic Concepts Of Development
4. Embryonic Development Of Drosophila
5. Flower Development Of Arabidopsis
6. On Genetic Control Of Behavior

II. Syllabus (Total Teaching Hours: 45)

UNIT-1: Germ Cells and Fertilization (9hr)

1.Germ Cells

2. Spermatogenesis,

Oogenesis3.Fertilization

and Gastrulation

UNIT-2: Basic & Molecular Aspects of Development (9hr)

1. Potency, commitment, specification , induction ,

competence2.Maternal effect gene Gap gene, Pair rule gene

3. Segment polarity genes, Homeotic genes

UNIT-3: Genetics of Embryonic Development in Drosophila (9hr)

1. Overview of Drosophila development

2. Zygotic genes

3. Segment formation

UNIT- 4: Flower Development in Arabidopsis (9hr)

1. Development of Arabidopsis

2. Role of Homeotic Selector Gene

3. ABC model of Arabidopsis

UNIT-5:Genetic Control Of Behavior (9hr)

1. Introduction, Behavior in Invertebrates, Honeybee,
2. Drosophila – Genetic basis of alcoholism, genetic basis for sexual orientation.
3. Courtship behavior in various animals.

III. Skill Outcomes

On successful completion of the practical course students shall be able to

1. Learn Dissection of Drosophila Larvae
2. Development of Chick Embryo
3. Role of SHH Signaling

SEMESTER-IV

COURSE 11: DEVELOPMENTAL AND BEHAVIORAL GENETICS

Practical Credits: 1 2 hrs/week

IV. Practical Syllabus Hours 2 hours per week = 30 hours

1. Study of development in chick embryo
2. Dissection of the imaginal disc in Drosophila larvae
3. Life cycle of drosophila, husbandry and handling.
4. Role of SHH signaling in chick development
5. Observation of living and plastic embedded chick

embryos6.The maternal effect gene in drosophila

V. REFERENCES

1. The cell – Bruce Alberts
2. Emery's Elements of Medical Genetics- Robert. F. Mueller, Ian. D. Young.
3. Principles of Development - Wolpert
4. Principles of Genetics – Snustad, Simmons, Jenkins.

Co-curricular Activities

a) Suggested Co-curricular Activities

1. Assignments
2. Group Discussions and Seminar On Related Topics
3. Power Point Presentation on Developmental Genes

SEMESTER-V

COURSE 5: HUMAN GENOME PROJECT AND GENOMES

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:

1. On Genome Organization
2. On Different Mapping Techniques
3. On Human Genome Project
4. On Genome By Understanding The Function Of Genes
5. On Molecular Phylogenetics

II. Syllabus (Total Teaching Hours: 45)

UNIT 1 GENOME ORGANIZATION AND STUDY (9hr)

1. Genome – general features, features of eukaryotic nuclear genomes
2. Genomes, transcriptomes and proteomes
3. Genome diversity – significance of genomes – bacteria, yeast, Caenorhabditis, Homo sapiens, Arabidopsis.

UNIT 2 MAPPING GENOMES (9hr)

1. Genetic mapping – pedigree analysis, DNA markers – RFLPs, SSLPs, SNPs
2. Physical mapping – restriction mapping, FISH, radiation hybrid mapping, STS mapping
3. Sequencing genome- assembly of contiguous DNA sequence, shotgun method, clonecontigmethod, whole-genome shotgun sequencing

UNIT 3 GENOME PROJECTS (9hr)

1. Human genome project, HapMap Project, 1000 genome project, ENCODE project
2. Other genome projects.
3. Applications and proposed benefits of HGP –ELSI.

UNIT 4 UNDERSTANDING GENOME SEQUENCE (9hr)

1. Locating the genes in a genome sequence
2. Determining the functions of individual genes
3. Transcriptome – microarrays, Proteome – protein profiling

UNIT 5 MOLECULAR PHYLOGENETICS (9hr)

1. Phenetics and cladistics
2. Reconstruction of DNA based phylogenetic tree
3. Applications of molecular phylogenetics – evolutionary relationship betweenhumans and primates; origin of AIDS; human pre - history.

III . Skill Outcomes

On successful completion of practical course students shall be able to1.Purification Techniques

2. PCR
3. Sequence Alignment Techniques
4. Gene Finding Tools
5. Proteomics

SEMESTER-V

COURSE 5: HUMAN GENOME PROJECT AND GENOMES

Practical Credits: 1 2 hrs/week

IV . PRACTICAL SYLLABUS Hours 2 hours per week = 30 hours

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

V. SUGGESTED READINGS

1. Human Genome Project by James Toriello .
2. Understanding the Human Genome Project by Michael A Palladino.
3. Human Genes and Genomes: Science, Health, Society by Leon E Rosenberge, DianeDrobnis Rosenberg.
4. From Genes to Genomes: Concepts and Applications of DNA Jeremy W Dale, Malcolm von Schantz, Nick Plant .
5.Genomes 3 by Terence A Brown.
6. Principles of Gene Manipulation and Genomics by Sandy B Primrose and RichardTwyman.

VI. Co-curricular Activities

a) Suggested Co-curricular Activities

1. Assignments
2. Group Discussions and Seminar On Related Topics
3. Visit to Bioinformatics Lab
4. Conduction of Workshop And Guest Lecture Related To Bioinformatics

SEMESTER-V

COURSE 6: CELLULAR AND MOLECULAR IMMUNOLOGY

Theory Credits: 3 3 hrs/week

I. LEARNING OUTCOMES

Upon successful completion, each student will have the basic knowledge:
1. On Vaccines
2. On Immunity
3. On Monoclonal Antibody
4. On Presentation And Processing Of Cells
5. On Immunological Techniques

II. Syllabus (Total Teaching Hours: 45)

Unit 1 (9hr)

I. Introduction to Immune System, types of immunity-innate and adaptive2.Cellular components of immunity — Lymphoid cells ,Myeloid cells

3. Lymphoid organs- Primary lymphoid organs (Bone marrow & thymus); secondary lymphoidorgans (lymph node and spleen)

Uint 2 : (9hr)

1. Antigens- Immunogens, epitopes, Haptens and types of adjuvants
2. Humoral and MHC immune responses
3. Basic structure of Immunoglobulin- Immunoglobulin domains-variable region and constant region domains; isotypes, allotypes, idiotypes ,Immunoglobulin classes and its functions- IgG, IgM, IgA, IgD, Ig E

Unit -3 (9hr)

1. Polyclonal antibodies, Monoclonal antibodies- its production and applications2.Structure and organization of MHC class I and class II molecules.
3. Cell-mediated Immunity ,Hypersensitivity- Types (I, II, III & IV)
4. Immunodeficiency disorders- primary immunodeficiency disorders (SCID), secondary immunodeficiency disorders (AIDS)

Unit -4 (9hr)

1. Vaccines- historical background and principle; passive & active immunization.attributes of effective vaccines
2. Types of vaccines- live attenuated and inactivated killed vaccines. sub-unit vaccines, DNA vaccines, edible vaccines

Unit-5 (9hr)

1.General features of ag-ab reactions- Agglutination, neutralization, complement fixation, opsonisation. Immunoprecipitation, immunoelectrophoresis, immunodiffusion Tests

2 ELISA — Types , Immuno fluorescence assays (direct & indirect) Principle and applications

3.Western blot -Principle, procedure and applications , Flow cytometry -Principle, methodologyand applications

III .Skill Outcomes

On successful completion of the practical course students shall be able to

1. Blood Grouping
2. Immunological Technique

SEMESTER-V

COURSE 6: CELLULAR AND MOLECULAR IMMUNOLOGY

Practical Credits: 1 2 hrs/week

IV Practical's Syllabus Hours 2 hours per week = 30 hours

1. ABO blood typing
2. Differential count of lymphocytes
3. Single Radial Immunodiffusion
4. ELISA
5. Agglutination
6. Haemagglutination test
7. Coomb's test
8. Western Blot

V . References

1. Essential Immunology by I.Roitt,Publ:Blackwell
2. Immunology by G. Reever & I.Todd,Publ:Blackwell
3. Immuno diagnostics by S.C. Rastogi, Publ:NewAge
4. Immunology by Richard A.Golds by, Thomas J Kindt, Barbaraa. Osborne,
5. Janiskuby
6. Fundamental immunology by WilliamE.Paul
7. Basic Immunology by Bhoosreddy G.L. and WadherB.J.
8. Text book of immunology by BarujBenacerraf
9. Immunology by Kuby:Publ:Freeman

VI .Co-Curricular Activities

1. Assignments.
2. Charts on complement pathway, MHC I & II
3. Group discussions and Student seminars.
4. Visit to diagnostic labs