BHARATHIAR UNIVERSITY: COIMBATORE - 641 046

M. Sc. HUMAN GENETICSAND MOLECULAR BIOLOGY (CBCS PATTERN) (For the students admitted during the academic year 2017-2018 batch and onwards)

Duration of the course: 2 Years Eligibility Condition:

Annexure No;61A

SCAA Dated: 03.07.2017

B.Sc. Genetics B.Sc. Biochemistry

B.Sc. Animal Sciences B.Sc. Biotechnology

B.Sc. Microbiology/Environmental Sciences B. Pharm

B.Sc. Forensic Sciences B.Sc. Bioinformatics

B.Sc. Zoology B.E./B. Tech. Biotechnology

BAMS/BHMS Ayurveda and Homeopathy

B. Tech. Genetic Engineering

All Branches in Life Sciences B. Tech. Industrial Biotechnology

MBBS/B.Sc., Nursing B.Sc., Animal Science and Zoology

B.Sc., Advanced Zoology and Biotech B.Sc., Agricultural Biotechnology

M.Sc., Human Genetics and Molecular Biology - Semester Wise Syllabus and Scheme

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Semester	Core	Subject Code	Title of the Paper	Hours/ Week	Internal	External	Total Marks	Total Credits
I	Core-I	-	Biochemistry	4	25	75	100	4
	Core-II	-	Cell Biology	4	25	75	100	4
	Core-III	-	Principles of Human Genetics	4	25	75	100	4
	Core-IV	-	Molecular Genetics	4	25	75	100	4
	Core Practical-	-	Practical-I (Biochemistry, Cell Biology, Human Genetics, Molecular Genetics)	6	25	75	100	4
	Elective-I	-	Genomics and Proteomics (or) r-DNA Technology	4	25	75	100	4
	Supportive-I	-	Genetics and Society	2	12	38	50	2
	Cana M	I	Harris Catalan C		25	7.5	100	1
	Core-VI	-	Human Cytogenetics	4	25	75	100	4
	Core-VI	-	Medical Genetics	4	25	75	100	4
TT	Core-VII	-	Developmental and Behavioral Genetics	4	25	75 75	100	4
II	Core-VIII	-	Biostatistics	4	25	75	100	4
	Core Practical- II	-	Practical-II (Human Cytogenetics, Medical Genetics, Developmental and Behavioral Genetics and Biostatistics)	6	25	75	100	4
	Elective-II	-	Bio Instrumentation (or) Nanobiology	4	25	75	100	4
	Supportive-II	-	Principles of Genetics	2	12	38	50	2
	Core-IX	-	Immunogenetics	4	25	75	100	4
	Core-X	-	Cancer Genetics	4	25	75	100	4
	Core-XI	-	Neurogenetics and Epigenetics	4	25	75	100	4
III	Core-XII	-	Bioethics and Biosafety	4	25	75	100	4
	Core Practical- III	-	Practical-III (Immunogenetics, Cancer Genetics, Genetic counseling, Bioethics and Biosafety)	6	25	75	100	4
	Elective-III	-	Stress and Biomarkers (or) Stem cell biology	4	25	75	100	4
	Supportive-III	-	Genetic toxicology	2	12	38	50	2
IV	Core-XIII	-	Genetic Counseling	4	25	75	100	4
	Project and		Project and Viva Voce	-	-	-	150	6
	Viva		Hospital visits/ Summer internship*	-	-	-	15	
			Research /Review Papers in Genetics	_	_	-	15	2
			Industrial Visit I (I Year) *	_	-	-	10	
			Industrial Visit II (II Year) *	-	-	-	10	
			Total				2250	90
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^{*} Report to be submitted along with Project Work

Practical Components:

The M.Sc., HGMB Core Practical Examination having the following Marks:

Internal Marks: 25 Time: 1.30 minutes

Major Practical		10 Marks
Minor Practical		5 Marks
Spotters (A, B, C, D and E)	5x2	10Marks

Total = 25 Marks

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External Marks: 75

Major Practical		20Marks
Minor Practical		10Marks
Minor Practical		10Marks
Spotters (A, B, C,D and E)	5x4 =	20Marks
Record &Viva		10+5 Marks

Total = 75Marks

Theory Components:

The M.Sc., HGMB Core and Elective theory Examination having the following Marks.

Core and Elective Papers: Maximum Marks-100

Internal Marks: 25

-15 Marks Test Assignment - 5 Marks Seminar - 3marks

External Marks: 75

Section-A:10x1=10 Marks (Question No. 1 to 10)

Choose the best Answer type. Answer all questions. All questions carry equal marks.

Section-B: 5x5 = 25 Marks (Either or type - Question No. 11 to 15)

Answer all questions. All question carry equal marks. Each answer should not exceed 2 pages.

Section-C: 5x8 = 40 Marks (Either or type-Question No. 16 to 20)

Answer all questions. All Question carry equal marks. Each answer should not exceed 4 page

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Supportive Papers: Maximum Marks-50

Internal Marks: 12

Test -6 Marks Assignment -3 Marks Seminar -3 Marks

External Marks: 38

Section-A: 5x1=5 Marks (Question No. 1 to 5)

Choose the best Answer type. Answer all questions. All questions carry equal marks.

Section-B: 3x3 = 9 Marks (Either or type-Question No. 6 to 8)

Answer all questions. All Question carry equal marks. Each answer should not exceed 1 page.

Section-C: 4x6 = 24 Marks (Either or type-Question No. 9 to 12)

Answer all questions. All Question carry equal marks. Each answer should not exceed 2 pages.

Core-I

BIOCHEMISTRY

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Unit-I

Introduction to Biochemistry, Structure of atoms, molecules and chemical bonds, Stabilizing interactions. Principles of biophysical chemistry, Bioenergetics, oxidativephosphorylation coupled reaction, group transfer, biological energy transducers.

Unit-II

Amino acids: Structure, Classification, Properties, Isoelectric pointand Zwitter ions. Biosynthesis of essential and non essential amino acids. Proteins: Structure, Classification, Conformation of proteins (Ramachandran plot). Principles of catalysis, enzymes and enzyme kinetics, enzyme regulation, mechanism of enzyme catalysis, isozymes.

Unit-III

Carbohydrates: Classification, Structure of Mono, Oligo and Polysaccharides. Glycoproteins: Synthesis of Glycosides. Lipids: Classification, Structure, Lipids and Biological membranes. Biosynthesis of Triglycerides, Cholesterol.

Unit-IV

Nucleic acids:DNA structure and Properties, Biosynthesis of Purines and Pyrimidines, Types of RNA, Replication of DNA, Mode of DNA replication. Conformation of nucleic acids (A-DNA, B-DNA and Z-DNA),t-RNA, micro-RNA.

Unit-V

Glycolysis and TCA cycle, Glycogen breakdown and synthesis, Gluconeogenesis, HMP Shunt.Co-ordinated control of metabolism, Oxidation of fatty acids.

- 1. Biochemistry, by D.Voet and J.G. Voet, 2004. John Wiley and Sons, USA Biochemistry, by R.H. Garrett and C.M. Grisham, (3rd Edition) 2007Saunders College Publishers.
- 2. Principles of Biochemistry by Albert L. Lehninger (4th edition) 2004. CBS Publishers and Distributors, New Delhi.
- 4. Biochemistry by LubertStryer (4th edition) 2000. Freeman International Edition.
- 5. Biochemistry by KeshavTrehan, 1990. Wiley Eastern Publications.
- 6. Fundamentals of Biochemistry by J.L.Jain et al. (4th edition) 1994. S.Chand and Company.
- 7. The Biochemistry of Nucleic acid, Tenth Edition, Roger L.P. Adams, John T. Knowler and David P. Leader, 1992. Chapman and Hall Publications.
- 8. Biochemistry. S. C. Rastogi, 2nd edition. 2003. Tata McGraw Hill Publishing Company, Ltd., N. Delhi.
- 9. Biochemistry: The Molecular Basis of Life. Trudy McKee and James R. McKee. Third Edition.2003.

Core-II CELL BIOLOGY

Unit-I

Cellular organization:Structure of cell organelles-An overview, Cell wall and membrane structure. Membrane constituents:phospholipids, glycolipids, cholesterol, membrane proteins, receptors and phospholipases, phospholipid bilayer:Structure, asymmetry, fluid mosaic model of random diffusion of membrane components, domains in membrane,natural and artificial membranes passive movements of solutes, ion distribution, mediated permeation, ionophores.

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Unit-II

Membrane transport of small molecules and the ionic basis of membrane excitability, principles of membrane transport, carrier proteins and active membrane transport, ion channels and electrical properties of membranes, Cell junctions and cell adhesion molecules, basement membrane, extracellular matrix.

Unit-III

Structure of nuclear envelope, the nuclear pore complex, Selective transport of proteins to and fro from nucleus, Regulation of nuclear protein import, transport of RNA's. Internal organization of nucleus, Chromosomes.

Unit-IV

Cell Communication: General principles of Cell signaling, types and Mechanisms, Cell surface receptors, GPCR Molecular structure and functions, Enzyme linked Receptors, Activated Tyrosine kinase and MAP kinase pathways. Cell Division: Overview of cell cycle, Mitosis and Meiosis. Cell Cycle control in mammalian cells, Checkpoint in cell cycle regulation

Unit-V

Cell biology of Cell aging process and its significance, Molecular Mechanism of Cell death: Cell necrosis and Apoptosis. CASPASE types and molecular mechanisms, proapoptotic regulators, Inhibitors of Apoptosis, Molecular biology of Survivin and Bcl2 family members.

- 1. Cell and Molecular Biology by Lodish, 2004.
- 2. The Cell by Gerald Karp, 2002.
- 3. Cell, A molecular Approach by Cooper, 2000.
- 4. Molecular Cell Biology, Alberts, 2000.
- 5. Cell and Molecular Biology, De Robertis, 2001.
- 6. Principles of Biochemistry by Albert L. Lehninger (4th edition) 2004. CBS Publishers and Distributors, New Delhi.
- 7. Cell Biology, Genetics, Molecular Biology, Evolution and Ecology (English) 14th Edition by Verma and Agarwal.

Core-III PRINCIPLES OF HUMAN GENETICS

Unit-I

History and development of human genetics, genes, law of segregation, law of independent assortment, chromosomal basis of segregation and independent assortment, linkage, crossing over.

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Unit-II

International system of Human Chromosome Nomenclature, Nomenclature of aberrant karyotypes, common syndromes due to numerical chromosomal changes, Structural alterations (translocations, duplications, deletions, microdeletion, fragile sites). Chromosomal basis and non chromosomal basis of sex determination.

Unit-III

Monogenic traits, autosomal inheritance, dominant, recessive, Sex-linked inheritance, Sex-limited and sex-influenced traits, mitochondrial inheritance, MIM number, consanguinity and its effects, Complex traits, approaches to analysis of complex traits, role of family and shared environment, monozygotic and dizygotic twins and adoption studies.

Unit-IV

Pedigree, gathering family history, pedigree symbols, construction of pedigrees, presentation of molecular genetic data in pedigrees, Complications to the basic pedigree patterns, genomic imprinting and uniparental disomy, spontaneous mutations, mosaicism and chimerism, male lethality, X-inactivation.

Unit-V

Inborn errors of metabolism:Phenylketonuria. Neurogenetic disorders: Alzheimer's diseaseMuscle genetic disorders:Duchenne Muscular Dystrophy. Genetic disorders of Haematopoitic system: Sickle cell anemia. Multifactorial disorders: Diabetes mellitus. Mitochondrial syndromes, Management of genetic disorders.

- 1. Human Genetics, Lewis, 1999.
- 2. Basic human Genetics, Mange and Mange, 1999.
- 3. Molecular and genetic analysis of human traits, Maroni, 2001.
- 4. An introduction to molecular genetics, Pasternack, 2000.
- 5. Human Molecular genetics, Strachan and Read, 2003.
- 6. An introduction to genetic analysis, Griffiths and Miller.
- 7. Lewin, Genes IX, 9th Edition Jonesand Bartlett 2007.
- 8. Snustadand Simmons, Principles of Genetics, 4th Edition, Wiley' 2005.
- 9. Alberts*et al.*, Molecular Biology of TheCell 2nd Edition, Garland 2007.

CORE-IV

MOLECULAR GENETICS

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Unit-I

Fundamentals of genes and chromosomes, general concept of a gene, gene families, non-coding genes, repetitiveDNA, replication, transcription, translation and post translational changes in prokaryotes and Eukaryotes.

Unit-II

Fundamentals of DNA cloning and molecular hybridization: Cell based DNA cloning, DNA hybridization assays, PCR based DNA cloning and DNA analyses. Typesof mutations and nomenclature. Mutagenesis and DNA repair: Types of DNA damage, Endogenous and exogenous origins of DNA damage, DNA repair pathways: Error-prone, Mismatch, photoreactivation, excision and SOS.

Unit-III

Homologous recombination: Models and molecular mechanisms, Gene conversion: Molecular mechanisms, site specific recombination, transposons and transposition mechanisms.

Unit-IV

Features of the human Genome: Organization and expression of the human genome, human multigene families. The human genome project: Mapping of the human genome: Physical mapping Genetic mapping. Footprints of evolution, mutation and instability of human DNA.

Unit-V

Dissecting and manipulating genes: Studying human gene structure and function and creating animal models of disease, gene theory and other molecular genetic based therapeutic approaches

- 1. Tom Strachan and Andrew. P. Read, Human Molecular Genetics, Bios" Scientific Pub UK. (1996).
- 2. Watson, J.D., Hopkins, N.H., Roberts, J.W., Steitz, J. and Weinter, A.M., Molecular Biology of Genes (4th edition) 1987. The Benjamin/Cummings publishing Company Inc., Joky.
- 3. Lewin, B. Genes VI (1997). Oxford University Press, Oxford, New York, Tokyo.
- 4. Darvell, J.et.al., Molecular Cell Biology (7th edition) 2002. Garland Publishing Iwc., New York.
- 5. Lewin, Genes IX, 9th Edition Jones and Bartlett 2007.
- 6. Principles of Genetics Gardner, Simmons, Snustad 8th Edition 2006.
- 7. Molecular Biology by Glick and Pasternick, 2003.

PRACTICAL-I

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BIOCHEMISTRY

- 1. Determination of blood glucose
- 2. Estimation of total cholesterol
- 3. Estimation of total protein and albumin
- 4. Estimation of AST, ALT and ALP
- 5. Estimation of Urea
- 6. Estimation of Creatinine
- **7.** TLC separation of amino acids

CELL BIOLOGY

- 1. Uses of Microscope and Micrometry
- 2. Preparation of blood smear
- 3. Counting of RBC and WBC using Heamocytometer
- 4. Slides for Mitosis and Meiosis
- 5. Preparation of medium and cultivation of Human cell lines
- 6. MTT assay
- 7. DNA Fragmentation Assay

HUMAN GENETICS AND MOLECULAR GENETICS

- 1. Isolation of Blood genomic DNA
- 2. Isolation of RNA
- 3. Isolation of Plasmid DNA
- 4. Estimation of DNA and RNA
- 5. Restriction digestion and ligation
- 6. Southern blotting
- 7. Polymerase Chain Reaction
- 8. Retrieval of sequences from Nucleic acid databases
- 9. Database similarity search tools, BLAST

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Elective-I

GENOMICS AND PROTEOMICS

Unit-I

Genomics, genes and chromosomes. Genome: Structure and organization of prokaryotic and eukaryotic genome, genetic and physical mapping. Assembly of a contiguous, DNA sequence, clone contig approach, whole genome shot gun sequencing. Human Genome Project: Importance and impact.

Unit-II

Genomics of *E. coli*, *Arabidopsis thaliana* and *Musmusculus*. Pharmacogenomics: High throughput screening for discovery and identification of drugs. Drug targets and developmentSNP analysis.

Unit-III

Function prediction of Gene, computational and experimental analysis. Transcriptomics, transcriptome, yeast transcriptome and the human transcriptome, link between the transcriptome and proteome. Transcripts analysis, SAGE, non-array based whole transcriptome analysis, differential display, Yeast two hybrid systems.

Unit-IV

Proteomics: Tools for proteome analysis, 2D, PAGE, Mass spectrometry, MALDI, TOF, TANDOM, MS, LC, MS, protein microarray, SAGE. Protein: Protein interactions and uses of their databases. Peptide finger printing: Techniques for protein purification, sequencing of proteins.

UNIT-V

Biological Databases: Overview, applications, gene and protein sequence databases, GenBank, EMBL, DDBJ, and PDB. Sequence alignment and sequence analysis: Concept of local and global sequence alignment, Pair-wise sequence alignment, BLAST, Multiple sequence alignment, homology, analogy.

- 1. Brown, T.A., 2006, Genomes, John Wiley and Sons, Pvt. Ltd., Singapore. Campbell A,Heyer.
- 2. 2004, Discovering Genomics, Proteomics and Bioinformatics, Pearson Education, New Jersey.
- 3. Liebler, Daniel, C., 2002, Introduction to proteomics tool for the new biology, Humana Press, New Jersey.
- 4. Lesk, A.M. 2007. Introduction to Bioinformatics, Oxford University Press, Oxford.
- 5. Old, R.W. and Primrose, S.B. 2006. Principles of Gene Manipulation, Blackwell Science Publication, Berlin.
- 6. Pennington, S.R, Dunn,M,J., 2002, Proteomics from Protein sequence to function, Viva Books Pvt., Ltd, New Delhi.
- 7. Introduction to Bioinformatics, TramontanoA, Chapman and Hall.
- 8. Understanding Bioinformatics, Zvelebil M and Baum JO, Taylor and Francis.

Elective-I

r-DNA TECHNOLOGY

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UNIT-I

Genes within the cells, genetic elements that control gene expression, restriction and modification enzymes (Restriction enzymes, DNA ligases, Klenowfragment, T₄DNA polymerase, Polynucleotide kinase, Alkaline phosphatase), safety guidelines of recombinant DNA research.

UNIT-II

Construction of genomic DNA and BSC libraries, BSC-Y libraries, design of linkers and adaptors. Characteristics of plasmid and phage vectors, prokaryotic and eukaryotic expression vectors, Insect, yeast and mammalian vectors.

UNIT-III

DNA sequencing (Maxumand Gilbert, Sangers, Pyro, sequencing, Shotgun sequencing method), Protein sequencing, RNA sequencing, Southern and northern and western blotting, *In situ* hybridization, Site-directed mutagenesis, DNA labelling, DNA fingerprinting (RAPD, RFLP, AFLP).

UNIT-IV

Isolation of DNA, mRNA and total RNA, polymerase chain reactions (PCR) and modified PCR, gene isolation, gene cloning, screening and expression of cloned gene, transposons and gene targeting.

UNIT-V

Production of insulin, human growth factor, gene therapy (antisense and ribozyme technology), human genome project and its application. Large scale gene expression analysis (Microarray for DNA and protein), strategies for genome sequencing.

- 1.Old and Primrose, Principles of Gene Manipulation, 3rd Ed, Blackwell Scientific Publishers.
- 2. Genetic Engineering by S. Rastogi and N. Pathak, Oxford Univ. Pub.
- 3. Recombinant DNA Technology: Setubal: Introduction to computational Molecular Biology. Cengage Learning India(P) Limited.
- 4. D.M. Glover, Genetic Engineering, Cloning DNA, Chapman and Hall, New York, 1980.
- 5.B. R. Glick and J.J. Pasternak, MolecularBiotechnology: Principles and Applications of Recombinant DNA, ASM press.
- 6. Watson, J.D., Gilman, M., Witkowski, J., Zoller, M., Recombinant DNA, ScientificAmerican Books, New York, 1992.
- 7. H.K. Das, Text Book of Biotechnology, 1st ed, 2004, Wiley Publishers.
- 8. Brown, T.A., Genetics a Molecular Approach, 4th Ed. Chapman and Hall, 1992.
- 9. D. M. Glover and B.D. Hames, DNA cloning: A Practical Approach, IRL Press.
- 10. Brown TA, Genomes, 3rd ed. Garland Science 2006.

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Supportive-I

GENETICS AND SOCIETY

Unit-I

The history and impact of Genetics in Medicine and Society, Medical Genetics, Early beginnings, Centers for Genetics and Societyaround the world, an overview. Genes and Pedigrees in a Population. Creation of Awareness about Genetic diseases and disorders.

Unit-II

Human Genome Project, Beginning and Organization of the HGP, Sequencing of the Human Genome, Promises and Achievements, Ethical, Legal and Social issues of the HGP, Other Genome Projects initiated as a direct consequence of HGP completion, Human Genome Diversity Project.

Unit-III

Advances in Modern Genetics: Some of the areas of concern in Modern Genetics, GM crops, personal DNA data, Gene Therapy, Pharming, Ethical and Legal issues in Medical Genetics.

- 1. Genetics, A Conceptual Approach, 4th ed., B.A. Pierce, Palgrave Macmillan, 2012.
- 2. Emery's Elements of Medical Genetics, 14th ed., P.D. Turnpenny and S. Ellard, Churchill Livingstone, 2012.
- 3. Introduction to Genetics, A Molecular Approach, T. Brown, Garland Science, 2012.
- 4. Genome Duplication, Concepts, Mechanism, Evolution and Disease, M.L. De Pamphilis and S.D. Bell, Garland Science, 2011.
- 5. Human Molecular Genetics, 4th ed., T. Strachan and A. Read, Garland Science, Taylor and Francis Group, 2011.
- 6. A Guide to Genetic Counseling,2nd ed., W.R. Uhlmann, J.L. Schuette and B.M. Yashar, Wiley, Blackwell, 2009.

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Core-V HUMAN CYTOGENETICS

Unit-I

History of human chromosome research, Denver conference (1940), Chicago conference (1966), Paris conference (1971), nomenclature of human chromosome. Identification of Human diploid chromosome, peripheral blood cultures, banding techniques: G,Q,C and Rband, identification of 23 pairs of human chromosomes by band position.

Unit-II

Chromosomal, autosomal and sex chromosomal syndromes, structural chromosomal syndromes. Syndromes and disorders, molecular pathology of monogenetic diseases, Cystic fibrosis. Inborn errors of metabolism: Phenylketonuria and Galactosemia.

Unit III

Genetics in medical practice, genetic principles and their application in medical practice, case studies (Interacting with patients, learning family history and drawing pedigree chart).

Unit-IV

Prenatal diagnosis: Chorionic villi sampling, foetoscopy, ultrascopy, amniocentesis. Postnatal diagnosis: Peripheral blood leucocyte culture, sister chromatid exchange, fragile site, Mitotic index, Genetic Counseling

Unit-V

Hereditary forms of cancer, oncogenes and cancer, chromosomes and cancer. Cancer and the environment: physical, chemical and biological carcinogens. Genetic predisposition to sporadic and non-sporadic cancer.

- 1. Human Heredity Principles and issues, by Michael R. Cumming's. 3rd Edition.
- 2. Genetics Medicine, by Karl. H. Muench Elsevier Pb. London.
- 3. Human Genetics by Elof Axel Carlson, TATA McGraw, Hill Pb. New Delhi.
- 4. Attwood, T.K. and Parry Smith, D.J. 1999, Introduction to Bioinformatics, Longman Publications, Pearson Education Ltd., New Delhi.
- 5. Baxevanis, A.D. and Francis Ouelletle, B.P., 1998, Bioinformatics, A Practical Guide to the Analysis of Genes and Proteins, Wiley, Interscience Publication, New York.
- 6. Bishop, M.J. and Rawlings, C.J., 1987, Nucleic Acid and Protein Sequence Analysis, A Practical Approach, IRL Press, Oxford.
- 7. Brown, T.A., Genomes, 1999, John Wiley and Sons Inc., New York.
- 8. Zhang, W.E.I. and Shmulevich, I.Y.A. 2002, Computational and Statistical Approaches to Genomics, Kluwer Academic Publishers, London.

MEDICAL GENETICS

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Core-VI Unit-I

An overview of the genetic basis of syndromes and disorders, monogenic diseases with well known molecular pathology, Cystic fibrosis, TaySach's syndrome, Marfan syndrome.

Unit-II

Inborn errors of metabolism- Genetic bases and Classification, Phenylketonuria, Maple syrup urine syndrome, Mucopolysaccharidosis, Galactosemia. Genome imprinting Syndromes: Prader-Willi and Angelman syndromes, Beckwith-Wiedemann Syndrome.

Unit-III

Neurogenetic disorders: Major regions of human brain and nerve conduction, Charcot-MarieTooth syndrome, spinal muscular atrophy. Syndromes due to triplet nucleotide expansion: Alzheimer's disease. Muscle genetic disorders: Dystrophies (Becker Muscular Dystrophy) myotonias, myopathies.

Unit-IV

Genetic disorders of Haemopoitic systems: Overview of Blood cell types and haemoglobin, Sickle cell anemia, Thalassemia, Hemophilia. Genetic disorders of Eye: Colour Blindness, Retinitis pigmentosa, Glaucoma, Cataracts.

Unit-V

Complex polygenic syndromes: Hyperlipidemia, Atherosclerosis, Diabetes mellitus, mitochondrial syndromes, Management of genetic disorders.

- 1. Clinical Genetics, A short course by Wilson, 2000.
- 2. Principle and Practice of Medical Genetics, Rimoinet al., 2002.
- 3. Genes in Medicine, Rasko and Doumes, 1995.
- 4. An introduction human molecular genetics, Pasternack, 2000.

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Core-VII DEVELOPMENTAL AND BEHAVIORAL GENETICS

Unit-I

Early development, fertilization, types of cleavage, gastrulation: Cell movement and formation of germ layers in frog, chick and mammals. Concept of determination, competence and differentiation. *Drosophila*: Maternal genes and formation of body axes and signaling pathways in development, segmentation genes, homeotic genes function, imaginal disc development and sex determinations.

Unit-II

Differentiation of germ cells and gametogenesis, fertilization and implantation, development of vertebrate nervous system, formation of neural tube, formation of brain regions, Axes formation and HOX genes, genetic determination of sex in mammals, stages of human embryonic development. Reproductive failure and infertility and assisted reproduction

Unit-III

Programmed rearrangements in genes, chromatin diminution, endoreplication cycles, gene amplification, genome imprinting, congenital malformations and teratogenesis, epigenetic regulation. Regeneration, Senescence, Embryonic stem cells and their applications.

Unit-IV

Nature and behavior, Genetic experiments to investigate animal behavior, identifying genes for behavior, investigating the genetics of human behavior, Twin and adoption study designs, interpreting heritability, Linkage and association studies.

Unit-V

Neurogenetic study designs: Genetic and environmental manipulations, learning and memory. Psychopathology, dementia, schizophrenia, mood disorders, anxiety disorders. Disorders of childhood personality and personality disorders, antisocial personality, criminal behavior. cognitive disabilities, mental retardation, learning disorders, Communication disorders.

- 1. Scott F. Gilbert, Developmental Biology, VIII edition, Sinauer Associates Inc., Publishers, Sunderland, Massachusetts USA (2006).
- 2. Bruce Alberts, A. Johnson, J. Lewis, M. Raff, K. Roberts, P. Walter (2008). Molecular Biology of the cell, V edition, John Wiley and sons Inc., 2008.
- 3. Benjamin Lewin (2010), Genes X, Jones and Bartlett Publishers, England
- 4. Principles of Developmental biology, Wolpert, 2002
- 5. Synopsis of psychiatry, Kaplan and Sadock, 1998
- 6. Behavioural genetics, Plominet al., 2001

Core-VIII

BIOSTATISTICS

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UNIT-I

Statistical population and sample in biological studies, variables, qualitative and quantitative, discrete and continuous. Sampling methods: Probability and non-probability methods, Frequency distribution, Representation of data, tables, diagram and graph.

UNIT-II

Measures of central tendency, mean, median and mode. Measures of dispersion, range, mean deviation, quartile deviation, standard deviation, variance, standard error. Probability, addition and multiplication rules, Bayes theorem, Probability distribution, binomial, Poisson and normal distribution.

UNIT-III

Testing of hypothesis: Null and alternate hypothesis, test for significance for large samples and test for significance for small samples. Chi-square test, test of independence, goodness of fit and homogenecity.

UNIT-IV

Correlation: Types, methods of correlation, graphic method, Karl Pearson's Correlation, SpearmanRank Correlation. Regression analysis, equation, estimation of unknown value from known value.

UNIT-V

Sign test for paired data, rank sum test. Kolmogorov-Smirnov, test for goodness of fit, comparing two populations. One sample run test, rank correlation. ANOVA, One way and two way classification.

- 1. Gupta S.P. 1987. Statistical Methods. Sulton Chand and Sons Publishers, New Delhi.
- 2. Attwood, T.K. and Parry, D.J.Smith, D.J. 2002. Introduction to Bioinformatics. Pearson Education (Singapore) Ptc. Ltd.
- 3. Palanichamy, S. Manoharan, M. 1994. Statistical methods for Biologists, Palani Paramount Publications, Tamil Nadu.
- 4. Biostatistics : A foundation for Analysis in the Health Sciences 7/E Wayne W. Daniel, Wiley Series in Probability and Statistics.
- 5. Introductory Statistics. Fifth Edition. (2004) Prem S. Mann. John Wiley and Sons (ASIA) Pte Ltd.
- 6. Basic Statistics-Aprimer for Biomedical Sciences- (Olive Jean Dunn).

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PRACTICAL-II

HUMAN CYTOGENETICS

- 1. Problems related to Mendelian laws
- 2. Pedigree analysis
- 3. Peripheral blood leukocyte culture for chromosomal studies
- 4. Mitotic indices
- 5. Sister chromatid exchange, determination
- 6. Micronucleus test
- 7. Chromosomal disorders: Numerical and Structural

HUMAN MEDICAL GENETICS

- 1. Gene polymorphism study by using RAPD and RFLP
- 2. COMET Assay
- 3. ELISA
- 4. Western blotting analysis
- 5. PCR detection of mutation using specific primers

DEVELOPMENTAL AND BEHAVIORAL GENETICS

- 1. Live Observation of *Drosophilla melanogaster* embryo
- 2. Study of gene expression during development with Lac-Z reporter gene in embryos
- 3. Gene expression in embryos *D. melanogaster*
- 4. Live Observation of Chick embryo
- 5. Dissection and mounting of Imaginal discof Drosophilla melanogaster
- 6. Case studies, learning disorders, Mental retardation

BIOSTATISTICS

1. Learning of SPSS software

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Elective-II

BIOINSTRUMENTATION

UNIT-I

Centrifugation:Principles of centrifugation, different types of instruments, rotors and its applications. Chromatography: Principles and application of adsorption, partition and ion, exchange chromatography, gel filtration, affinity, high performance liquid chromatography and gas liquid chromatography.

UNIT-II

Electrophoresis: Moving boundary and zonal electrophoresis, gel electrophoresis (Native PAGE, SDS PAGE, agarosegel electrophoresis, Real Time PCR), isoelectric focusing technique. Immuno-electrophoresis, ELISA and RIA.

UNIT-III

Radioactivity: Disintegration of radionuclides, half-life of radioactive compounds, determination of radioactivity by Geiger Muller counter and scintillation counting, isotopic tracer techniques and autoradiography. Applications of radio isotopes in biological and medical sciences.

UNIT-IV

Spectrophotometry: Beer, Lamberts law, extinction coefficient and its importance, design of colorimeter and spectrophotometer. Principles of atomic absorption spectrophotometry andits application in Biology. Principles and applications of x-ray diffraction and NMR in structure determination.

UNIT-V

Principles and Applications of Light, Phase Contrast, Fluorescence Microscopy, Scanning and Transmission Electron Microscopy, Confocal Microscopy, Cytophotometry and Flow Cytometry, advances of microscopy.

- 1. Skoog, D.A. *et al.*, "Principles of Instrumental Analysis", 5th Edition, Thomson / Brooks, Cole, 1998.
- 2. Braun, R.D. "Introduction to Instrumental Analysis", Pharma Book Syndicate, 1987.
- 3. Biophysical chemistry, Upadhyay, Upadhyay and Nath.
- 4. Instrumental methods of chemical analysis, P.K. Sharma.
- 5. Handbook of Biomedical Instrumentation, R.S. Khandpur, Tata McGraw Hill.
- 6. Practical Biochemistry, Principles and techniques, Wilson. K and Walker. J.
- 7. A Biologist's guide to principle and techniques of practical biochemistry, Brigan L. Williams.
- 8. Experimental methods in Biophysical chemistry, Nicolau, C.

Elective-II

NANOBIOLOGY

Annexure No;61A

SCAA Dated: 03.07.2017

UNIT-I

Scientific revolution, atomic structures, molecular and atomic size, emergence of Nanotechnology, carbon age, new form of carbon, Challenges in Nanotechnology.

UNIT-II

Carbon nanotubes, metals (Au, Ag) and Metal oxides (TiO₂, CeO₂, ZnO). Physicochemical characteristics of nanomaterials. Infra red spectroscopy (IR), UV-visible, Absorption and Diffraction analyses X-ray diffraction. Scanning Electron Microscope (SEM), Transmission Electron Microscopy (TEM).

UNIT-III

Nanomoleculardiagnostics and Biosensor. Nanodiagnostics: Nanoparticles for molecular diagnostics, DNA nanomachines, Nanobiosensor, CNT biosensor, DNA nanosensor, Nanowire biosensor, application of nanodiagnostics.

UNIT-IV

Nanopharmaceutical:Nanobiotechnology for drug discovery, protein and peptide based compounds for cancer and diabetes, drug delivery, nanoparticle based drug delivery, lipid nanoparticles, vaccination, cell therapy, Gene therapy.

UNIT-V

Health impact of Nanomaterials: Source of nanoparticles, Handling of Nanomaterials, entry routes into the human body: Lungs, inhalation, deposition and translocation, intestinal tract, skin and eye. Nanoparticle interaction with biological membrane, Neurotoxicology.

- 1. M. Wilson, K. Kannangara, G Smith, M. Simmons, B. Raguse, Nanotechnology: Basic science and Emerging technologies, Overseas Press India Pvt Ltd, New Delhi, First Edition, 2005.
- 2. C.N.R. Rao, A. Muller, A.K. Cheetham (Eds), The chemistry of nanomaterials: Synthesis, properties and applications, Wiley VCH VerlagGmbhandCo, Weinheim, 2004.
- 3. Kewal K. Jain, The Hand book of Nanomedicine, Humana Press, Springer 2008.
- 4. Dr. ParagDiwan and AshishBharadwaj (Eds), Nano Medicines, Pentagon Press, 2006.
- 5. Challa S.S.R. Kumar, Nanomaterials for medical diagnosis and therapy, Viley, VCH, 2007.
- 6. Nancy A. Monteiro, Riviere and C. Lang Tran, Nanotoxicology: Characterization, Dosing and Health Effects, Informa Healthcare, 2007.
- 7. Kumar, Challa S. S. R. (ed.) Nanomaterials, Toxicity, Health and Environmental Issues, Wiley, VCH, Weinheim, 2006.

Annexure No;61A SCAA Dated: 03.07.2017

Supportive-II

PRINCIPLES OF GENETICS

Unit-I

Mendelian principles: Dominance, segregation, independent assortment, deviation from Mendelian inheritance, Concept of gene, Allele, multiple alleles, pseudoallele, complementation tests.

Unit-II

Extensions of Mendelian principles, Codominance, incomplete dominance, gene interactions, pleiotropy, genomic imprinting, penetrance and expressivity, phenocopy.

Unit-III

Extensions of Mendelian principles, sex linkage, sex limited and sex influenced characters. Extra chromosomal inheritance, Inheritance of mitochondrial and chloroplast genes, maternal inheritance.

- 1. Genetics, A Conceptual Approach, 4th ed., B.A. Pierce, Palgrave Macmillan, 2012.
- 2. Genetics: A molecular perspective, 1st edition W.S. Klug and M.R. Cummings, Benjamin Cummings, 2002.
- 3. Principles of Genetics, 6th ed. (Course Smart), D.P. Snustad and M.J. Simmons, John Wiley and Sons, 2012.

Core-IX

IMMUNOGENETICS

Annexure No;61A

SCAA Dated: 03.07.2017

Unit-I

Historical perspective, lymphoid organs, lymphocytes. Cells of immune system, T and B cell activation and maturation, Haematopoiesis, Haematopoitic stem cells, programmed cell death and necrosis. Immunoglobulins, Class switching, Antigens. The molecular basis of antigen and antibody interactions. Types of immunity and immune responses.

Unit-II

Cytokines, interleukins, complement system, the classical pathway, alternate pathway and the membrane attack pathway. Immunostimulation, Immunosupression and its clinical significance, Immunopotentiation, adjuvants.

Unit-III

MHC gene in man and mouse, Genomic map, gene expression, antigen presentation and processing by MHC class I and class II molecules. Autoimmune diseases, transplantation Immunology, tissue typing and organ transplantation, tumour Immunology, immunobiology of HIV infection.

Unit-IV

Immunization, active and passive. Vaccines: whole organism vaccine, synthetic peptide vaccine, multivalent subunit vaccine, anti idiotype vaccine, designer vaccine, edible vaccine, DNA vaccine, recombinant vector vaccine. Production and applications of monoclonal antibodies, genetically engineered monoclonal antibodies, Abzymes.

Unit-V

Radio-Immuno Assay, ELISA, Immunofluoresence technique, Immune-histochemistry, Karyotyping. Molecular medicines in cancer therapy. Microarray as a tool for detection of human genetic disorders.

- 1. Kuby, J., 2008, Immunology, W.H. Freeman and Co., New York.
- 2. Roit, I.N., Brostaff, J.J. and Male, D.K., 2007, Immunology, C. Mosby, St.Louis.
- 3. Van Dam, Mieras, M.C.E., de Jeu, W.H., de Vries, J., Currell, B.R., James, J.W., Leach, C.K. and Patmore, R.A., 20044, Technological Applications of Immunochemicals, Butterworth, Heineman Ltd., Oxford.
- 4. Lewin, Genes IX, 9th Edition Jonesand Bartlett 2007.
- 5. Principles of Genetics Gardner, Simmons, Snustad 8th Edition 2006.
- 6. An introduction to molecular genetics, Pasternack, 2000.
- 7. Human Molecular genetics, Strachan and Read, 2003.

Core-X

CANCER GENETICS

Annexure No;61A

SCAA Dated: 03.07.2017

Unit-I

History, scope and current scenario of cancer research. Cancer: Types and their prevalence, Carcinoma, Lymphoma and Malignancy. Classification based on origin/organ: breast, colon, lung, prostrate, cervical and oral cancers. Molecular biology of tumor invasion and metastasis.

Unit-II

Cell transformation and tumourigenesis, oncogenes, tumour suppressor genes, DNA repair genes and genetic instability, epigenetic modifications, telomerase activity, centrosome malfunction. Tumour progression: angiogenesis and metastasis.

Unit-III

Oncogenes and human cancers: Role of proto-oncogenes in regulating cell growth and survival, mechanisms of activation of oncogenes, point mutations, fusion genes, gene amplification, chromosome rearrangements, promoter insertion, tumor suppressor genes: Role in cell cycle regulation, Knudson's two, hit hypothesis.

Unit-IV

Non-random chromosome abnormalities in hematologic neoplasms, chronic myeloid leukemia, Chronic lymphocytic leukemia, Acute myeloid leukemia, Acute lymphoblastic leukemia, Myelodysplastic syndromes, Myeloproliferative disorders, Hodgkin's disease, non, Hodgkin's lymphoma, Burkitt's lymphoma.

Unit-V

Familial cancers:Retinoblastoma, Wilm'stumour, Li-Fraumeni syndrome, colorectal cancer and breast cancer. Cancer therapy: At cellular level, at gene level, at protein level. Principles of cancer biomarker and their applications, chemotherapeutics for cancer, Phytotherapy for cancer.

- 1. The Biology of Cancer, R.A. Weinberg, Garland Science, Taylor and Francis Group, 2007.
- 2. Cancer Biology, 3rd ed., R.J.B. King and M.W. Robbins, Pearson Education Ltd., 2006.
- 3. Cancer cytogenetics, chromosomal and molecular genetic aberrations of tumor cells, 3rd ed., S.Heim and F. Mitelman, Wiley, Blackwell Inc., 2009.
- 4. Human cytogenetics: malignancy and acquired abnormalities, a practical approach, 3rd ed., D.E. Rooney, Oxford University Press, 2001.
- 5. ISCN 2013 An International System for Human Cytogenetic Nomenclature (2013), Recommendations of the International Standing Committee on Human Cytogenetic Nomenclature, L.G. Shaffer, J. McGowan, Jordan and M. Schmid, S. Karger, 2013.
- 6. Introduction to the Cellular and Molecular Biology of Cancer,4th ed., M.A. Knowles and P.J. Selby, Oxford University Press, 2005.

CORE XI

NEUROGENETICS AND EPIGENETICS

Annexure No;61A

SCAA Dated: 03.07.2017

UNIT I

Organization of the nervous system, Histology of the nervous tissue: Supporting cells and Neurons. Neurophysiology: Resting membrane potential, Membrane potentials and Signals. Synapse, Postsynaptic potentials and synaptic integration. Neurotransmitters and Receptors. Neuroendocrine system.

UNIT II

Autosomal (recessive and dominant) and X-linked neurological diseases. Neurodegenerative diseases, Unstable mutation (repeat expansion) causing spinocerebellar ataxias, Huntington's disease, Myotonic dystrophy, Fragile-X syndrome. Metabolic defects causing neurological diseases (Tay-Sach's and Gaucher's diseases). Diagnostic procedures for assessing neurogenetic diseases.

UNIT III

Degenerative brain diseases: Cerebrovascular accidents or Stroke and Alzheimer's disease History, Causes, Signs and symptoms, Pathophysiology, Diagnosis, Preventions and Management, Animal models, Therapeutics-Novel therapeutics. Parkinson disease: Causes, Pathophysiology, Diagnosis and Treatment.

UNIT IV

Epigenetics. Mechanisms of DNA methylation, Histone modifications, Chromosomal position effect and gene variegation, Epigenetic control of gene activity, Analysis of gene-specific DNA methylation, Methods of assessing genome-wide DNA methylation. Model organism of epigenetics: Drosophila

UNIT V

Effects of diet and environmental agents on epigenetic processes. Role of epigenetic in immune diseases and disorders.Imprinting disorders in humans. Epigenetic therapy.

- 1. Elaine N. Marieb and KatjaHoehn (2012). Human Anatomy and Physiology. Pearson Publisher.
- 2. Nicholas Wood (2012). Neurogenetics A Guide for Clinicians. Cambridge University Press.
- 3. Thomas T. Warner and Simon R. Hammans (2009). Practical Guide to Neurogenetics. Elsevier Ltd.
- 4. Tollefsbol T (2011). Handbook of Epigenetics. Elsevier Publications
- 5. David C. Allis, Marie-Laure Caparros et al., (2015). Epigenetics. Cold Spring Harbor Laboratory Press.

Annexure No;61A **SCAA Dated: 03.07.2017**

Core-XIIBIOETHICS AND BIOSAFETY

Unit-I

Introduction to bioethics in biotechnology, ethics, bioethics, biotechnology, Positive effects, Negative effects, Ethics in biotechnology, Toxic Soils, Biological Pest Controls. Fast Growing Trees, Fast Growing fish, The Monarch Butterfly Story, Consumer traits, food safety, Environmental concerns and Economic and Social Concerns.

Unit-II

BiosafetyRegulations: National and International Guidelines.Introduction,Regulation framework in various countries,USA,European Union,Canada,Australia,South Africa,Asian Region, International Guidelines.

Unit-III

CPCSEA Guidelines for Laboratory Animal Facility.

Goal, Veterinary care, Animal procurement, Quarantine, Sterilization and separation, Surveillance, diagnosis, treatment and control of disease, Animal care and technical personnel, Personal hygiene, Animal experimentation involving hazardous agent, Multiple surgical procedures on single animal, Duration of experiments, Physical restraint, Physical plant, Physical relationships of animal facilities to laboratories, Functional areas, Physical relationship of animal facilities to laboratories, Functional area, Physical facilities, Environment, Animal husbandry, Activity, Food, Bedding, Water, Sanitation and cleanliness, Assessing the effectiveness of sanitation, Waste disposal, Pest control, Emergency.

Unit-IV

GLP and Bioethics, Introduction, National Good Laboratory Practice (GLP) Programme, The GLP authority functions, Good Laboratory Practices, The Aspiration, Role of a Sponsor, quality standards for Clinical Trials, Clinical Trials worldwide.

Unit-V

Intellectual Property Rights, An introduction, Origin of the Patent Regime, Early patterns Act and History of Indian Patent System: The Present Scenario, Basis of Patentability, Patent Application Procedure in India, Patent Granted Under Convention Agreement, Patent Procedure, Opposition to Grant of Patent, Grant and Sealing, Exclusive Rights, Grant of Exclusive Rights, Special Provision for selling or distribution, Suits relating to infringements, Compulsory License, Termination of Compulsory License, Relief under TRIPS agreement.

References

1. Bioethics, by Shaleesha A. Stanley (2008). Published by Wisdom Educational Service, Chennai.

PRACTICAL-III

Annexure No;61A

SCAA Dated: 03.07.2017

IMMUNOGENETICS

- 1. Serum antigen antibody interaction
- 2. Blotting techniques
- 3. Blood grouping test
- 4. Electrophoretic separation of serum proteins
- 5. Complement mediated haemolysis
- 6. Isolation and enumeration of lymphocytes from human blood
- 7. Determination of lymphocyte viability by Trypan blue dye exclusion test
- 8. Estimation of serum lysozyme and total peroxidase secretion
- 9. Detection of Antibody using ELISA
- 10. Purification of monoclonal antibody

CANCER GENETICS

- 1. Characterization of cancer cell lines
- 2. Detection of oncogene expression adopting Western Blot analysis
- 3. Survey of cancer incidence in India through oncological databases
- 4. MAP kinase activity (Demonstration)
- 5. Characterization of BCl2activity
- 6. Construction of phylogenetic tree using known DNA and Protein sequence
- 7. Sequence similarity analysis for protein and nucleic acid using online bioinformatics tools
- 8. Submission of sequence to NCBI or DDBJ

BIOETHICS AND BIOSAFETY

- 1. Visit to Research institutes holding animal house facility
- 2. Visit to pharmaceutical industry and report submission
- 3. CPCSEA, GLP, IPR, Group discussion, report submission

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ELECTIVE-III

STRESS AND BIOMARKERS

Unit-I

Introduction, definition, sub disciplines, environmental toxicants, routes of entry of xenobiotics.

Unit-II

Toxicity tests, need for conduct of toxicity tests, bioassay, types of acute toxicity tests, terminologies in toxicity tests, chronic toxicity tests.

Unit-III

Bioassay, basic requirements, test organisms, test solutions, test procedures, data analysis joint toxicity.

Unit-IV

Biomarkers: Introduction, background, biomarker selection and development, methods, types, specific and non-specific, organ and tissue specific.

Unit-V

Physiological biomarkers, definition, direct enzyme inhibition, endocrine, blood chemistry energetic, growth rate, histopathology.

- 1. Principles of Ecotoxicology, 3rd edition. C.H. Walker, S.P. Hopkin, R.M. Siblyand D.B. Peakall. Taylor and Francis, New York. 315 pp. 2006.
- 2. Principles of Biochemical Toxicology, 3rd ed. J.A. Timbrell. Taylor and Francis, New York. 394 pp. 2000.
- 3. Huggett, R.J., Kimerle, R.A., Mehrle, P.M.Jr., Bergman, H.L., eds.: Biomarkers: Biochemical, Physiological, and Histological Markers of Anthropogenic Stress. Lewis Publishers, Boca Ratan, FL, 1992.
- 4. McCarthy, J.F., Shugart, L.R., eds.: Biological Markers of Environmental Contaminants. Lewis Publishers, Boca Ratan, 1990.
- 5. Peakall, D.B.: Animal Biomarkers as Pollution Indicators, Ecotoxicological Series 1. Chapman and Hall, London, 1992.

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Elective III STEMCELL BIOLOGY

Unit I

Introduction and Scope of stem cells:definitions, Concepts of stem cells, differentiation, maturation, proliferation, pluripotency, self, maintenance and self, renewal, significations in measuring stem cells, preservation and storage protocols.

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Unit II

Intestinal stem cells, Mammary stem cells, Skeletal muscle stem cell, keratinocyte stem cells of cornea, skin and hair follicles, tumor stem cells, factors influencing proliferation and differentiation of stem cells, Role of hormone in differentiation.

Unit III

Embryonic stem cells, blastocyst, innercell mass, Culturing of ES cells in lab, laboratory tests to identify ES cells, stimulation ES cells for differentiation, properties of ES cells, human ES cells, Monkey and Mouse ES cells.

Unit IV

Identification, Manipulating differentiation pathways, stem cell therapy Vs cell protection, stem cell in cellular assays for screening, stem cell based drug discovery platforms, drug screening and toxicology, stem cell banking.

Unit V

Gene therapy, genetically engineered stem cells, stem cells and Animal cloning, transgenic animals and stem cells, Therapeutic applications, parking disease, Neurological disorder, limb amputation, heart disease, spinal cord injuries, diabetes, burns Matching the stem cell with trans plant recipient, HLA typing Alzheimer's disease, spinal cord injuries tissue engineering application, production of complete organ, kidney, eyes, heart, brain, Stem cell case study

- 1. Embryonic Stem cells by Kursad and Turksen. 2002. Humana Press.
- 2. Stem cell and future of regenerative medicine. By committee on the Biological and Biomedical applications of Stem cell Research.2002.National Academic press.
- 3. Stem cells, Elsevier: CS Potten.

Supportive-III

GENETIC TOXICOLOGY

Annexure No;61A

SCAA Dated: 03.07.2017

Unit I

General Principles of Mutagenicity, Testing and Regulatory Control of Environmental Chemicals. Monitoring of Chemical Mutagens in the environment, Classification of genotoxic agents.

Unit II

Genetic effects of environmental agents and genotoxic agents in various occupations. Various assay procedures to determine genotoxicity, Methods of evaluation of mutagens, Microbial to mammals.

Unit III

Epidemiological approach to evaluate genetic hazards: Occupational Epidemiology and Reproduction, Monitoring for genetic disease in the new born: Transplacentalgenotoxic agents

- 1. Genetic Damage in Man, Caused by Environmental Agents. Berg. K (1979) Academic Press.
- 2. Principles of Genetic Toxicology. Brusick D (1980), Plenum Press.
- 3. Evaluation of Mutagenic and Carcinogenic Potential of Environmental Agents. (1982). Environmental Mutagen Society of India, Bombay.
- 4. Chemical Mutagens, Principles and Methods for their Detection. Hollaender. A, and Serres F.J., Volume 1-10, Plenum Press.
- 5. Cytogenetic Assays of Environmental Agents. Hsu, T.C (1982), Oxford and IBH, New Delhi.
- 6. Handbook of Mutagenicity Test Procedures. Kilbey, B.J., Lehgator, M., Nichols, W and Ramel, C (1984) Elsevier.
- 7. Carcinogens and Mutagens in Environment. Stich, H.F.(1982) CRC Press Inc.
- 8. Environmental Mutagens and Carcinogens. Sugimura, T., Kondo, S and Takebe, H (1982), Alan, R., Liss Inc.,
- 9. Chemical Mutagenesis in Mammals and Man. Voge, F and Rohroborn, G (1970), Springer, Verlag, Berlin.

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Supportive-I GENETICS AND SOCIETY

Unit-I

The history and impact of Genetics in Medicine and Society, Medical Genetics, Early beginnings, Centers for Genetics and Society around the world, an overview. Genes and Pedigrees in Population.

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SCAA Dated: 03.07.2017

Unit-II

Human Genome Project, Beginning and Organization of the HGP, Sequencing of the Human Genome, Promises and Achievements, Ethical, Legal and Social issues of the HGP, Other Genome Projects initiated as a direct consequence of HGP completion, Human Genome Diversity Project.

Unit-III

Advances in Modern Genetics:Some of the areas of concern in Modern Genetics,GM crops, personal DNA data, Gene Therapy, Pharming, Ethical and Legal issues in Medical Genetics.

- 1. Genetics, A Conceptual Approach, 4th ed., B.A. Pierce, Palgrave Macmillan, 2012.
- 2. Emery's Elements of Medical Genetics, 14th ed., P.D. Turnpenny and S. Ellard, Churchill Livingstone, 2012.
- 3. Introduction to Genetics, A Molecular Approach, T. Brown, Garland Science, 2012.
- 4. Genome Duplication, Concepts, Mechanism, Evolution and Disease, M.L. De Pamphilis and S.D. Bell, Garland Science, 2011.
- 5. Human Molecular Genetics, 4th ed., T. Strachan and A. Read, Garland Science, Taylor and Francis Group, 2011.
- 6. A Guide to Genetic Counseling,2nd ed., W.R. Uhlmann, J.L. Schuette and B.M. Yashar, Wiley,Blackwell, 2009.

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Supportive-II

PRINCIPLES OF GENETICS

Annexure No;61A

Unit-I

Mendelian principles: Dominance, segregation, independent assortment, deviation from Mendelian inheritance, Concept of gene, Allele, multiple alleles, pseudoallele, complementation tests.

Unit-II

Extensions of Mendelian principles, Codominance, incomplete dominance, gene interactions, pleiotropy, genomic imprinting, penetrance and expressivity, phenocopy.

Unit-III

Extensions of Mendelian principles, sex limited and sex influenced characters. Extra chromosomal inheritance, Inheritance of mitochondrial and chloroplast genes, maternal inheritance.

- 1. Genetics, A Conceptual Approach, 4th ed., B.A. Pierce, Palgrave Macmillan, 2012.
- 2. Genetics: A molecular perspective, 1st edition W.S. Klug and M.R.Cummings, Benjamin Cummings, 2002.
- 3. Principles of Genetics,6th ed. (CourseSmart), D.P. Snustad and M.J. Simmons, John Wiley and Sons, 2012.

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Supportive-III

GENETIC TOXICOLOGY

Unit I

General Principles of Mutagenicity, Testing and Regulatory Control of Environmental Chemicals. Monitoring of Chemical Mutagens in the environment, Classification of genotoxic agents.

Unit II

Genetic effects of environmental agents and genotoxic agents in various occupations. Various assay procedures to determine genotoxicity, Methods of evaluation of mutagens, Microbial to mammals.

Unit III

Epidemiological approach to evaluate genetic hazards: Occupational **Epidemiology** and Reproduction, Monitoring for genetic disease in the new born: Transplacentalgenotoxic agents

- 1. Genetic Damage in Man, Caused by Environmental Agents. Berg.K (1979) Academic Press.
- 2. Principles of Genetic Toxicology. BrusickD (1980), Plenum Press.
- 3. Evaluation of Mutagenic and Carcinogenic Potential of Environmental Agents. (1982). Environmental Mutagen Society of India, Bombay.
- 4. Chemical Mutagens, Principles and Methods for their Detection. Hollaender. A, and SerresF.J., Volume 1-10, Plenum Press.
- 5. Cytogenetic Assays of Environmental Agents. Hsu, T.C (1982), Oxford and IBH, New Delhi.
- 6. Handbook of Mutagenicity Test Procedures. Kilbey, B.J., Lehgator, M., Nichols, W and Ramel, C (1984) Elsevier.
- 7. Carcinogens and Mutagens in Environment. Stich, H.F. (1982) CRC Press Inc.
- 8. Environmental Mutagens and Carcinogens. Sugimura, T., Kondo, S and Takebe, H (1982), Alan, R., Liss Inc.,
- 9. Chemical Mutagenesis in Mammals and Man. Voge, F and Rohroborn, G (1970), Springer, Verlag, Berlin.

Core-XIII GENETIC COUNSELLING

Unit-I

History of Human Genetics: Pedigrees, gathering family history, Pedigree symbols, Construction of pedigrees, Presentation of molecular genetic data in pedigrees, Pedigree analysis of monogenic traits, autosomal inheritance, dominant, recessive, sex-linked inheritance, X-linked recessive, dominant, Y-linked, sex-limited and sex-influenced traits, Mitochondrial inheritance, MIM number.

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Unit-II

Complications to the basic pedigree patterns: Genomic imprinting and uniparental disomy, Spontaneous mutations, Mosaicism and chimerism, Male lethality, X-inactivation, Consanguinity and its effects in the pedigree pattern, Allele frequency in population, Complex traits, polygenic and multifactorial.

Unit-III

Approaches to analysis of complex traits, Nature *vs* nurture, Role of family and shared environment, Monozygotic and dizygotic twins and adoption studies, Polygenic inheritance of continuous (quantitative) traits, normal growth charts, Dysmorphology, Polygenic inheritance of discontinuous (dichotomous) traits, threshold model, liability and recurrence risk, Genetic susceptibility in complex traits, Alcoholism, cardiovascular disease, diabetes mellitus, obesity and epilepsy, Estimation of genetic components of multifactorial traits, empiric risk, Heritability, coefficient of relationship, Application of Bayes theorem.

Unit-IV

Genetic counselling: Historical overview (Philosophy and ethos) and components of genetic counselling. Indication for and purpose, Information gathering and construction of pedigree Medical genetic evaluation (Basic components of Medical History, Past medical history, social and family history).

Unit-V

Components of genetic counselling: Physical examination (General and Dysmorphology examination, documentation). Legal and ethical considerations, Patterns of inheritance, risk assessment and counselling in common Mendelian and multifactor syndromes Prenatal and pre-implantation screening and diagnosis, indications for prenatal diagnosis. Indications for chromosomal testing. Noninvasive methods (Ultrasound, embryoscopy, MRI, etc.). Invasive methods, Prenatal screening for Down's syndrome (maternal serum) and Neural tube defect, Preimplantation genetic diagnosis, Ethical issues in pre-natal screening and diagnosis.

- 1. Genetics, A Conceptual Approach, 4th ed., B.A. Pierce, Palgrave Macmillan, 2012.
- 2. Emery's Elements of Medical Genetics, 14th ed., P.D. Turnpenny and S. Ellard, 2012.
- 3. Introduction to Genetics, A Molecular Approach, 1st ed., T.A Brown, Garland Science, 2012.
- 4. Human Molecular Genetics, 4th ed., T. Strachan and A. Read, Garland Science, Taylor and Francis Group, 2011.
- 5. A Guide to Genetic Counseling, 2nd ed., W.R. Uhlmann, J.L. Schuette and B.M. Yashar, Wiley, Blackwell, 2009.
- 6. Practical Genetic Counseling-7th ed., P. S. Harper, CRCPress., 2010.
- 7. Genetics Society and Clinical Practice, P.S. Harper and A.J. Clarke, 1st ed., Garland Science, 1997.