ANDHRA PRADESH STATE COUNCIL OF HIGHER EDUCATION
CBCS PATTERN FOR HUMAN GENETICS

STRUCTURE OF B.Sc (HUMAN GENETICS) PROGRAM UNDER CBCS
REVISED SYLLABUS - 2020

YEAR	SEMESTER	PAPER	TITLE	MARKS	CREDITS
1st	I	HGT- I	GENETICS & HUMAN HERIDITY	100	3
		HGP-I	PRACTICAL	50	2
	II	HGT-II	HUMAN GENETICS AND CYTOGENTICS	100	3
		HGP-II	PRACTICAL	50	2
2nd	III	HGT-III	HUMAN MOLECULAR GENETICS	100	3
		HGP-III	PRACTICAL	50	2
	IV	HGT-IV	RECOMBINANT DNA TECHNOLOGY	100	3
		HGP-IV	PRACTICAL	50	2
		HGT-V	STATISTICS AND INFORMATICS IN HUMAN GENETICS	100	3
		HGP-V	PRACTICAL	50	2
3rd	V ELECTIVE PAPERS *ANY ONE PAIR OF ELECTIVE PAPER A OR B OR C		A - PAIR		
		HGT A 1	CLINICAL GENETICS & GENETIC COUNCELING	100	3
		HGP A 1	PRACTICAL	50	2
		HGT A 2	LABORATORY DIAGNOSIS IN GENETICS	100	3
		HGP A 2	PRACTICAL	50	2
			B - PAIR		
		HGT-B 1	HUMAN GENOME PROJECT AND GENOMES	100	3
		HGP B 1	PRACTICAL	50	2
		HGT B 2	MOLECULAR TECHNIQUES IN GENETIC ENGENEERING	100	3
		HGP B 2	PRACTICAL	50	2
			C - PAIR		
		HGT C 1	DEVELOPMENTAL & BEHAVIORAL GENETICS	100	3
		HGP C 1	PRACTICAL	50	2
		HGT C 2	MOLECULAR PATHOLOGY IN HUMAN DISEASES	100	3

		HGP C 2	PRACTICAL	50	2

ANDHRA UNIVERSITY
HUMAN GENETICS
FIRST YEAR - SEMESTER-I
HGT-I GENETICS & HUMAN HEREDITY
(Revised Syllabus-2020)
THEORY

Unit 1: Mendelian Genetics and Extensions

1.1 Physical basis of Heredity. Cell divison –Mitosis & Meiosis

1.2 Mendelian Principles of inheritance – Law of segregation, Law of independent assortment –animal examples; Mendelian inheritance of human traits; Chromosome theory of inheritance.

1.3 Incomplete dominance and codominance; Multiple alleles, Lethal alleles, Pleiotropy, Penetrance and Expressivity – human examples

1.4 Two gene interactions – Epistatic, nonepistatic interactions; Polygenic inheritance in man and other animal organisms.

1.5 Genes and environment –norm of reaction, phenocopies, developmental noise

Unit 2 Sex Linked Inheritance and Sex Determination

2.1 Sex Linked Inheritance - Sex linked inheritance in Drosophila and human; Sex limited and Sex influenced inheritance

2.2 Sex Determination - Sex determination in Drosophila – Genic balance theory

2.3 Sexdetermination in eukaryotes –heterogametic, homogametic, haplodiploidy, role of environmental factors, mosaics

2.4 Sex determination in mammals- and role of human Y chromosome

Unit3Extrachromosomal Inheritance

3.1 Mitochondrial inheritance (petite mutations);Mitochondrial inheritance in man

3.2Maternal inheritance-shell coiling in snail, Ephestia pigmentation

3.3Infective heredity- symbionts in Drosophila,Kappa particles in *Paramecium.*

3.4Epigenetics and genome imprinting in humans

Unit 4: Linkage, crossing over and chromosome mapping

4.1 Linkage and chromosome mapping in eukaryotes – cytological basis of crossingover; recombination frequency,two factor andthree factor crosses; interference and coincidence; Mitotic recombination

4.2 Linkage and chromosome mapping in prokaryotes – bacteria and bacteriophages – transformation, transduction, conjugation; gene mapping in bacteria.

4.3 Genetic definition of gene - Complementation test, intrageniccomplementation,rII locus of phage T4

Unit 5: Variation in Chromosome number and structure

5.1 Specialized chromosomes -Lampbrush chromosomes. Polytene chromosomes: Supernumerary chromosomes.

5.2 Variation in chromosome structure - Deletion, Duplication, Inversion, Translocation, Position effect

5.3Variation in chromosome number - Euploidy and Aneuploidy in man

HGP-I (PRACTICLS)

1. Mendel's laws through seed ratios& Drosophila mutants.
2. Statistical tests in genetic analysis - application of laws of probability (product rule, sum rule, binomial probability); chi square test and its application in the analysis of genetic data.
3. Study of linkage, recombination, 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 anaemia, Xeroderma Pigmentosum, Albinism. Tests for red-green Colour blindness, Widow's peak, Rolling of tongue, Hitchhiker's thumb and Attached ear lobe.
6. Incomplete dominance and gene interaction through seed ratios
7. Blood Typing: ABO groups & Rh factor.
8. Study of aneuploidy: Down's, Klinefelter's and Turner's syndromes.
9. Mitosis &Meiosis through temporary squash preparation.
10. Smear technique to demonstrate sex chromatinin buccal epithelialcells.

Suggested Readings

1. Gardner, E.J., Simmons, M.J., Snustad, D.P. (1991). Principles of Genetics, John Wiley & sons, India.8th edition.
2. Snustad, D.P. and Simmons, M.J. (2010). Principles of Genetics, John Wiley & Sons Inc., India.5th edition.
3. Klug, W.S., Cummings, M.R., Spencer, C.A. (2012). Concepts of Genetics. Benjamin Cummings, U.S.A. 10th edition.
4. Griffiths, A.J.F., Wessler, S.R., Carroll, S.B., Doebley, J. (2010). Introduction to Genetic
Analysis. W. H. Freeman and Co., U.S.A. 10th edition.

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HUMAN GENETICS
BSc FIRST YEAR - SEMESTER-II
HGT-II: HUMAN GENETICS & CYTOGENETICS
(Revised Syllabus-2020)
THEORY

Unit 1 Basic Human Genetics – Monogenic traits

1.1 History of Human Genetics.
1.2 Pedigrees – family history, symbols, construction of pedigree
1.3 Monogenic traits - autosomal inheritance, sex-linked inheritance,sex-limited and sex influenced inheritance, mitochondrial inheritance
1.4 Complications in pedigree patterns – non-penetrance, expressivity,pleiotropy,genetic heterogeneity,genomic imprinting, uniparental disomy, male lethality, X inactivation,consanguinity

Unit 2 Basic Human Genetics – Complex traits

2.1 Approaches to analysis of complex traits - Nature vs nurture, monozygotic and dizygotic twins
2.2 Polygenic inheritance of continuous traits – normal growth charts, dysmorphology
2.3 Polygenic inheritance of discontinuous traits – threshold model, liability and recurrence risk
2.4 Genetic susceptibility in multifactorial disorders – diabetes

Unit 3 Genetic Mapping of Mendelian and Complex characters

3.1Identifying recombinants and non-recombinants in pedigrees
3.2 Genetic and physical map distances – genetic markers, mapping of genetic traits
3.3 Two point mapping – LODscore analysis, multipoint mapping, homozygositymapping
3.4 Genetic mapping of complex traits – difficulties in mapping, allele sharing methods, sib pair analysis, allelic association, linkage disequilibrium mapping

Unit 4 Human Chromosomes

4.1 History of human cytogenetics
4.2Cell division cycle – mitotic process, meiotic process
4.3 Human karyotype – banding, nomenclature of banding
4.4 Nomenclature of aberrant karyotypes

Unit 5 Chromosome anomalies

5.1 Common syndromes due to numerical chromosome changes
5.2 Common syndromes due to structural alterations (translocations, duplications, deletions, microdeletions, fragile sites)
5.3Common chromosome abnormalities in cancer

HGP-II (PRACTICALS)

1. Preparation of pedigree charts for blood group, tongue rolling,ear lobes and colorblindness
2. Genetics of codominant genes – blood groups.
3. Barr Body analysis.
4. Dermatoglyphics
5. Polygenic inheritance – finger print ridge count
6. Preparation of metaphase chromosome spread using peripheral blood sample.
7. Sterilization techniques for leukocyte culture
8. Inoculation and Culture of human leucocytes
9. Preparation of metaphase plates and their staining and analysis
10. Human karyotyping – numericals on chromosome number.
11. Camera-lucida drawing of chromosomes.
12. Micrometric analysis of chromosomes.
13. Study of various abnormal karyotypes observed in humans.
14. G- banding of metaphase plates and their analysis
15. Sister Chromatid exchange analysis from peripheral blood

SUGGESTED READINGS:

1. Human Genetics: Concept and Application by Ricki Lewis 10th Edition
2. Vogel and Motulsky's Human Genetics: Problems and Approaches
3. The Principles of Clinical Cytogenetics by Steven L. Gersen, Martha B. Keagle 3rd edition.
4. Human Cytogenetics: Constitutional Analysis: a Practical Approach by Denise E. Rooney.

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HUMAN GENETICS
BSc SECOND YEAR - SEMESTER-III
HGT-III: HUMAN MOLECULAR GENETICS
(Revised Syllabus-2020)
THEORY

Unit 1 DNA , RNA and Protein Structure

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

2.1 Central dogma of molecular biology

2.2 RNA transcription

2.3 RNA processing

2.4 Translation, post-translation processing

Unit 3 DNA replication, recombination, Mutagenesis and DNA repair

3.1 DNA replication – semiconservative, semi-discontinuous, DNA replication machinery

3.2 DNA recombination

3.3 DNA mutagenesis

3.4 DNA repair

Unit 4 Human Chromosome Organization

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

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 repeats

HGP-III(PRACTICALS)

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 on wt and repair deficient E.coli strains.
8. Preparation of Human chromosome spread and banding

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 Sudbery
5. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf, Johannes Zschocke.

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CBCS PATTERN FOR HUMAN GENETICS

HUMAN GENETICS
BSc SECOND YEAR - SEMESTER-IV
HGT-IV: RECOMBINANT DNA AND STEM CELL TECHNOLOGY
(Revised Syllabus-2020)
THEORY

Unit 1 Cell Based Cloning

1.1 Restriction endonucleases and other enzymes used in manipulating DNA molecules
1.2 Cloning vectors – plasmid vectors, lambda and cosmid vectors, P1 phage vectors, YAC, BAC, M13 or phagemid vectors, expression vectors
1.3 Introducing recombinant DNA into recipient cells
1.4 DNA libraries -generation of genomic and cDNA libraries; chromosomal DNA libraries

Unit 2 Cloning Human disease genes

2.1 Cloning human disease genes- functional candidate gene cloning, positional candidate gene cloning
2.2 Detection of mutations in human genes –SSCP analysis, DGGE, chemical mismatch cleavage
2.3 Detection of mutation in human gene – DNA sequencing, heteroduplex analysis, protein truncation

Unit 3 Applications of rDNA technology

3.1 DNA fingerprinting – use of mini-satellites for DNA fingerprinting, single locus probes, STRs
3.2 Genetic testing – prenatal testing, neonatal screening, diagnosis of genetic disease in children after birth, pre-symptomatic testing.
3.3 In vivo, in vitro gene therapy; vehicles for gene therapy; gene therapy for heritable and non- heritable genetic diseases.

Unit 4 Biology of stem cells

4.1 Historical perspectives, concept of stem cells
4.2 Cellular and molecular features of stem cells
4.3 Embryonic stem cells and germ stem cells
4.4 Fetal adult stem cells and cancer stem cells

Unit 5 Applications

5.1Medical need for stem cells and preservation of stem cells
5.2.Genetically engineered stem cells for gene therapy

5.3 Stem cell therapy – neurodegenerative disorders,cardiovascular disorders,metabolic disorders, hematopoietic disorders, organ disorders, autoimmune disorders,reproductive failures

HGP-IV(PRACTICALS)

1. Isolation of plasmid DNA from *E. coli* cells.
2. Digestion of plasmid DNA with restriction enzymes.
3. Estimation of size of a DNA fragment after electrophoresis using DNA markers
4. Construction of restriction digestion maps from data provided
5. Recovery of DNA from low-melting temperature agarose gel
6. Preparation of competent cells of E.coli
7. Transformation of competent E.coli cells with plasmid DNA
8. Amplification of a DNA fragment by PCR.
7. Complementation of beta−galactosidase for Blue and White selection.
8. Southern blotting
9. Western blotting.
10. Culturing cells – aseptic techniques, media
11. Subculturing and cell lines
12. Cryopreservation

Suggested Readings

1. Gene Cloning and DNA Analysis (2010) 6th ed., Brown, T.A., Wiley-Blackwell publishing(Oxford, UK), ISBN: 978-1-4051-8173-0.
2. Principles of Gene Manipulation and Genomics (2006) 7th ed., Primrose, S.B., and Twyman, R. M., Blackwell publishing (Oxford, UK) ISBN:13: 978-1-4051-3544-3.
3. Molecular Biotechnology: Principles and Applications of Recombinant DNA (2010) 4th ed., Glick B.R., Pasternak, J.J. and Patten, C.L., ASM Press (Washington DC), ISBN: 978-1-55581-498-4 (HC).
4. Human Molecular Genetics by Sudbery.

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BSc THIRD YEAR – SEMETER-V
HGT-V: STATISTICS AND INFORMATICS IN HUMAN GENETICS
THEORY

Unit 1 Descriptive Statistics

1.1 Methods of presentation and interpretation of data – frequency distribution, graphical representation of data, histogram, frequency polygon, frequency curve.

1.2 Measures of Central tendency – mean, median, mode

1.3 Measures of Dispersion - standard deviation, variance, coefficient of variation.

Unit 2 Elementary Probability

2.1 Mathematical definition of probability of an event, Use of permutations and combinations in calculations of Probability

2.2 Conditional probability, Additive and Multiplication law of Probability, Random Variables, Mathematical expectation and variances

2.3 Probability Distributions: Binomial, Poisson and normal distributions.

2.4 Bayes theorem

Unit 3 Correlation analysis, test of significance and ANOVA

3.1 Correlation and regressionanalysis— Relationship between variables

3.2 Test of significance – statistical and scientific hypothesis, null and alternative hypothesis, procedure of hypothesis testing,

3.3 Test of significance – student's t test, chi-square test, F test

3.4 ANOVA – general idea of one way and two way analysis

Unit 4 Computers, operating systems and Internet

4.1 Principles of computer operations –basic computer architecture, hardware architecture

4.2 Principles of computer operations – software architecture, operating systems, Programming languages –traditional and scripting languages, Java, markup languages, application programs

4.3 Communication and Networks – network architecture, standards for exchange of information, internet services - email, WWW search engines

Unit 5 Bioinformatics

5.1 History of Bioinformatics

5.2 Databases and search tools – NCBI, EBI, GenomeNet; Databasemining tools – BLAST

5.3 Database archives – nucleic acid sequence databases, genome databases and genome browsers, protein sequence databases, databases of protein families, databases of structures, expression and proteomic databases, bibliographic databases

5.4 Gateways to archives –ENTREZ, PIR, ExPASy

HGP-V(PRACTICALS)

1. Frequency distribution
2. Various types of graphs
3. Mean, Median, Mode
4. Standard deviation, variance and coefficient of variation
5. Testing of hypotheses regarding population mean
6. Testing of hypotheses about the difference between population means
7. Chi-square test
8. Testing of Correlation Coefficient
9. Fitting of simple linear regression
10. One-way ANOVA&Two-way ANOVA
11. Internet basics
12. Sequence retrieval (protein and gene) from NCBI, Structure download (protein and DNA) from PDB
13. Molecular file formats - FASTA, GenBank, Genpept, GCG, CLUSTAL, Swiss-Prot,FIR

Suggested Readings

1. Fowler, J., Cohen, L. and Jarvis, P. (1998). Practical Statistics for Field Biology. John Wiley and Sons, 2nd ed. .
2. Bland, M. (2006). An Introduction to Medical Statistics. Oxford University Press, 3rd ed.
3. Finney, D.J. (1980). Statistics for Biologists.Chapman and Hall Ltd.
4. Wayne, W, Daniel (1999). Biostatistics: A Foundation for Analysis in Health Sciences. John Wiley and Sons, 7th ed.

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BSc THIRD YEAR – SEMESTER-V
THERE WILL BE THREE PAIRS OF EACH DOMAIN OF CORE COURSE.

STUDENT HAS TO CHOOSE ONE PAIR FROM EACH DOMAIN.

ELECTIVE THEORY
A - PAIR
HGT A1: CLINICAL GENETICS & GENETIC COUNCELING

UNIT-1 GENETIC DISORDERS I

- 1.1Monogenic diseases – Cystic fibrosis, Tay-Sachs syndrome, Marfan syndrome
  1.2Inborn errors of metabolism – Phenylketonuria, Maple syrup urine syndrome,galactosemia
  1.3 Genome imprinting syndromes –Prader Willi and Angelman syndrome

UNIT-2 GENETIC DISORDERS II

2.1 Genomic syndromes – Neurofibromatosis I
2.2Neurogenetic disorders – Charcot Marie Tooth syndrome, spinal muscular atrophy, alzhemiers diseases, syndromes due to triplet nucleotide expansion
2.3 Muscle genetic disorders – dystrophies, myotonias, myopathies

UNIT-3 GENETIC DISORDERS III

3.1 Genetic Disorders of Haemopoitic systems- sickle cell anaemia, thalassemias, hemophilia
3.2 Genetic disorders of eye – colorblindness, retinitis pigmentosa, glaucoma
3.3. Complex polygenic syndromes – artherosclerosis, diabetes mellitus
3.4 Mitochondrial syndromes

UNIT-4 GENETIC COUNCELLING

4.1 Role of genetic counseling
4.2 Causes and factors for seeking counselling
4.3 Dysmorphology
4.4 Prenatal and preimplantation diagnosis

UNIT-5 PRACTICAL GENETIC COUNCELING

5.1 Process of genetic counselling - Constructing a family tree, diagnostic information, risks and odds, estimation of risks
5.2 Genetic counselling in Mendelian disorders
5.3 Genetic counselling in Non-Mendelian disorders
5.4 Ethical and legal issues in genetic counselling

HGP A1: CLINICAL GENETICS & GENETIC COUNCELING

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 tumor 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

SUGGESTED READINGS

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

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BSc THIRD YEAR – SEMESTER-V
HGT A 2 : LABORATORY DIAGNOSIS IN GENETICS

UNIT-1 BASICS OF CELL CULTURE & INSTRUMENTATION

1.1 Techniques of cell cultures (short term lymphocyte, primary and secondary cell cultures, maintenance of cell lines)
1.2Spectrophotometer,centrifugation
1.3 ELISA, radioactivity detection
1.4 Mass spectrometry
1.5High performance liquid chromatography

UNIT-2 TECHNIQUES IN CHROMOSOME ANALYSIS

2.1 Techniques of chromosome analysis - (a) Chromosome preparation from cultured lymphocytes, cell lines and solid tumors (b) Karyotyping, C-,G-banding and fluorescence banding, nomenclatures of bandings (c) *In-situ* hybridization techniques (d) Meiotic chromosomes in mouse testis
2.2Chromosomal anomalies and disorders - Numerical (polyploidy, aneuploidy, autosomal, sex- chromosomal) - Structural (deletion, duplication, translocation, inversion, isochromosome, ring chromosome) - Chromosomal abnormalities in cancer
2.3Microscopy -Metaphase chromosome preparations from bone marrow of mouse, rat, human,Sex chromatin (buccal mucosa, hair bud),Comet assay, Micronuclei assay, Chromosome preparation from chorionic villi, Sister Chromatid Exchange (SCE)

UNIT-3 GENETIC DISORDERS & MOLECULAR TECHNIQUES FOR DISEASE IDENTIFICATION

3.1 Genetic Disorders - Classification of genetic disorders, Single gene Disorders (Cystic Fibrosis, Marfan's syndrome), Multifactorial disorders (Diabetes, Atherosclerosis, Schizophrenia)
3.2 Molecular Techniques PCR-RFLP, Multiplex-PCR, SSCP,MALDI-TOF
3.3. Disease identification and Genetic tests for following disorders: (a) Thalassemia, Fanconi , Sickle Cell anaemia, Fragile-X syndrome, Alzheimer's disease (b) Duchenne Muscular Dystrophy/Becker's Muscular Dystrophy, Huntington's disease (c) Allelic susceptibility test for multifactorial disorders (Neural Tube Defect, Cleft Lip and Palate, Cardio Vascular Disorder, Male infertility)

UNIT-4 BIOCHEMICAL GENETIC DIAGNOSIS

4.1 Biochemical tests: sugar, albumin, urea, protein, globulin, vitamin ;
4.2 Biochemistry and diagnostic tests of following diseases -Duchenne Muscular Dystrophy (DMD) (Creatine phosphokinase-CPK),Phenylketonuria-PKU (phenylketone) ,G6PD deficiency syndrome (G6PD) ,Endocrine disorders related to thyroid and reproduction (TSH, T3, T4, Estradiol, Testosterone, LH, FSH)

UNIT-5 GENETIC COUNCELLING AND PRENATAL DIAGNOSIS

5.1 Causes and factors for seeking counselling
5.2Dysmorphology
5.3 Ethical and legal issues in genetic counselling
5.4 Prenatal and preimplantation diagnosis

HGP A 2 : LABORATORY DIAGNOSIS IN GENETICS

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.Fluorescence *in-situ* Hybridization (FISH)
6.Meiosis in mouse testis
7.Sex chromatin (buccal mucosa, hair bud)
8.Comet assay
9.Micronuclei assay
10.Chromosome preparation from chorionic villi, stem cells, cell line
11.Sister Chromatid Exchange (SCE)
12.Molecular markers for tumor detection
13.Bcr-abl (RT-PCR)
14.Genetic counseling (pedigree analysis in disease conditions, risk calculation)
15.Prenatal diagnosis of Thalassemia
16.Y-chromosome micro deletion
17.Biochemical tests for sugar, albumin, Creatine phosphokinase-CPK, glucose 6 phosphate dehydrogenase-G6PD

Suggested Reading

1. Primrose, SB and Twyan RM. *Principles of gene manipulation and genomics*. 7th edition.Blackwell Science, 2006.

2. Watson, Myers and Caudy. *Recombinant DNA: Genes and Genomes-A short course*. 3rd edition. 2006. Freeman W.H. and Company.

3. Fundamentals of Molecular Diagnositics by David E. Bruns, Edward R. Ashwood, Carl A Burti4. Human Genetics: From Molecules to Medicine by Christian Patrick Schaaf,JohannesZschocke, Lorraine Potocki

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BSc THIRD YEAR – SEMESTER-V
B - PAIR
HGT- B 1 : HUMAN GENOME PROJECT AND GENOMES

Unit 1 Genome Organization and Study

1.1 Genome – general features, features of eukaryotic nuclear genomes

1.2 Genomes, transcriptomes and proteomes

1.3 Genome diversity – significance of genomes – bacteria, yeast,Caenorhabditis,Homo sapiens, Arabidopsis.

Unit 2 Mapping Genomes

2.1 Genetic mapping – pedigree analysis, DNA markers – RFLPs, SSLPs, SNPs

2.2 Physical mapping – restriction mapping, FISH, radiation hybrid mapping, STS mapping

2.3 Sequencing genome- assembly of contiguous DNA sequence, shotgun method, clonecontig method, whole-genome shotgun sequencing

Unit 3 Genome Projects

3.1 Human genome project, HapMap Project, 1000 genome project, ENCODE project

3.2 Other genome projects.

3.3 Applications and proposed benefits of HGP –ELSI.

Unit 4 Understanding Genome sequence

4.1 Locating the genes in a genome sequence
4.2 Determining the functions of individual genes
4.3. Transcriptome – microarrays
4.4 Proteome – protein profiling

Unit 5 Molecularphylogenetics

5.1 Phenetics and cladistics
5.2 Reconstruction of DNA based phylogenetic tree
5.3 Applications of molecular phylogenetics – evolutionary relationship between humans and primates; origin of AIDS; human pre history.

HGP- B 1 : HUMAN GENOME PROJECT AND GENOMES

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

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, Diane Drobnis 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 Richard Twyman.

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BSc THIRD YEAR – SEMESTER-V

HGT-B 2 : MOLECULAR TECHNIQUES IN GENETIC ENGENEERING

UNIT-1 NUCLEIC ACID ISOLATION AND AGAROSE GEL ELECTROPHORESIS

(9 hours)

Conventional and kit method for isolation of nucleic acids-Plasmid DNA-Genomic DNA from Bacterial cells,Plant cells,animal cells-RNA isolation and m-RNA purification –Agarose purification-Agarose gel electrophoresis-Staining techniques –Pulse feild gel electrophoresis

UNIT-2 PCR TECHNIQUES (9 hours)

Principle of Polymerase Chain Reaction (PCR)-Components of PCR reaction and optimization of PCR –Gene specific primer- Inverse PCR, Hot-start PCR ,Loop mediated PCR – Reverse transcription PCR and Real time PCR.Chemistry of primer synthesis

UNIT-3 HYBRIDIZATION METHODS (9 hours)

Probes –Labelling of probes-Radio active and non-radio active probes-Detection techniques,Southern hybridization,Northern hybridization,Western blotting

UNIT-4 DNA SEQUENCING AND GENE SYNTHESIS (9 hours)

Sangers's method of DNA sequencing – Manual and automated methods. Pyroseuencing-massive parallel 454-sequencing,illumina sequencing,SOLID sequencing,single molecule sequencing

UNIT-5 PROTEIN TECHNIQUES (9 hours)

Electrophoresis of protein –native and denaturing conditions,capillary and gel electrophoresis,3D gel electrophoresis,ELISA ,yeast hybrid system-one hybrid system,phage display

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HGP-B 2 : MOLECULAR TECHNIQUES IN GENETIC ENGENEERING

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
9. SDS-Gel electrophoresis
10. Southern blotting
11. Northern blotting
12. Western blotting

REFERENCES

1.Fredrick M.Ausubel.Roger Brent,Robert E Kingstone,David D. Moore,Seidman J. G,John A.Smith and Kevin Struhl, "Current Protocols in Molecular Biology",John Wiley & Son,Inc.2003.

2.Daniel C.Liebler "Introduction to Proteomics",Human Press,2002.

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HUMAN GENETICS
BSc THIRD YEAR – SEMESTER-V
C - PAIR
HGT C 1: DEVELOPMENTAL AND BEHAVIORAL GENETICS

UNIT-1: Germ Cells and Fertilization

Germ Cells

Spermatogensis

Oogenesis

Fertilization and Gastrulation

UNIT-2: Molecular Aspects of Development

Maternal effect gene

Gap gene

Pair rule gene

Segment polarity genes

Homeotic genes

UNIT-3: Genetics of Embryonic Development in Drosphila

Overview of Drosphila development

Zygotic genes and segment formation

UNIT- 4: Flower Development in Arabidopsis

Development,Role of Homeotic Selector Gene

UNIT-5:GENETIC CONTROL OF BEHAVIOUR

Introduction, Behaviour in Invertebrates,Honeybee, Drosophila – Genetic basis of alcoholism, genetic basis for sexual orientation.Courtship behaviour in various animals.

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HGP C 1: DEVELOPMENTAL AND BEHAVIORAL GENETICS

1.Study of development in chick embryo

2.Dissection of imaginal disc in Drosophila larvae

3.life cycle of drosophila,husbandary and handling.

4.Role of SHH signaling in chick development

5.Observation of living and plastic embedded chick embryos

6.The maternal effect gene in drosophila

REFERENCES

The cell – Bruce Alberts

Emery's Elements of Medical Genetics- Robert. F. Mueller, Ian. D. Young.

Principles of Development - Wolpert

Principles of Genetics – Snustad, Simmons, Jenkins.

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HUMAN GENETICS
BSc THIRD YEAR – SEMESTER-V

HGT C 2 : MOLECULAR PATHOLOGY IN HUMAN DISEASES

Unit 1 Human diseases I

1.1 Etiology, pathology and symptoms of genetically inherited diseases – PKU, alkaptonuria, galactosemia, Von Gierke disease, LeschNyhan syndrome, Gout, sickle cell anaemia, beta thalassemia, diabetes

1.2 Mode of infection, symptoms and epidemiology of disease causes by viruses (HIV, Hepatitis B, Rabies, HSV-1)

1.3 Mode of infection, symptoms and epidemiology of disease caused by bacteria – typhoid, syphilis, TB

Unit 2 Human diseases II

2.1 Mode of infection, symptoms and epidemiology of disease caused by fungi – aspergillosis,histoplasmosis.

2.2 Mode of infection, symptoms and epidemiology caused by protozoa –malaria, amoebiasis.

2.3 Cancer genetics - tumor suppressor genes, oncogenes, Molecular basis of oncogenesis

Unit 3 Basic Instrumentation principles and techniques

3.1 Principles of electrophoresis and immunoblotting

3.2 Principles of DNA sequencing and methods of genotyping and mutation analysis

3.3 Principles and applications of PCR

3.4 In situ hybridization techniques – ISH, FISH

Unit 4 Genetic testing for hereditary disorders

4.1 Genetic testing for thalassemia

4.2 Genetic testing for familial colorectal cancer

4.3 Genetic testing for familial breast and ovarian cancer

4.4 EGFR mutation in lung cancer, HER2 amplification in breast cancer, FISH test for early bladder cancer detection,KRAS mutation detection for colorectal cancer

Unit 5 Molecular diagnosis of infectious diseases

5.1 Principles of HPV testing and methods of genotyping

5.2 Hepatitis B virus infection – testing for viral load and HBV DNA mutants detection

5.3 Molecular techniques -NestedPCR, Real Time PCRfor different clinical applications

HGP VIII-C 2 : Molecular Pathology in Human Diseases

1. Preventing contamination.
2. Extract and assess the purity of DNA.
3. Agarose gel electrophoresis
4. Set up PCR.
5. Evaluate Southern blot data
6. Analyze PCR product using agarose gel electrophoresis and interpret results
7. Demonstration of karyotyping
8. Isolate cellular RNA, purify mRNA
9. Set up RT-PCR using commercial kit
10. Analyze RT-PCR results by agarose gel.

SUGGESTED READING

1. Basic Concepts of Molecular Pathology Series: Molecular Pathology Library,Vol. 2Cagle, Philip T. Allen, Timothy C. (Eds.)Springer 2009

2. Molecular Pathology: The Molecular Basis of Human Disease; William B. Coleman, Gregory J. Tsongalis (Eds.);Academic Press;

3. Genomics and Personalized Medicine Huntington F. Willard, Geoffry S. Ginsburg; Elsevier 2009

4. Medical Genetics, 4th Edition;Lynn B. Jorde, John C. Carey, and Michael J. Bamshad,Mosby

5. DNA from A to Z & Back Again; Carol A. Holland and Daniel H. Farkas; AACC Press 2008

6. Molecular Genetic Pathology, 1st ed.; Liang Cheng and David Zhang; Humana Press 2008