

## HUMAN GENETICS SYLLABUS FOR CREDIT BASED SEMESTER SYSTEM (BASIC)

No.	Course Code	Name of the course	Hours	Credits
1	HG – 401	Cell Biology	3+1	04
2	HG - 402	Techniques in Cytogenetics and Tissue Culture	3+1	04
3	HG – 403	Genetics	3+1	04
4	HG – 404	Biological Chemistry	3+1	04
5	HG – 405PR	Practical – 1	6	04
6	HG – 406PR	Practical – 2	6	04
		<b>Total</b>	<b>28</b>	<b>24</b>
1	HG – 407	Molecular Biology	3+1	04
2	HG - 408	Bioinformatics and Biostatistics	3+1	04
3	HG – 409	Immunotechnology	3+1	04
4	HG – 410	Genetic Counseling and Management	3+1	04
5	HG – 411PR	Practical – 3	6	04
6	HG – 412PR	Practical – 4	6	04
		<b>Total</b>	<b>28</b>	<b>24</b>
1	HG – 501	Genetic Toxicology and Cancer Genetics	3+1	04
2	HG - 502	Genetic Engineering	3+1	04
3	HG – 503	Clinical and Molecular Genetics	3+1	04
4	HG – 504	Stem Cell Biology and Nanosciences	3+1	04
5	HG – 505PR	Practical – 5	6	04
6	HG – 506PR	Practical – 6	6	04
		<b>Total</b>	<b>28</b>	<b>24</b>
1	HG – 507S	Seminars / Industrial Visits	4	04
2	HG - 508A	Assignments / Group Discussions	4	04
3	HG – 509PT	Dissertation and Viva	20	16
		<b>Total</b>	<b>28</b>	<b>24</b>
		<b>Grand Total</b>	<b>112</b>	<b>96</b>

## **HUMAN GENETICS SEMESTER – I**

### **HG-401 : CELL BIOLOGY**

#### **UNIT – 1:**

General Principles of Microscopy, Light microscopy, Electron microscopy, Phase contrast, Fluorescence, Polarized and confocal microscopy, special techniques in electron microscopy, Flow cytometry.

#### **UNIT – II:**

From molecules to first cell, from prokaryotes to eukaryotes, from unicellular to multicellular organisms, cell colony, cell cohesion, internal environment or homeostasis of cells, Plasma membrane, Mitochondria, Cytoskeleton.

#### **UNIT – III**

Golgi complex, Endoplasmic reticulum, Ribosomes, Lysosomes and diseases, Peroxisomes, Nucleus and nucleolus.

#### **UNIT – IV**

Cell cycle, Apoptosis, cell-cell communication, receptor ligand interaction, signal transduction, role of second messengers and G-proteins, Ion calcium channels, extra and intracellular interactions, cell aggregations.

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### **HG-402:TECHNIQUES IN CYTOGENETICS AND TISSUE CULTURE**

#### **UNIT – I**

History and development of Human Cytogenetics, Centromere and kinetochore, telomere and its maintenance, Chromosome banding techniques, sister chromatid exchanges, FISH

#### **UNIT – II**

Idiogram, Karyotyping, Numerical and structural aberrations, Cytogenetic nomenclature, Chromosomal disorders . Common syndromes due to numerical changes and structural disorders. Genetics of fetal wastage.

#### **UNIT – III**

Basics of tissue culture, culture environment, factors affecting tissue culture, contamination and its remedies, cryopreservation, PBLC

#### **UNIT – IV**

Culture initiation, maintenance, suspension and monolayer cultures, primary and secondary cultures, cell lines, organ culture.

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## **HG – 403: GENETICS**

### **UNIT – I**

Structure of DNA. Histone proteins, Nucleosome, Solenoid structure, Molecular organization of DNA in chromosomes. Heterochromatin and Euchromatin. Human mitochondrial DNA. DNA replication – nuclear and mitochondrial, Transcription, Translation, control of gene expression – Eukaryotic.

### **UNIT – II**

Introduction to Genetics, Mendel and his experiments: law of segregation, Law of independent assortment, Applications of laws of probability (product rule, sum rule), Allelic variation and gene function- Dominance-Recessive relationships.

### **UNIT – III**

Penetrance and expressivity, phenocopy, Gene interactions and modifying genes, Pleiotropy, Mechanism of sex determination, Sex linked inheritance, Linkage and crossing over.

### **UNIT – IV**

Concepts of genome organization - split genes, overlapping genes, unique sequences, repetitive sequences, pseudogenes, Transposons, conserved genes. Population Genetics

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## **HG – 404: BIOLOGICAL CHEMISTRY**

### **UNIT – I**

Essential and non-essential amino acids, small peptides and their biomedical importance. Proteins: structure, conformation, function relationship, protein degradation, metabolism and genetic disorders. Enzymes: General properties, kinetics, mechanism of action, regulation of enzyme activity, enzyme inhibition.

### **UNIT – II**

Carbohydrates: Structure and conformations, Glycolysis, Krebs cycle, Gluconeogenesis, Glycogenesis and glycogenolysis, metabolism and its genetic disorders.

### **UNIT – III**

Lipids: Fatty acids, synthesis and oxidation of fatty acids, ketogenesis, Metabolism of cholesterol, Lipoproteins: role in lipid transport and storage, Metabolism and its genetic disorders.

### **UNIT – IV**

Hormones: Characteristics, Mechanism of action of peptide and steroid hormones, hormone receptors, Metabolism and its genetic disorders.

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**HG-405PR : Practical I : Based on topics covered in HG 401 and 402**

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**HG-406PR : Practical II : Based on topics covered in HG 403 and 404**

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**Reference Books for HG Semester I:**

<b>Sr. No.</b>	<b>Book Title</b>	<b>Author</b>	<b>Publisher</b>	<b>Year</b>
1	Cell and Molecular Biology	Karp	John Wiley	2002
2	Cell Biology	Pollard & Earnshaw	Saunders	2002
3	Cell Biology - A Laboratory Manual (Vol. I - III)	Celis	Academic Press, USA	1994
4	Molecular Biology of the Cell	Alberts et al.	Garland Science, USA	2002
5	Molecular Cell Biology	Lodish et al.	Scientific American Books Inc., USA	1998
6	Techniques in microscopy and Cell Biology	Sharma	Tata Mac Grow Hill Publishing Company Ltd., New Delhi	1991
7	The Cell : A Molecular Approach	Cooper & Hausman	A.S. M. Press, Washington D.C.	2006
8	Theory and Practice of Histological Techniques	Bancroft & Stevens	Churchill Livingstone, New York	1990
9	Biochemistry & Mol. Biology	Elliott & Elliott	Oxford Press	2005
10	Biochemistry Vol. 1 & 2	Voet & Voet	Wiley	2004
11	Essential Genetics	Hartl et al.,	Wiley & Sons	2002
12	Essentials of Genetics	Klug & Cummings	Prentice Hall	2003
13	Fundamental Genetics	Ringo	Cambridge Univ. Press	2006
14	Fundamental of Biochemistry Life at Molecular Level	Voett et al.	Willey Asia Student Ed.	2006
15	Genetics	Strickberger	Prentice Hall of India Pvt. Ltd.	1999
16	Genetics	Russell	The Benjamin / Cummings Publishing Inc.	2002
17	Genetics : A Conceptual Approach	Benjamin	Freeman	2003
18	Harpers Illustrated Biochemistry	Murray et al.	Prentice Hall	2003
19	Lehninger's Principles of Biochemistry	Nelson and Cox	W.H. Freeman	2005
20	Medical Biochemistry	Baynes and Dominiczak	Mosby	2005
21	Modern Genetic Analysis	Griffith's et al.	Freeman	1999
22	Principles of Biochemistry	Zubay et al.	Brown W. M. C. Co.	1995
23	The Eukaryotic chromosome	Bostok and sumner	Elsevier	1980
24	Chromosome structure and function	Risley	Reinhold	1985
25	Human Chromosomes. Manual of Basic techniques	Verma and Babu	Pergamon	1989
26	Chromosome aberration: Basic and applied aspects	Obe and Natarajan	Springer	1990
27	Chromosome	Sumner	Blackwell	1990

## **HUMAN GENETICS SEMESTER – II**

### **HG-407: MOLECULAR BIOLOGY**

#### **UNIT - I:**

Enzymes used in DNA technology, Isolation and purification of DNA (genomic and plasmid) and RNA, Electrophoresis: Agarose, PAGE, Pulse-field electrophoresis, capillary electrophoresis, 2D electrophoresis.

#### **UNIT – II:**

Polymerase chain reaction and its applications, DNA sequencing, Oligonucleotide synthesis, ELISA. Blotting techniques- Southern, northern and western.

#### **UNIT – III**

Cloning vectors, Cloning techniques, Recombinant DNA technology applications with examples. Centrifugation: types of rotors, clinical, high speed and ultracentrifuges

#### **UNIT – IV**

Colorimetry and spectrophotometry: Beer-Lambert law, absorption spectrum, Fluorescence spectrum, Introduction to mass spectrophotometry, Chromatography: Paper, Thin layer, Column, Ion-exchange, Gel-permeation, Affinity and HPLC.

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### **HG-408: BIOINFORMATICS AND BIostatISTICS**

#### **UNIT – I**

Basics of computer, uses of computer, internet searches, operating system, virtual libraries. Basic concepts of biological databases.

#### **UNIT – II**

Compliances of databases, Gene and protein sequence databanks, Access to sequence databases on the internet, storing sequences in computers, Simple pattern searching in sequences, Uploading the data to existing databases. Web based resources and databases. Primer designing.

#### **UNIT – III**

Data and data presentation, concepts of population, sample and parameter and statistical analysis, measures of central tendency: Mean, mode and median, variants, variance and variation, Standard deviation and standard error, Basic of Probability theory and theory of distribution, Normal distribution and normal curve.

#### **UNIT – IV**

Coefficient correlation and regression, Null hypothesis and alternative hypothesis, tests of significance, ‘t’ test, chi square and applications, testing for genetic segregation, linkage and heterogeneity, ANOVA- one way and two way analysis, Non-parametric test.

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## **HG-409: IMMUNOTECHNOLOGY**

### **UNIT – I**

Introduction to immune system, cells and organ of immune system, clonal selection, Immunogens, Antigen presentation.

### **UNIT - II**

Ag-Ab interaction, immunoglobulins, Generation and regulation of Immune response, TCRs tolerance, Complement system. , HLA complex,

### **UNIT – III**

Disorders of Human Immune system, Primary and secondary immuno-deficiencies, Autoimmune disorders and role of MHC in disease susceptibility, Hypersensitive reactions.

### **UNIT – IV**

Cytokine related diseases, Immune response to infectious diseases and malignancy, Concept of immunotherapy, vaccines, transplantation immunology.

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## **HG- 410: GENETIC COUNSELING AND MANAGEMENT**

### **UNIT – I**

Patterns of inheritance: classical and non classical, Overview of genetic counseling, components of genetic counseling, information gathering and construction of pedigrees and their interpretation.

### **UNIT – II**

Ethics, psychological counseling, Patient education, Risk communication and decision making, Understanding genetic testing, Medical documentation.

### **UNIT -III**

Risk assessment and counseling in common Mendelian and multifactorial syndromes, Management of genetic disorders, Genetic case pathway & preventive management guidelines.

### **UNIT - IV**

Indications of prenatal diagnosis, indications for chromosomal testing, Techniques - Non-invasive methods & Invasive methods, Amniocentesis, Chronic Villus Sampling, Ultrasound, Fetoscopy, Maternal blood screening, PGD, Ethical and legal issues in adopting prenatal diagnosis.

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**HG-411PR : Practical III : Based on topics covered in HG 407 and 408**

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**HG-412PR : Practical IV : Based on topics covered in HG 409 and 410**

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**Reference Books for HG Semester II:**

Sr. No.	Book Title	Author	Publisher	Year
1	Human cyrogenetics: A practical approach (Vol. I & II)	Rooney and Czepulkowski	IRL Press	1992
2	Emery's Elements of medical genetics	Mueller and Young	Churchill	1998
3	An introduction to molecular human genetics	Pasternak	Fritzgerald	2000
4	Practical biochemistry. Principle and techniques	Wilson & Walker	Cambridge	2000
5	An introduction to genetic analysis	Grifith's et al.	Freeman	2004
6	Gene VIII	Lewin	Pearson	2004
7	Bioinformatics. Methods and protocols.	Misner & Krawetz	Humana press	2000
8	Immunology	Roitt et al.	Mosby	2000
9	Recombinant DNA and Biotechnology	Krenzer and Massey	ASM press	2000
10	Cellular and Molecular Immunology	Abbas et al.	Saunders	2001
11	Essential Molecular biology (Volume I & II)	Brown	Oxford University Press	2001
12	Immunobiology	Janeway et al.	Churchill	2001
13	Molecular Cloning	Sambrook	Coldspring Harbour	2001
14	Short Protocols in Molecular Biology	Ausubel	Willey	2002
15	Immunology	Kuby	Freeman	2003
16	Recent Advances in Bioinformatics	Khan & Kanum	Ukraaz Publication	2001
17	Statical Method in Bioassay	Finney	Grrifin	1991
18	Statical Techniques in Qualitative Genetics	Jain	TATA	1982
19	Applied Statistics	Mukhopadhyay	Books and Allied	2000
20	Fundamentals of Statistics	Gupta	Himalaya	2005
21	Fundamentals of Biostatistics: Practical Approach	Datta	Kanishka	2002

## **HUMAN GENETICS SEMESTER – III**

### **HG-501 : GENETIC TOXICOLOGY AND CANCER GENETICS**

#### **UNIT 1:**

Endogenous metabolism and DNA damage, Exogenous factors: irradiation and carcinogens, DNA lesions and genomic instability, Genotoxicity – classification of genotoxic agents – genotoxicity testing. Dominant lethal assay and Micronucleus test, *In vitro* chromosomal aberrations and SCE's in peripheral blood lymphocyte cultures and HGPRT assay in cultures, DNA adduct analysis.

#### **UNIT 2:**

DNA damage induced mutagenesis, Gene activation following exposure to DNA damaging agents, Cell cycle arrest in response to DNA damage, DNA Damage signaling genes. Direct reversal repair, Excision repair, Translation repair, mismatch repair, Recombination repair; Hereditary Diseases caused by defective DNA, Defective cellular responses to DNA damage resulting in hereditary Diseases, Defective repair of DNA damage and hereditary diseases.

#### **UNIT 3:**

Cancer cell characteristics, modes of cancer, types of cancer, Cell signaling in cancer cells. Regulation of cancer cell, Cell transformation and tumourigenesis, Tumour progression: angiogenesis and metastasis,

#### **UNIT 4:**

Familial cancers, Genetic predisposition to sporadic cancer, Chromosomal aberrations in neoplasia, Tumour specific markers, Cancer and environment: physical, chemical and biological carcinogens, Cancer - treatment and prevention.

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### **HG-502 : GENETIC ENGINEERING:**

#### **UNIT 1:**

Construction of genomic and cDNA libraries, Positional cloning: RFLP mapping, chromosome walking and jumping. Restriction mapping.

#### **UNIT 2:**

Principles of hybridizations and hybridization based techniques (Southern, Northern and *in situ* hybridizations), Expression based screening, Interaction based screening: colony, plaque, yeast two-hybrid system, S1 nuclease and RNase mapping.

#### **UNIT 3:**

Screening and characterization of clones, Preparation of probes, Promoter characterization: promoter analysis through reporter genes, DNA foot-printing, DNA fingerprinting, Mutagenesis, Gene transfer techniques, Microarray.



**UNIT 4:**

Gene therapy: somatic and germ line gene therapy, The Human Genome project (HGP), Single Nucleotide Polymorphisms (SNPs), Proteome and Proteomics, DNA drugs and vaccines. Recent advances in the field of Human Genetics.

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**HG-503 : CLINICAL AND MOLECULAR GENETICS:**

**UNIT 1:**

Monogenic diseases with well known molecular pathology, Inborn errors of metabolism and their genetic bases, Genome imprinting Syndromes, Uniparental disomy, mosaicism.

**UNIT 2:**

Genetic disorders of Haemopoetic systems, Genetic disorders in skeleton and skin, Complex polygenic syndromes, multi-factorial syndromes.

**UNIT 3:**

Muscle genetic disorders, mitochondrial syndromes, Genetic disorders of eye.

**UNIT 4:**

Genetic basis of male and female infertility, Diagnostic Molecular Genetics. Neurogenetic disorders.

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**HG-504 : STEM CELL BIOLOGY AND NANOMEDICINE:**

**UNIT 1:**

Stem cells - Introduction, types, isolation, growth and maintenance, confirmation, differentiation.

Molecular Genetics (Basis) of stem cells, Embryonic and Adult.

**UNIT 2:**

Applications and recent advances in stem cell research and therapeutics.

Neuronal, skin, dental, bone, cardiac, reproductive, muscles, glands, etc.

**UNIT 3:**

GLP, GCP, GMP, Intellectual property rights and protection (IPR & IPP). Patent of materials and products of biological origin, Quality assurance. Accreditation in Medical genetics. Application in Genetics.

**UNIT 4:**

Nanotechnology – Definition, Introduction to Nanotechnology, Nanomolecules of the cells, methods for use of nanomolecules, drug delivery, applications in biomedical genetics.

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**HG-505PR : Practical V : Based on topics covered in HG 501 and 502**

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**HG-506PR : Practical VI : Based on topics covered in HG 503 and 504**

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**Reference Books for HG Semester III:**

<b>Sr. No.</b>	<b>Book Title</b>	<b>Author</b>	<b>Publisher</b>	<b>Year</b>
1	DNA Science	Micklos & Freyer	Coldspring	1990
2	Recombinant DNA	Watson et al.	Freeman	1992
3	Gene Cloning: An introduction	Brown	Stanley Thormes	1995
4	Gene Regulation	Latchman	Chapman	1995
5	Manipulation and Expression of recombinant DNA	Robertson et al.	Academic Press	1997
6	Molecular biotechnology	Glick and Pasternak	ASM press	1980
7	Applied Molecular Genetics	Miesfield	Wiley	1999
8	Casearett and Doulls Toxicology	Klassen	MCGraw Hill	6th Ed.
9	Diagnostic Cytogenetics	Rolf: Dierer Wegner (ed.)	Springer	1999
10	Gene VIII	Lewin	Pearson	2004
11	Human Chromosomes. Manual of Basic techniques	Verma and Babu	Pergamon	1989
12	Human Molecular Genetics	Strachan & Read	Wiley	1999
13	Mutagenecity testing: A Practical Approach	Venitt and Parry	IRL Press	1984
14	Nanobiotechnology			
15	Culture of Animal Cells: A manual Of Basic Technique	Freshney,RI	Wiley-Liss,New York	2006
16	<a href="http://www.nih.gov.in">www.nih.gov.in</a>			
17	<a href="http://www.isscr.org/public">http://www.isscr.org/public</a>			
18	Nanotechnology	Niemeger CM & CA. MIRLCIU	Wiley	2004
19	Bionanotechnology	D.S. Goodsell	Wiley	2004
20	Advanced Nanotechnology (I&II)	S. K. Prasad	Discovery	2008
21	The Nanoscope Encyclopedia of Nanoscience and Nanotechnology (I & VI)	Dr. Parag Diwan	PENTAGON	2005
22	The Potential of Stemm cells: An Inventory	Nikolaus Knoepffler	Ashgate	2007
23	Clinical Cancer Genetics, Risk counseling and management	Kenneth-Liss	Wiley-Liss	1998
24	Principles of cancer Genetics	Fred Bung	Springer	2008
25	Molecular Biology of Cancer	Lauren Pecorino	Oxford Uni. Press	2008

## **HUMAN GENETICS SEMESTER – IV**

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**HG-507S : Seminars and Industrial Visits during the Semester.**

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**HG-508 : Assignments / Group Discussions.**

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**HG-509PT: Project Dissertation and Viva-voce**

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**TIME TABLE**  
**HUMAN GENETICS (SEMESTER – ) YEAR 201 -201 .**

BMT-I	TIME	MON	TUE	WED	THUR	FRI	SAT
<b>THEORY</b> (09:00 TO 11:00)	<b>09:00</b> <b>10:00</b>						
	<b>10:00</b> <b>11:00</b>						
<b>11:00</b> <b>11:30</b>	<b>R E C E S S</b>						
<b>PRACTICAL</b> (11:30 TO 02:30)	<b>11:30</b> <b>12:30</b>						
	<b>12:30</b> <b>01:30</b>						
	<b>01:30</b> <b>02:30</b>						
<b>THEORY</b>	<b>02:30</b> <b>03:30</b>						