MSc (Human Disease Genetics)

A course-outline submitted by

the Centre for Human Genetics

to

Bangalore University

July 2018

Outline of the MSC course

ADMISSIONS

CHG conducts an admission process, detailed below, which has been followed since the institute's MSc programme was initiated in 2015, with the concurrence of the Government of Karnataka and the Governing Body of the institute.

Eligibility

1. The student is required to obtain at least 50% in his/her Bachelor's programme overall.
2. Bachelor's degree in any branch of science/technology/medicine (with degrees such as BSc, BE,
3. BTech, BPharm, MBBS, BDS, BVSc and BAMS)
4. The eligible subject areas are: Life sciences (zoology, botany, genetics, human biology, general life sciences, ecology, environmental biology), bioinformatics, biotechnology, chemistry, physics, mathematics, statistics, any branch of engineering, pharmaceutical sciences, agriculture, medicine, dentistry, horticulture, forestry and veterinary sciences.
5. There is no age bar for applying.
6. Applicants who have had a break in their education or have been working for a few years are also eligible.

Entrance Test

CHG conducts a national level online entrance test.
It is an objective type test (without negative marking). The syllabus for the test is basic physics, maths, chemistry, biology and English language comprehension.

Interview

Students are called for a personal interview based on the cut off marks in the entrance test.
Approximately 60 students are called for the interview.
The interview is held at CHG in Bangalore.
Interviews of students through Skype call is allowed.

Admission Score

1. The Admission Score is based on the interview and the candidate's previous academic performance.
2. The interview contributes 50% to the Admission Score.
3. An average of the candidate's marks secured in 10th, 12th and the Bachelor's degree contributes to the remaining 50% of the Admission Score.

Reservation Policy

The reservation policy applicable in Karnataka is followed.

Fee Structure

The fee structure of the institute will be decided in consultation with the University.

SCHEME OF EVALUATION

Theory

1. Each theory course will be evaluated by both internal assessment (30%) and final assessment (70%).
2. Two internal assessments will be conducted for each 4 credit theory course and one internal assessment for each 2 credit theory course. Internal assessments will consist of tests/seminars/assignments or can be based on class participation.
3. For the hard core theory course HDG11 and the soft core course HDG16, the final assessment will be based on essays (two essays of 2500 and 1000 words each or one essay of 3000 to 4000 words) submitted by each student. Details of the final assessment for the remaining theory courses are mentioned below.
4. A minimum attendance of 75% is compulsory for every theory course.

Laboratory

1. Each practical course will be evaluated by both internal assessment (30%) and final assessment (70%).

2. The internal assessment will be as follows: For the 4 –credit practical courses, 20 marks will assigned for the record and 10 for laboratory participation/assignments. For course HDGP2, which is a 2 credit laboratory course, 10 marks will be allotted for the record and 5 for laboratory participation/assignments.
3. For HDGP4, 7 and 10 'dry lab' courses where there are no records, internal assessment will be conducted in the form of assignments.
4. For HDGP10 the final exam will involve a sitting at a clinical session, followed by solving of problems. Details of the final assessments for the remaining laboratory courses are mentioned below.
5. A minimum attendance of 75% is compulsory for every laboratory course.

Term paper

1. The term paper will be researched and written individually.
2. The topic will be allotted during semester III to facilitate students to initiate it during the semester break and continue during semester IV.
3. Faculty members of CHG will serve as guides.
4. This course in writing will be evaluated both by internal assessment (30%) and final assessment (70%).
5. The internal assessment will be as follows: the first assessment for 15 marks will conducted by the guide, based on his/her interactions with the student. The second assessment for 15 marks will be based on a presentation on the proposed plan of the paper judged by a minimum of three CHG faculty members.
6. The term paper will be evaluated by two examiners from CHG for 70 marks.

Research methodology paper

1. The research methodology paper will be written and submitted by the end of the semester III and will comprise a research proposal on the project work that the student intends to undertake during the fourth semester.
2. It will be written individually.
3. Faculty members of CHG will serve as guides.
4. Students may wish to conduct the research in collaboration with other research institutes, start-ups or larger companies. In these cases, representatives from these other institutions could serve as co-guides.
5. This course will be evaluated both by internal assessment (30%) and final assessment (70%).
6. The internal assessment will be as follows: 15 marks for a presentation on the research proposal judged by a minimum of three CHG faculty members.
7. The research proposal will be evaluated by two examiners from CHG for 35 marks. In the event that the student is guided by more than one faculty member of CHG, or by representatives from other institutions, the research proposal will be evaluated separately by all guides and an additional faculty member from CHG.

Project work

1. Project work will be carried out individually.
2. Projects will be allotted by the end of semester II. Some preliminary work may be carried out during semester III, to aid in the writing of the research methodology paper.
3. In-house projects are preferred.
4. Faculty members of CHG will serve as guides.
5. Students may be allowed to carry out the project work in collaboration with other research institutes, start-ups or larger companies.
6. Co-guides from the collaborating institution/company are allowed.
7. Internal assessment on project work will be made by the guide/s for 30 marks and will be based on the student’s day to day performance in the laboratory.
8. The dissertation will be evaluated by two examiners from CHG for 70 marks.
9. The project viva voce examination will be held at CHG by a minimum of three faculty members for 50 marks (35 marks for the presentation and 15 marks for the viva voce).

SCHEME OF FINAL THEORY EXAMINATION
(Hard Core – 4 credits)

Time 3 Hours	Max. Marks 70
Section A Answer the following objective type questions (20/25)	20 x 1 =20
Section B Answer the following short answer type questions (10/12)	10 x 2 = 20
Section C Answer the following long answer type questions (3/6)	3 x 10 = 30

SCHEME OF FINAL THEORY EXAMINATION
(Soft Core – 2 credits or Hard Core – 2 credits)

Time 2 Hours	Max. Marks 35
Section A Answer the following objective type questions (5/6)	5 x 1 =05
Section B Answer the following short answer type questions (5/6)	5 x 2 = 10
Section C Answer the following long answer type questions (4/5)	4 x 5 = 20

SCHEME OF FINAL PRACTICAL EXAMINATION
(For courses HDGP1, HDGP3, HDGP5, HDGP6 and HDGP9)

Experiment type	Marks (70)
Major Experiment/s	40
Minor Experiment/s	20
Viva voce	10

SCHEME OF FINAL PRACTICAL EXAMINATION
(For course HDGP2)

Experiment type	Marks (35)
Major Experiment/s	20
Minor Experiment/s	10
Viva voce	5

SCHEME OF FINAL PRACTICAL EXAMINATION
(For courses HDGP4, HDGP7 and HDGP10)

Experiment type	Marks (35)
Major problem/s or code/s	20
Minor problem/s or code/s	10
Viva voce	05

M.Sc. (Human Disease Genetics) (CBCS)
Effective from the academic year 2018-2019
SCHEME OF INSTRUCTION AND EXAMINATION, SEMESTER SYSTEM

Course code	Course title	Course Type	Hours/week	Duration of Exam (hrs)	Marks IA	Marks EA	Marks Total	Credits
I Semester								
HDG01	Cell Biology	Theory , H Core	2	2	15	35	50	2
HDG02	Fundamentals of Genetics	Theory , H Core	2	2	15	35	50	2
HDG03	Human cytogenetics	Theory , H Core	2	2	15	35	50	2
HDG04	Biostatistics	Theory , H Core	2	2	15	35	50	2
HDG05	Human Embryology, Anatomy and Physiology	Theory , H Core	4	3	30	70	100	4
HDG06	Science writing and presentation	Theory , S Core	3	2	15	35	50	2
HDGP1	Practical Module I – Genetics	Lab , H Core	8	4	30	70	100	4
HDGP2	Practical Module II – Basic cell immortalization techniques	Lab , H Core	4	3	15	35	50	2
HDGP3	Practical Module III – Human cytogenetics	Lab , H Core	8	4	30	70	100	4
HDGP4	Practical Module IV – Biostatistics and programing in R	Lab , H Core	4	3	15	35	50	2
	Total hours of instruction/week		**39**		**Total marks and credits**		**600**	**26**
II Semester								
HDG07	Biochemistry	Theory , H Core	2	2	15	35	50	2
HDG08	Molecular Biology	Theory , H Core	4	3	30	70	100	4
HDG09	Principles of Development	Theory , H Core	4	3	30	70	100	4
HDG10	Bioinformatics	Theory , H Core	2	2	15	35	50	2
HDG11	Principles of evolution and population genetics	Theory, H core	2	Essay evaluation	15	35	50	2
HDGP5	Practical Module V – Biochemistry and Molecular Biology	Lab , H Core	8	4	30	70	100	4
HDGP6	Practical Module VI – Cell Biology	Lab , H Core	8	4	30	70	100	4
HDGP7	Practical Module VII – Bioinformatics	Lab, H Core	4	2	15	35	50	2
	Total hours of instruction/week		**34**		**Total marks and credits**		**650**	**24**
III Semester								
HDG12	Human Molecular Genetics	Theory , H Core	4	3	30	70	100	4
HDG13	Biochemical genetics	Theory , H Core	4	3	30	70	100	4
HDG14	Essentials in Immunology	Theory , H Core	2	2	15	35	50	2
HDG15	Genetic counselling and ethics	Theory , H Core	2	2	15	35	50	2
HDG16	Use of model organisms to study human disease	Theory , S Core	3	Essay evaluation	15	35	50	2
HDGP8	Practical module VIII – Research methodology	Lab , H Core		Proposal evaluation	15	35	50	2
HDGP9	Practical Module IX - Molecular Diagnostics	Lab , H Core	8	4	30	70	100	4
HDGP10	Practical module X – Genetic counselling and ethics	Lab , H Core	4	3	15	35	50	2
HDG17	Course will be held in other suitable departments	OE	4	3	30	70	100	4
	Total hours of instruction/week		**31**		**Total marks and credits**		**650**	**26**
IV Semester								
HDG18	Cancer biology	Theory , H Core	2	2	15	35	50	2
HDG19	Human genome organization	Theory , H Core	2	2	15	35	50	2
HDG20	Genetics of infertility and assisted reproduction technology	Theory, H Core	2	2	15	35	50	2
HDG21	Introduction to disease biology	Theory , H Core	2	2	15	35	50	2
HDGP11	Scientific writing and presentation – term paper submission	Lab , H Core		Paper evaluation	30	70	100	4
HDGP12	Dissertation work	Lab , H Core		Report evaluation Viva	30 15	70 35	100 50	8 2
	Total hours of instruction/week		**8**		**Total marks and credits**		**450**	**22**
					Grand total of marks and credits		**2350**	**98**

Syllabus

SEMESTER 1

Course code and title		Type	Core	Credits
HDG01	Cell Biology	T	H	2
HDG02	Fundamentals of Genetics	T	H	2
HDG03	Human cytogenetics	T	H	2
HDG04	Biostatistics	T	H	2
HDG05	Human Embryology, Anatomy and Physiology	T	H	4
HDG06	Science writing and presentation	T	S	2
HDGP1	Lab Module I – Genetics	L	H	4
HDGP2	Lab Module II – Basic cell immortalization techniques	L	H	2
HDGP3	Lab Module III – Human cytogenetics	L	H	4
HDGP4	Lab Module IV – Biostatistics and programing in R	L	H	2

Abbreviations: T- Theory, L- Lab, H- Hard core, S- Soft core

HDG01. CELL BIOLOGY TH2

Unit I - Cell Theory and Introduction to Cell Biology (2hrs): History and breakthroughs in cell biology properties and behaviour of cells, diversity of cell types; differences and similarities in the basic structure and functioning of prokaryotic and eukaryotic cells

Unit II - Biological membranes (2hrs): Origin and evolution of primitive cells; composition of biological membranes: Lipids and lipid modification, membrane proteins; functions, techniques used to study membranes and membrane proteins introduction to cell organelles and functions, covering mitochondria in detail.

Unit III - Cell organelles (2 hrs): Structure, function and diseases associated with cell organelles such as mitochondria, ER, Golgi.

Unit IV - Nuclear Architecture and Organization (3 hrs): Nuclear membrane, nuclear transport, nuclear organizer region, kinetochore and centrosome.

Unit V - Cell Division (5 hrs): Mitosis and meiosis, cell cycle, cell cycle controls, phases of cell cycle, techniques to study cell cycle

Unit VI - Cytoskeleton and cell motility (3hrs): Components of the cytoskeleton, organization in stationary and motile cells; functioning of the cytoskeleton and molecular motors; diseases associated with cytoskeleton

Unit VII - Protein and Small Molecule Trafficking (3 hrs): Receptor-mediated endocytosis; intra-cellular transport, lysosomes, organelle biogenesis; extra-cellular transport: biogenesis of membrane proteins, protein modification, glycosylation; pumps, channels and transporters.

Unit VIII - Principles of Cell Signalling (5 hrs): Basics of signal transduction—ligands, receptors, second messengers, effector molecules**;** regulation of signalling pathways—negative and positive regulation, feed-back regulation**;** major known signalling pathways and their functions; techniques to study cell signalling—reporter assays, phosphoprotein analysis

Unit IX - Senescence and Programmed cell death (1 hrs): Replicative Senescence, Hayflick limit and telomere maintenance**;** intrinsic and extrinsic pathways of apoptosis**;** de-regulation of these pathways in diseases**;** techniques to study senescence and apoptosis

HDG02. FUNDAMENTALS OF GENETICS TH2

Unit I - Heredity and Variation (2 Hrs): Definition of heredity and terminology used in the study of genetics; history and significance of the study of genetics; the different branches of genetics including an introduction to genetic diversity and evolution; the future of genetics.

Unit II - Mendelian laws or Principles of heredity and the Inheritance of Mendelian traits in Humans (3 Hrs): Mendel's success in studying inheritance; principles of segregation and independent assortment; concept of dominance and recessivity of traits, dihybrid

crosses and their genetic outcomes; qualitative and quantitative traits; patterns of inheritance in humans using well known examples such as the ABO blood group locus.

Unit III - Chromosomal basis of Inheritance (3 Hrs): Historical experiments establishing chromosomes to be the basis of inheritance; the relationship between Mendel's laws and chromosome transmission in mitosis and meiosis; effect of meiosis and recombination on the results of genetic crosses; mode of inheritance: autosomal and allosomal inheritance; X linked loci, for example, *white* gene inheritance in *Drosophila*; sex linkage, sex influenced characters, genes located on human sex chromosomes and associated diseases; an introduction to sex determination, in particular, XX and XY sex determination in humans.

Unit IV - Extensions and modifications of the Basic principles of Heredity (6 Hrs): Types of dominance, interaction between genes, modified di-hybrid ratios resulting from presence of lethal alleles, multiple alleles, genetic epistasis; inheritance of complex traits, genetics of multifactorial traits, locus/allelic/clinical heterogeneity; effects of inbreeding; extra-chromosomal inheritance**:** maternal inheritance in humans (mitochondrial inheritance, Leigh syndrome), cytoplasmic inheritance; environmental effects on phenotypes, concept of penetrance (illustrated by Huntington's disease) and expressivity; X inactivation, mosaicism and chimerism; pleiotropic gene function.

Unit V - Mapping of Genetic Loci (2 Hrs): Linkage, crossover and map distance, Sturtevant's linkage map, interference in crossover, X^2 test of linkage, mapping functions, genetic polymorphisms, chromosomal mapping.

Unit VI - Introduction to Human Pedigree analysis (2 Hrs): Constraints and special features of human biology and culture, constructions of pedigrees and symbols used, analysis of pedigrees; autosomal recessive traits, autosomal dominant traits, X- linked recessive traits, X- linked dominant traits and Y-linked traits; interpretations of pedigree analysis, genetic testing and risk assessment; studies of twins to assess the importance of genes and environment.

Unit VII - Identifying Human Disease Genes (2 Hrs): Discussion on positional cloning illustrated using as examples, Duchenne muscular dystrophy, cystic fibrosis and Huntington's disease; discussions on concepts of genome wide association, new genome technologies for discovering disease genes, the role of genetic variations in understanding human disease and personalized medicine.

Unit VIII - Identification of the Genetic Material and the Concept of a Gene (4 Hrs): Classical experiments: by Griffith; Avery, MacLeod and McCarty; Hershey and Chase; Beadle and Tatum; Benzer's deletion mapping and complementation; McClintock's jumping genes.

Unit IX - Introduction to Microbial Genetics (2 Hrs): Exchange of genetic material in bacteria: conjugation, transformation and transduction; natural gene transfer of antibiotic resistance, horizontal gene transfer, mapping of bacterial genes using phages, fine structure of bacteriophage genes; bacterial and viral gene maps.

HDG03. HUMAN CYTOGENETICS TH2

Unit I - Introduction to Cytogenetics and Clinical Cytogenetics (2 Hrs): History of human cytogenetics, confirmation of human chromosome number, morphology of human chromosomes, non-banding techniques, classification of human chromosomes into different groups (A-G), international system for human cytogenetic nomenclature, various conferences held to discuss chromosome nomenclature; karyotyping.

Unit II - Introduction to Cytogenetic Techniques (3 Hrs): Conventional banding patterns of chromosomes; specialized banding techniques – Q- banding, G- banding, C banding, silver staining for nucleolus organizer region (NOR), R-banding, sister chromatid exchange (SCE), chromosome analysis, chromosome band nomenclature, Identification and definition of chromosome landmarks, regions, bands and sub-bands, high resolution banding (HRB); immortalization of cells - Epstein-Barr virus (EBV) transformation of lymphocytes to generate lymphoblastoid cell lines.

Unit III - Application of Cytogenetics in Medical Genetics (**3 Hrs):** General principles, Chromosome abnormalities and human genetic diseases: numerical and structural (markers, isochromosomes, ring chromosomes, deletion, duplication, insertions, translocations and inversions) abnormalities; sex chromosome abnormalities, autosomal abnormalities, uniparental disomy, Chromosome breakage Studies (chromatid and chromosome breaks) and their Applications.

Unit IV - International System for Human Cytogenetic Nomenclature (ISCN) and Quality Assurance (2 Hr): General principles, specification of breakpoints, designating structural chromosome aberrations by breakpoints and band composition, short system for designating structural chromosome aberrations, two break rearrangements, three break, four break rearrangements and more complex rearrangements, detailed system for designating structural chromosome aberrations, additional symbols, derivative chromosomes, recombinant

chromosomes, questionable identification, uncertain breakpoint designations, alternative interpretations. Variations in heterochromatic segments, satellite stalks and satellites, fragile sites, inversions as normal variations.

Unit V - Introduction to Cancer Cytogenetics (2 Hrs): Application of cytogenetics in cancer diagnosis (karyotyping), analysis and interpretation of results, quality assurance, Clones and clonal evolution, definition of a clone, clone size, mainline, stemline, sideline, clonal evolution, composite karyotype, unrelated clones, modal number, constitutional karyotype, chromosome markers found in different Lymphomas and leukemias (CML, AML, APML, myelodysplastic syndromes etc.,) and solid tumors (Sarcomas and carcinomas).

Unit VI - Introduction to Molecular Cytogenetics (1 Hr): History of molecular cytogenetics, various molecular techniques applied in clinical cytogenetics, advantages and applications in clinical cytogenetics.

Unit VII - Clinical Applications of Fluorescence *in situ* Hybridization (FISH) (2 Hrs): Principles, procedure, labelling of DNA (Direct and Indirect methods), antibodies used to detect the probe signals, probe amplification, advantages of FISH, various tissue samples used for FISH study.
Kinds of FISH probes – Alpha satellite, telomeric, NOR specific, chromosome specific paint probes, unique sequence specific, repetitive sequence etc., and their applications in clinical diagnosis of various syndromes giving examples of normal and abnormal results.

Unit VIII - Application of FISH in Prenatal Diagnosis and confirming Microdeletion syndrome (2 Hrs): Principles and procedure involved, alpha satellite and unique sequence FISH probes used in prenatal diagnosis of genetic abnormalities on cultured and uncultured cells using appropriate examples.
Prenatal diagnosis of trisomies that could lead to live birth. Postnatal diagnosis of microdeletion syndromes (Prader-Willi, Angelman, Williams, DiGeorge etc.,) using FISH probes, confirmation of cryptic translocations by FISH using appropriate examples.

Unit IX - Application of FISH in Cancer Diagnosis (3 Hrs): Principles and procedure, details of FISH probes used in detecting various markers [(BCR/ABL, t(15;17), t(8;21) etc.,] found in Leukemia and solid tumors(HER-2/neu, C-myc, p53 etc.,), use of single fusion, dual fusion, break apart and multipanel probes used in cancer detection, confirmation of probe amplifications seen in breast cancer and solid tumors, section *in situ* hybridization used to study probe amplifications on tissue sections, RNA *in situ* hybridization on tissue sections using appropriate examples.

Unit X - Advanced Molecular Cytogenetic Techniques (3 Hrs): Principles and procedures involved and their applications in clinical diagnosis of genetic abnormalities (including complex chromosomal translocations – CCRs etc.,) - Primed *in situ* labeling (PRINS), comparative genomic hybridization (CGH), Spectral karyotyping (SKY), multicolor FISH (mFISH) and multicolor banding (mBAND), Fiber FISH, etc., using appropriate examples.

Unit XI - Quality Assurance (2 Hrs): Interpretation and Reporting of normal and abnormal reports using International System for Human Cytogenetic Nomenclature (ISCN) for FISH.

HDG04. BIOSTATISTICS **TH2**

Unit I - General Introduction and Probability (4 Hrs): Data types, descriptive statistics versus inferential statistics; measures of central tendency: mean, median, mode**;** measures of spread**:** variance, standard deviation, coefficient of variation**;** tabulation and visual display of data**;** introduction to probability: sample space, events, definition of probability, permutations and combinations, events and operations on events, conditional probability, concept of independence, Bayes' rule and screening tests, Bayesian inference; ROC curves; prevalence and incidence.

Unit II - Distributions (3 Hrs): Introduction; random variables and their properties**;** permutations and combinations; the Binomial distribution: expectation (mean) and variance; the Poisson distribution: expectation and variance, computation of Poisson probabilities; the Gaussian distribution and its properties

Unit III - Estimation (3 Hrs): Introduction; the relationship between population and sample; random-number tables, randomized studies; estimation of the mean of a distribution; estimation of the variance of a distribution; estimation for the Binomial distribution; estimation for the Poisson distribution; the Central Limit Theorem; the concept of confidence interval; confidence interval for the mean and its interpretation.

Unit IV - Hypothesis Testing (4 Hrs): One-sample inference, one-sample test for the mean of a normal distribution: one-sided alternatives; one-sample test for the mean of a normal distribution: two-sided alternatives; the relationship between hypothesis testing and confidence intervals; Bayesian inference; one-sample $\chi 2$ test for the variance of a normal distribution, one-sample inference for the binomial distribution, one-sample inference for the Poisson distribution; two-sample inference; the paired t-test; interval estimation for the comparison

of means from two paired samples; two-sample t-test for independent samples with equal and unequal variances, interval estimation for the comparison of means from two independent samples (equal variance case); testing for the equality of two variances.

Unit V - Categorical Data (4 Hrs): Two-sample test for binomial proportions, Fisher's exact test; two-sample test for binomial proportions for matched-pair data (McNemar's test); estimation of sample size and power for comparing two binomial proportions, R×C contingency tables; chi-square goodness-of-fit test, the kappa statistic.

Unit VI - Nonparametric Methods (2 Hrs): the sign test, the Wilcoxon signed-rank test, the Wilcoxon rank-sum test.

Unit VII - Regression and Correlation Methods (4 Hrs): General concepts; fitting regression lines, assessing the goodness of fit including the method of least squares; inferences to be made from the parameters of regression lines; interval estimation for linear regression; the correlation coefficient, inferences from correlation coefficients; multiple regression, partial and multiple correlation, rank correlation, interval estimation for rank correlation coefficients.

Unit VIII - Multisample Inference (4 Hrs): Introduction to the one-way analysis of variance (ANOVA): fixed effects model**,** hypothesis testing; comparisons of specific groups, using one-way and two-way ANOVA; The Kruskal-Wallis test; the random effects models in one-way ANOVA, the intra-class correlation coefficient; mixed models.

HDG05 HUMAN EMBRYOLOGY, ANATOMY AND PHYSIOLOGY TH4

PART 1 - HUMAN ANATOMY and PHYSIOLOGY

Unit I - Introduction to tissue organization in relation to human physiology (5 Hrs): Levels of organization and organ systems of the body; different kinds of tissues; cavities of the body (cranial cavity, vertebral cavity, thoracic, abdominal and pelvic cavities).
Epithelial tissue: Structure of epithelia, different kinds of simple and compound epithelia and tissue specific variations, example keratinised versus non-keratinised.
Different types of Connective tissue and special connective tissue: Components of connective tissue, cellular and matrix components; components and features of proper connective tissue, fluid connective tissue (blood and lymph) and supporting connective tissue (cartilage and bone); distribution of these tissues in various organs; special mention on classification of bones, example Pneumatic bones, sesamoid bones, developmental classification, structural classification; growth of bones.
Muscular tissue: Structure, classification and function of different kinds of muscles: cardiac, smooth and different kinds of skeletal muscles according to colour and direction of organization; structure of myofibrils and the neuro-muscular junction; common diseases that affect the muscles.
Glands: General structure of a gland, exocrine, endocrine and paracrine glands; epithelial glands classified according to structure (simple and compound duct, tubular or alveolar structure) and secretions (example serous, mucous); diseases that affect the glands.
Skin: Structure, function and components of the skin; structure and function of the appendages of the skin such as nails, hair, sweat glands and sebaceous glands; diseases that affect the skin.
Unit II - Fluid composition of the body (2 Hrs): Body fluid compartments, intracellular, extracellular and interstitial; blood cells and their lineage
Cardiovascular and lymphoid system: gross anatomy of the heart and major blood vessels; arterial and venous systems, valves, structure and histology of vessels; maintenance of blood pressure, conduction system of heart; blood capillaries, movement of blood cells through capillaries; lymphatic tissue, major lymph vessels and organs, lymph capillaries, spleen, tonsils, thymus; genetic disease that affect the cardiovascular system.

Unit III - Digestive system (3 Hrs): parts of the digestive system and their functions, innervation of gastrointestinal tract, accessory organs of digestion, gastrointestinal hormones, physiology of digestion, absorption and satiation.

Unit IV - Urinary system (2 Hrs): parts of the urinary system and their functions, cellular organization of the kidney, physiology of urine formation, glomerular filtration, tubular reabsorption, maintenance of water balance; physiology of human urinary system compared with that of other land animals.

Unit V - Respiratory system (3 Hrs): parts of the respiratory system and their functions, nasal cavities, tracheobronchial tree; passage of air, physiology of respiration, ventilation and gaseous exchange in lungs, oxygen-haemoglobin dissociation curve, transport of carbon dioxide; nervous control of respiration.

Unit VI - Essentials of the nervous system (4 Hrs): structure and physiology of neurons, glia and ganglia; structure and physiology of synapses and neurotransmitters; divisions of the nervous system: central nervous system (CNS), peripheral nervous system (PNS) and autonomic nervous system (ANS), elaborating briefly on their various components and the functional areas of cerebrum, cerebellum and

brainstem and special sense organs such as eyes and ears; structure of ventricles and physiology of the cerebrospinal fluid; structure and physiology of the choroid plexuses, meninges and blood brain barrier; blood supply to the brain and spinal cord.

Unit VII - Endocrine system (4 Hrs): Structure and secretions of Pituitary gland, Thyroid gland, parathyroid glands, Suprarenal and Adrenal glands, Pancreas; Hormones regulating calcium, energy and water homeostasis, body temperature and growth.

Unit VIII - Reproductive System (3 Hrs): Components and functions of the male and female reproductive system and accessory organs, such as the mammary gland**;** cellular organization of important organs of the reproductive system such as the uterus, ovary, testis and mammary glands; physiology of the male and female reproductive systems, hormonal cycles and puberty.

PART 2 - HUMAN EMBRYOLOGY

Unit I - Fertilization and assisted reproductive technologies (3 Hrs): formation of gametes, process of fertilization; diseases causing sterility; in-*vitro* fertilization techniques

Unit II - Early embryogenesis (3 Hrs): cleavage, the germinal stage, gastrulation, formation of the germ layers and body cavities; body axis specification; brief mention of the different kinds of birth defects and prenatal diagnosis; description of the phylotypic pharyngula stage. **Development of the placenta.**

Unit III - Development of the skeletal system (3 Hrs): vertebral column and ribs, commonly occurring genetic diseases affecting the skeletal system.
Unit IV - Development of the bronchial apparatus, face, nose and palate (1 Hr): normal development and commonly occurring diseases including cleft palate.

Unit V - Development of the special sense organs (2 Hrs): eyes, ears.

Unit VI - Development of the nervous system (3 Hrs): Neurulation and subsequent development of the spinal cord and major areas of the brain; commonly occurring anomalies affecting the development of the nervous system example hydrocephaly, anencephaly.

Unit VII - Development of the gastrointestinal tract (2 Hrs): divisions of the gut tube and their anlage; epithelial-mesenchymal interactions in the specification of different parts of the gut tube; partitioning of foregut into oesophagus and respiratory diverticulum; formation and rotation of stomach and midgut, the occurrence of herniation; formation of liver, spleen and pancreas; commonly occurring genetic diseases resulting in deviations in normal development.

Unit VIII - Development of the cardio-vascular system (3 Hrs): formation of the heart fields and then heart tube, cardiac loop; brief description of formation of the cardiac septa; vascular development: vasculogenesis and angiogenesis; formation of the aortic arches and then vascular pattern; formation of the venous system; foetal circulation and circulatory changes after birth; anomalies resulting in fused septa and thinning of capillaries.

Unit IX - Development of the urogenital system (3 Hrs): development of the pronephros, mesonephros and metanephros; formation of the kidney from the metanephric mesoderm and uretric bud; development of the gonads: formation of the genital ridges, migration of germ cells to somatic gonads; development of the testis, ovary and uterus; hormonal influence on development of the genital system; genetic diseases of the urogenital system affecting kidney function and sterility.

Unit X - Development of the respiratory system (3 Hrs): formation of the lung buds, branching of the trachea and formation of lung lobes; maturation of lungs, association of terminal bronchiole sacs with blood capillaries.

HDG06. SCIENCE WRITING AND PRESENTATION TS2

Unit I - Basics of good writing (13 Hrs): English grammar in the current context of writing: nouns and pronouns; adjectives; verbs; adverbs; prepositions; conjunctions, articles; order of words; sentence structure, clauses, punctuation, vocabulary; idioms; figures of speech; active vs. passive voice, etc.; writing skills: paragraph construction; common mistakes while writing, better sentences, story writing; precis writing.

Unit II - Structure of scientific articles (11 Hrs): differences in structure between reviews and papers**;** composition of an abstract, title, introduction, methods, results and discussion; preparing figures and figure legends, citations; importance of engaging the reader, telling a story and providing the right emphasis on important results; avoiding plagiarism; adapting to different journal requirements; attempts will also be made to critique published articles.

Unit III - Constructing arguments (11 Hrs): Learning how to recognize the argument being made in a passage, learning how to represent it graphically using rationale online software and then paraphrasing it. Learning how to critically analyse the strengths and weaknesses of an argument and then to write such that sufficient emphasis is given to each. Articles from editorial sections of the journal *Science* will be used to practice these exercises.

Unit IV - Other forms of professional writing (1 Hr): composing professional emails and letters for various purposes; preparing an effective resume.

Unit V - Effective oral presentations (2 Hrs): essential principles of a good talk, importance of engaging the audience, telling a story and providing the right emphasis on important results; preparing effective presentations: preparing images, graphs and tables, appropriate use of colour and font size; talks for different audiences.

Unit VI - Other modes of scientific communication (1 Hr): Short accounts of scientific discoveries for the layman; social media and science communication; using social media to transmit a scientific discovery that has influenced you; the importance of engaging with the community and communicating scientific knowledge to the community.

HDGP1. LAB MODULE I - GENETICS **LH4**

1. **Introduction to Laboratory course on Genetics:** Understanding laboratory ecosystems, 'do's and don'ts' of laboratory safety, familiarization with instruments to be used; measurement of weight and volume, preparation of solutions, buffers, media, sterile areas, sterilization methods; record keeping.
2. **Growth curve analysis:** Culturing bacteria to study the phases of growth; quantification of bacteria by plating serial dilutions and by spectrophotometry; plotting growth curves under different growth conditions, for example different culture media.
3. **Bacterial Conjugation**: Transfer of antibiotic resistance genes across different strains of bacteria by conjugation.
4. **Introduction to *Drosophila* genetics:** Life cycle of *Drosophila*, stages of growth and identification of anatomical structures; husbandry and handling of *Drosophila*; historical perspective and relevance of *Drosophila* in present day genetic analysis; an introduction to the database, Flybase.
5. **Analysis of *Drosophila* mutants**: Observation and identification of genetic markers of *Drosophila,* basics of setting up genetic crosses; phenotypes from monohybrid and dihybrid crosses in *Drosophila*; probability and chi square analysis of cross results.
6. **Identification of X-linked genes**: Inheritance of the *white* gene locus to demonstrate sex linkage; analysis of reciprocal crosses.
7. **Qualitative and Quantitative analysis of traits:** Phenotypes of mutants affected in movement (flight, gait), eye colour, geotaxis, photo taxis, wing shape, wing size etc.; temperature sensitive mutations.
8. **Genetic pathways, for example, genes involved in metabolism**: Demonstration of mutations or gene products involved in the synthesis of *Drosophila* eye pigments; separation of the constituent pigments of mutants using thin layer paper chromatography.
9. **Preparation of Cytogenetic map using polytene chromosomes**: Dissection of salivary glands from third instar larvae, spreading of polytene chromosomes, fixation using aceto-methanol fixative, observing banding pattern using Orcein stain and a light microscope.
10. **Creating a genetic map using *Drosophila* markers**: Setting recombination crosses and preparing a recombination map from recombination frequencies.
11. **Mapping of P-element insertions**: Mapping P-element insertions by segregation and linkage analysis, introduction to P-element enhancer and protein trap lines and their application in Genetics.
12. **Demonstration of the use of biochemical markers:** reporter gene (ß-Galactosidase) staining or immunohistochemistry to observe tissue specific expression of *vg-lacZ* or *wg-lacZ* insertion lines.
13. **Brief discussion on the use of the currently vast Genetic tool kit in *Drosophila* to understand gene function**: Use of methods such as spontaneous mutations, X-ray and EMS mutagenesis, P-element based mutagenesis, Crispr-Cas9, RNA interference.

HDGP2. LAB MODULE II – BASIC CELL IMMORTALIZATION TECHNIQUES **LH2**

1. **Basics of cell culture, Good laboratory practice (GLP), Techniques of cell cultures** – short-term lymphocyte culture, long-term culture, sub-culturing, primary and secondary cell cultures, maintenance, culture contaminations, detection of contaminations; *in vitro* transformation of human peripheral B-lymphocytes by Epstein-Barr virus to establish lymphoblastoid cell lines (LCLs).
2. **Culture of patient tissue samples**: blood/percutaneous umbilical blood sampling (PUBS)/amniotic fluid/chorionic villus specimen (CVS)/products of conception (POC)/Skin/bone marrow (BM)/solid tumor, etc.

3. **Principles of Microscopy**: Parts of a light microscope, principle of the Koehler illumination system, phase contrast microscopy.

HDGP3. LAB MODULE III – HUMAN CYTOGENETICS **LH4**

1. **Karyotyping using karyotype workstation**– chromosome preparations from short term cultures and long term cultures and G-Banding. Identification of chromosomes under the microscope, use of software in capturing G-banded metaphases, chromosome analysis and karyotyping.
2. **Conventional and specialized cytogenetic techniques (C- banding and NOR staining)**: peripheral blood (PB) cytogenetics, prenatal cytogenetics (AF/CVS/POC/PUBS), culture from skin biopsy, cancer cytogenetics from Bone marrow, FNA culture and culture from tissue biopsy and chromosome preparations and karyotyping: case study analysis and inference. Analysis and interpretation of results.
3. **Chromosome breakage study**: Mitomycin C induced cultures, principles, procedure, applications and case studies.
4. **Quality Assurance:** Interpretation and Reporting of normal and abnormal reports using International System for Human Cytogenetic Nomenclature (ISCN).
5. **Fluorescence Microscopy** – Principles and use of Fluorescence microscopy
6. **FISH:** Specimen culture; preparation of slides for fluorescence *in situ* hybridization (FISH). Selection of probes, *in situ* hybridization, washings, detection of probes by antibodies, washings, observations using Fluorescence microscope, image capture and analysis using FISH workstation.
7. **Advanced FISH techniques**: SKY, mFISH and mBANDing
   SKY: Slide preparation, probe hybridization, probe detection, Methods in spectral (Color) image capture and analysis.
   Multiplex fluorescent *in situ* hybridization (**mFISH**): Slide preparation, probe hybridization, probe detection, Methods in multicolor image capture and analysis.
   Multicolor banding (**mBAND**): Slide preparation, probe hybridization, probe detection, Methods in multicolor image capture and analysis.
   Analysis and interpretation of results.
8. **Quality assurance**: Interpretation and Reporting of normal and abnormal reports using International System for Human Cytogenetic Nomenclature (ISCN) for FISH.

HDGP4. LAB MODULE I LAB MODULE IV – BIOSTATISTICS AND PROGRAMMING IN R **LH2**

1. R software installation and basic R usage.
2. Mathematical operations and string manipulation
3. Basic data structures: Vectors, data frames, lists and matrices
4. Logical statements and loops: If-else statements, for and while loops, break
5. Writing user defined functions and packages
6. Reading and writing tables and files
7. R graphics library: Line plots, histograms, pie charts, bar plots and other plots
8. Computation of statistical parameters
9. Correlation studies of data
10. Error analysis and error bars
11. Binomial, Poisson and Gaussian distributions and deviates
12. Demonstrations and study of Central Limit Theorem through data
13. Hypothesis testing and p-value computation
14. Parametric tests – Performing one and two parametric Z tests, family of t-tests, tests for proportion, chi-square tests
15. Analysis of variance – Performing one factor and two factor ANOVA
16. Non-parametric tests – Wilcoxon Mann Whitney tests, Kruskal Wallis test
17. Regression analysis: Least square linear regression with errors and non-linear regression of data
18. Using Biostrings library for RNA and DNA sequence analysis
19. Using Excel spreadsheet for statistics calculations.
20. Using GraphPad for basic biostatistics.

****

SEMESTER 2

Course code and title		Type	Core	Credits
HDG07	Biochemistry	T	H	2
HDG08	Molecular Biology	T	H	4
HDG09	Principles of Development	T	H	4
HDG10	Bioinformatics	T	H	2
HDG11	Principles of evolution and population genetics	T	H	2
HDGP5	Lab Module V – Biochemistry and Molecular Biology	L	H	4
HDGP6	Lab Module VI – Cell Biology	L	H	4
HDGP7	Lab Module VII – Bioinformatics	L	H	2

Abbreviations: T- Theory, L- Lab, H- Hard core

HDG07. BIOCHEMISTRY TH2

Unit I - Fundamental Chemistry (1 Hr): Atomic structure; chemical bonds and interactions (types of chemical bonds, bond angles, bond length and bond rotation, weak forces); properties of organic compounds, chirality in biomolecules; mole and molecular mass. Chemical properties of water; hydrogen bonding; the pH scale, acids, bases and buffers.

Unit II - Carbohydrates and Carbohydrate Metabolism (6 Hrs): Structure and properties of monosaccharides; D and L sugars; derivatives of sugars; glycosidic bond; oligosaccharides (disaccharides and trisaccharides); polysaccharides: homopolysaccharides (e.g., starch, cellulose and glycogen) and hetero polysaccharides (e.g., glycosaminoglycans); glycoproteins and peptidoglycans; functions of carbohydrates. Synthesis and degradation of glucose and glycogen in humans (including glycolysis, tricarboxylic acid cycle, glycogenesis, glycogenolysis, gluconeogenesis, and pentose phosphate pathway).

Unit III - Proteins and Protein Metabolism (4 Hrs): Classification, structure and properties of amino acids; peptide bond; classification of proteins; structural organization of proteins (primary, secondary, tertiary and quaternary) with haemoglobin and myoglobin as examples; the Ramachandran plot.
Amino acid metabolism- General reactions of amino acid metabolism- transamination, deamination and decarboxylation; the urea cycle and its regulation; amino acid biosynthesis.

Unit V - Lipids and their metabolism (4 Hrs): Classification; fatty acids (properties and types); triacyl glycerol; phospholipids; glycolipids (with cerebrosides and gangliosides as examples); glycosylphosphatidylinositol; cholesterol; prostaglandins; properties and reactions of lipids; the β-oxidation pathway- even and odd numbered saturated and unsaturated fatty acids; energetics of total oxidation; general scheme of biosynthesis of fatty acids.

Unit VI - Nucleic acids and their metabolism (4 Hrs): Structure of nitrogenous bases, nucleosides and nucleotides; phosphodiester bond; polynucleotides; double helical form of DNA (A, B and Z forms); triplex and quadruplex structures in DNA; RNA (mRNA, rRNA and tRNA); synthesis of purines and pyrimidines: *de novo* and salvage pathways, catabolism of purines and pyrimidines.

Unit VII - Enzymes (3 Hrs): Properties, classification and nomenclature; co-enzymes and co-factors; isozymes; regulation of enzyme activity; mechanism of enzyme catalysis; enzyme immobilization; enzyme kinetics: factors affecting the rate of enzyme-catalyzed reactions such as enzyme concentration, substrate concentration, pH and temperature, Michaelis-Menten equation, significance of Km and Vmax and their determination using Lineweaver-Burk (L-B) plots; enzyme inhibition: reversible and irreversible, reversible-competitive, non-competitive and uncompetitive inhibition with graphical representations using L-B plots.

Unit VIII - Bioenergetics (3 Hrs): Laws of thermodynamics, the concept of Gibb's free energy; generation and utilization of the energy currency, ATP in cells; mitochondrial electron transport chain- components and oxidative phosphorylation.

HDG08. MOLECULAR BIOLOGY TH4

Unit I - Biomolecules and central dogma of molecular biology (4 Hrs): Historical overview of discoveries identifying DNA as hereditary material and its structural organization (contributions of Mischer, Kossel, Levene, Griffith, Avery's group, Hershey and Chase, Chargaff, Franklin, Wilkins, Pauling, Watson and Crick); structure of DNA, RNA and proteins; central dogma; genetic code.

Unit II - DNA replication in prokaryotes and eukaryotes (6 Hrs): Modes of replication: conservative, semi-conservative and dispersive, Meselson and Stahl's experiment; defining a replicon and the origins of replication; leading and lagging strand synthesis at a growing replication fork, Okazaki's experiment; DNA polymerase and replication machinery in prokaryotes and eukaryotes; processes of initiation, elongation and termination; trombone model of replication; end replication problem of linear DNA, telomerase and the Shelterin

complex; early and late replicating DNA; visualizing replication; isolation of autonomous replicating sequences (ARS); coordinating regulation of DNA replication initiation with the prokaryotic and eukaryotic cell division cycle

Unit III - DNA Recombination (4 Hrs): Overview of recombination and gametogenesis, genetic and cytological evidence of crossing over; mechanisms of recombination: the Holliday model, the Meselson-Radding heteroduplex model and the double-strand-break model; gene conversion and branch migration; molecules in recombination: synaptonemal complex, ruvABC, recA and recBCD

Unit IV - Transcription in prokaryotes and eukaryotes (6 Hrs): Subunit organization and structure of prokaryotic RNA polymerase and eukaryotic RNA Polymerase I, II and III; core promoter elements, analysis of promoters: deletion analysis, DNase foot printing, base-modification and mutational assays; transcription initiation, prokaryotic initiation and role of sigma factor; general transcription factors (GTFs) and initiation in eukaryotes, binary and ternary complexes of RNA polymerase (RNAP), abortive initiation and DNA scrunching; transcription elongation, catalytic synthesis of RNA, role of bridge helix and trigger loop in nucleotide addition cycle and translocation, role of pol II carboxy terminal domain (CTD) in mRNA processing, proofreading and backtracking of RNA polymerase, role of prokaryotic GRE factor and eukaryotic TFIIS in release of RNA polymerase arrest; transcription termination: intrinsic and rho-dependent termination in prokaryotes, PAS-dependent termination in eukaryotes

Unit V - Post-transcriptional processing and nuclear transport (5 Hrs): Splicing: evidence of split genes, splicing mechanism and splicing signals, spliceosome assembly and function, role of RNA polymerase II CTD, self-splicing RNAs, tRNA splicing; mRNA capping: cap structure, synthesis and function; mRNA polyadenylation: function of poly(A), mechanism of polyadenylation, polyadenylation signals, poly(A)polymerase and turnover of poly(A); coordination of mRNA processing events: effect of individual events on the other events, coupling transcription termination with mRNA_3'-end processing, mechanism of termination; processing of rRNA; RNA editing, editing by nucleotide deamination; mRNA stability, stability of specific mRNAs such as Casein mRNA and transferrin receptor mRNA; gene silencing by RNA interference.

Unit VI - Translation (6 Hrs): Ribosome composition and structure; discovery of tRNA and structure of tRNA, recognition during translation (second genetic code), proof reading and editing by amino-acyl-tRNA synthetases; prokaryotic translation initiation: tRNA charging, dissociation of ribosomes, formation of initiation complexes, control of translation at the level of initiation; eukaryotic translation initiation: initiation factors, scanning model of initiation, regulation of translation at the level of initiation; prokaryotic and eukaryotic translation elongation and termination: direction of polypeptide synthesis, genetic code and codon bias, steps in elongation, structure of EF-Tu and EF-G, regulation by GTPases, termination codons, release factors; dealing with aberrant termination.

Unit VII - Post-translational processing of proteins (5 Hrs): Polypeptide cleavage, brief mention of chemical modifications such as ubiquitin and ribozymes.

Unit VIII - Brief overview of eukaryotic gene regulation (10 Hrs): Basic concepts of gene regulation, cis and trans regulators, positive and negative gene regulation, operons, *lac* operon as example; organization of the genome: size, complexity, coding and non-coding sequences, repeat sequences; packaging of DNA: structure of chromatin, heterochromatin and euchromatin, chromosomal territories, lampbrush and polytene chromosomes, higher order packaging and fractal globules; expression of the genome: activators and repressors, DNA-binding motifs, promoter proximal elements, enhancers, insulators and barrier elements, mediators, transcription factories, chromatin remodelling during transcription; epigenetic modifications and their role in gene expression: DNA modifications and histone modifications, histone code; molecular basis of imprinting and X-inactivation; the role of transcription factors in cellular differentiation, identity and reprogramming; RNA mediated gene regulation: microRNA, long non-coding RNA; techniques used to study gene expression and regulation: promoter assays, reporter assays, chromatin immunoprecipitation (ChIP), chromosome conformation capture (3C) and related techniques, hybridization techniques, yeast-two hybrid assay.

Unit IX - Mutations and repair (4 Hrs): types of mutations; chemical and physical mutagens and mode of action: induction of mutations by tautomerization, base analogues, deamination, base damaging agents such as aflatoxins, alkylating agents, oxidative damage and formation of pyrimidine dimers; mutations induced at repeat sequences: replication slippage, unequal crossing-over and unequal sister chromatid exchange; cellular repair pathways: base excision repair, nucleotide excision repair and mismatch repair, photo-reactivation, non-homologous end joining (NHEJ), recombination mediated repair and translesion synthesis; defects in repair and associated human diseases.

Unit X - Mitochondrial DNA (2 Hrs): Genome organization and codon usage; mitochondrial mutations and associated diseases.

HDG09. PRINCIPLES OF DEVELOPMENT TH4

Unit I - Developmental Biology: Basic Principles and Concepts (1 Hr). Introduction to the evolution of the body plan: acoelomate to coelomate, diploblastic and triploblastic organization of body plan; introduction to information flow from mother to egg; different sized embryos, yolk density and cleavage patterns; transition from a plastic, uncommitted state to a determined and then differentiated state;

introduction to the development of four model organisms, namely nematodes (*C elegans),* fruit flies (*D melanogaster),* fish *(D rerio),* frog *(X laevis),* and mammals (mouse and human); life cycles of these organisms.

Unit II - Origins of polarity in the embryo (6 Hrs): Differences in the origins of polarity between different kinds of embryos namely nematodes (*C elegans),* fruit flies (*D melanogaster),* fish *(D rerio),* frog *(X laevis),* and mammals (mouse and human); inheritance in most cases of polarity from the egg; specification of polarity in mammalian zygotes; differences in time at which dorso-ventral and anterio-posterior polarity is specified in all above five kinds of embryos.
Student discussions, schematic preparations and presentations (1Hr)

Unit III - Basics of pattern Formation (1 Hr): Formation of protein gradients during development, pattern formation within syncitia (syncytial *Drosophila* embryo) versus simple epithelia (cellularised *Drosophila* embryo); introduction to long range and short-range pattern formation.
Student discussions, schematic preparations and presentations (1Hr)

Unit IV - Formation of the germ layers (5 Hrs): Early specification of germ layers and the derivatives of each germ layer in insects (fruit fly) and vertebrates (fish, frog, mammals); long germ band and short germ band insects; Spemann's organizer in frog, fish and mammalian embryos; gastrulation in invertebrates and vertebrates; morphogenetic movements and cell shape changes accompanying the formation of each germ layer, apical constriction, cell movement during convergent extension (involvement of planar cell polarity and specific signalling pathways), delamination of cells; cytoskeletal distribution accompanying certain cell movements such as apical constriction and dorsal closure in *Drosophila* embryos; induction between germ layers
Student discussions, schematic preparations and presentations (3Hrs)

Unit V - Emergence of the Body Plan (16 Hrs): Segmentation in insects and vertebrates across the different germ layers; mechanisms and signalling pathways involved in segment formation in *Drosophila* embryos, somitogenesis and rhombomere formation in vertebrates; formation of compartment boundaries during development; segment identity specification by Hox genes; the emergence, development and patterning of limbs in *Drosophila* (wings) and vertebrates (formation and patterning of limb bud and formation of digits), signalling pathways involved during these processes**.**
Student discussions, schematic preparations and presentations (6Hrs)

Unit VI - Cell Migration (1 Hr): role of migration during development, epithelial to mesenchymal transition and vice versa during somitogenesis, neural crest formation and directed migration of individual cells; branching of tubular structures, for example, trachea formation in insect embryos and lung development in mammalian embryos.
Student discussions, schematic preparations and presentations (1Hr)

Unit VII - Specification of Cell Fate (1Hr): Signalling events accompanying cell fate specification in certain well characterized contexts, for example, Notch-Delta signalling in neuroblast specification; the role of lineage, asymmetric and symmetric cell divisions.

Unit VIII - Determination of size (4 Hrs): Regulation of body size, organ size and cell size; concept of allometry; coordination of growth with development; coordination of growth with cell division; regulation of cell size versus cell number; using *Drosophila* imaginal discs as an example, organ intrinsic and extrinsic (systemic) regulation of size; pathways that regulate growth (eg. insulin/TOR signalling pathways); bone development and growth in vertebrates.

Unit IX - Germ cell development (2 Hrs): Specification and migration of germ cells to form gonads in invertebrates and vertebrates; specification of germ cells in different regions of the *Drosophila,* fish, frog and mammalian embryos; conserved molecules involved in germ cell development across species (eg. *vasa*, *nanos*, mitochondrial ribosomes), suppression of somatic programme in primordial germ cells; signalling pathways that guide migration of primordial germ cells to the gonads during development.

Unit IX - Techniques used to study development (4 Hrs): Making transgenic *Drosophila, D. rerio* and mouse embryos; different kinds of mutagenesis methods: chemical mutagenesis, use of transposons (different kinds of transposable elements used in the different model organisms) and site directed mutagenesis (different uses for Cripr-Cas9 technology); *in vivo* expression of tagged wild type and mutant proteins; FLP-FRT and CRE-LOX technology in generating mosaics.

TH2

HDG10. BIOINFORMATICS

Unit I - Introduction to Bioinformatics and its Scope (1 Hr): An overview of activities in bioinformatics with emphasis on the types of information in modern biology and the need for databases and software.

Unit II - Operating Systems, Basics of Computer Programming, Languages, and Commands (5 Hrs): Use of Linux operating system and commonly used open software; introduction to basic Linux commands, programming concepts and commonly used programming languages

Unit III - Databases (6 Hrs): General concepts of databases; an overview of database types; common databases, related software and their utilities (Pubmed, Entrez Gene, Gene cards, Uniprot, dbEST, OMIM, Unigene, HPRD, NEBcutter, Primer3plus, Primer Blast); using DNA sequence databases for PCR primer designing and restriction site analysis.

Unit IV - Human genome analysis (4 Hrs): NGS analysis for single nucleotide polymorphism (SNP) detection; case studies; significance of SNPs; understanding the relationship between mutations, SNPs, insertions or deletions (indels), copy number variations (CNVs) and alleles; SNP and other variant databases and analysis of reference and sample sequences using the databases (dbSNP, Clinvar, dbVar, and Cosmic); genome wide association studies (GWAS); genomic versus exomic analysis. Concept of metagenomics; types of metagenomic approaches and their relative significances; significance of microbiomes to human health.

Unit V - Sequence analysis (5 Hrs): importance of sequence analysis; common methods used in sequence analysis and alignment; basic concepts of sequence similarity, identity and homology, definitions of homologs, orthologues and paralogs; common databases used to retrieve gene, mRNA and protein sequences; phylogenetic analysis with reference to nucleic acids and proteins, and their significance; comparison of operation and applications of BLAST and MSA, and interpretation of results; concepts in DNA and RNA motif analysis, relevant databases and software.

Unit VI - Transcriptome Analysis (5 Hrs): Significance of data and analysis at various levels: genome vs. exome vs transcriptome and proteome; commonly used databases for obtaining and analysing transcriptomic data; micro-array technology and analysis; discussions on RNA-seq analysis; miRNA analysis; ChIP-seq technology and related data analysis; DNA motif analysis.

Unit VII - Bioinformatics for Proteins (5 Hrs): Review of protein structures and domains; use of databases for analysing protein-structures, gene ontologies, protein-interactions and pathways; domain analysis; significance of interaction analysis and systems biology; concepts in homology modelling, drug discovery and design; biologicals, biosimilars vs. traditional active pharmaceutical ingredients (APIs) and concept of clinical trials.

HDG11. PRINCIPLES OF EVOLUTION AND POPULATION GENETICS TH2

Unit I - Origins (1 Hr): an introduction to the concept of evolution and the idea that all living forms are related; evidence for evolution, fossil record, classification and phylogeny, evolutionary transitions among genera, major transitions during evolution, Cambrian explosion and terrestrial life.

Unit II - Adaptation and Natural Selection (2 Hrs): phenotypic variation vs. genetic variation; theory of Natural Selection; the 'selfish gene' concept; individual selection vs. group selection; patterns in adaptation for example, parasite-host adaptation; biogeographic evidence for evolution, major patterns of distribution of species.

Unit III - Genes and evolution (1 Hr): mutation rate, adaptive vs deleterious mutations, mutations with no selective advantage, consequences of gene duplication, genetic variation and evolution.

Unit IV - Basic population genetics (2 Hrs): frequencies of alleles and genotypes, Hardy-Weinberg principle and its significance in evolution; genetic variation in natural populations, geographic variation and genetic distance.

Unit V - Genetic drift (4 Hrs): theory of genetic drift; inbreeding and its effects on genetic drift; models of gene flow and genetic drift; the 'neutral theory of molecular evolution'; alternatives to natural selection.
Population structure and Speciation: population structure and gene trees.

Unit VI - Phylogeny and Ontogeny (1 Hr): developmental patterns of evolutionary change, developmental constraints, non-adaptive characters and discontinuity of evolutionary change, evolution of novelty.

Unit VII - Evolution of Sex and Sexual selection (1 Hr): selective advantages of asexual reproduction versus sexual reproduction, sex ratios, evolution of sexual dimorphism, mate choice, signal detection.

Unit VIII - Evolution of Social behaviour (1 Hr): evolution of cooperation and apparent altruism.

Unit IX - Evolution of Life Histories (2 Hrs): life history traits, life span and senescence.

Unit X - Evolution of Homo sapiens (2 Hrs): Phylogenetic relationships, fossil record and origin of modern human populations, migration and genetic variation in human populations; the evolutionary future of humans; evolution of human behaviour and cultural evolution.

Unit XI - Human Disease and Evolution (3 Hrs).

Unit XIII - Presentations by students on recent papers on evolution and human health **(6 Hrs)**

HDGP5. LAB MODULE V: BIOCHEMISTRY & MOLECULAR BIOLOGY LH4

1. **Isolation and Estimation of Proteins:** Preparation of cell lysates from E. coli/ adenocarcinoma cell line (MCF7); estimation of total protein (Lowry/Bradford); analysis of protein of interest by western blotting.
2. **Isolation and Estimation of DNA**: Isolation of DNA from E. coli and human blood; estimation and agarose gel electrophoresis.
3. **Isolation and Estimation of RNA**: Isolation of RNA from adenocarcinoma cell line (MCF7) and blood; estimation and agarose gel electrophoresis.
4. **Molecular cloning:** Preparation of competent cells, transformation of E coli DH5α, preparation of plasmid DNA; restriction enzyme digestion, analysis of products through agarose gel electrophoresis; gel elution of restriction enzyme digested fragments; ligation, transformation and analysis of clones.
5. **Polymerase chain reaction (PCR):** Amplification of DNA sequences from genomic DNA/plasmid DNA/cDNA; optimization of conditions, eg. MgCl2 concentration and annealing temperature.

HDGP6. LAB MODULE VI - CELL BIOLOGY LH4

1. **Basics of cell culture:** Media preparation and filtration; sub-culturing and reviving frozen stocks.
2. **Cell counting, cell viability assay:** Trypan blue exclusion.
3. **Transfection using lipids:** A demonstration of the technique
4. **Cell viability assays:** MTT assay, IC50 determination
5. **Wound healing assay:** Scratch test performed using a confluent culture of a human cell line.
6. **Cell separation and sub-cellular fractionation methods:** A lecture on the theory behind and the usefulness these techniques
7. **Early development of *Drosophila melanogaster*:** collecting synchronised eggs, watching cellularisation and gastrulation in live embryos; fixing and staining staged embryos with nuclear dyes; using GFP-expressing strains that label organelles, cytoskeleton, and mitotic spindles during early development of *Drosophila* embryos and/or larvae.
8. **Later development of *Drosophila melanogaster*:** studying coordination of growth and cell division in mosaic wing imaginal discs: analysis of the size and cell number in wildtype clones marked with GFP-expression with the size and cell number in two sets of clones one co-expressing RBF and the other E2F induced at the same time in larval development as the wild type clones.

HDGP7. LAB MODULE VII – BIOINFORMATICS LH2

1. Operating Systems and Basics of Computer Languages and Commands: Use of Linux operating system, commonly used open software; introduction to Shell, Perl
2. Case studies to use common data mining resources: Pubmed, Entrez Gene, Genbank, Uniprot, HPRD, PubMed & Google Scholar
3. Phylogenetic analysis with reference to nucleic acids and proteins, and their significance; comparison of operation and applications of BLAST and MSA, and interpretation of results; demonstration of DNA and RNA motif analysis using relevant databases and software (JASPER, MEME).
4. Use of major tools for molecular cloning work: NEBcutter, Primer3plus, Primer Blast
5. Case studies of analysis using SNP databases (dbSNP); NGS analysis for SNP detection.
6. Case studies in clinically relevant databases (clinvar and OMIM).
7. Use of databases for protein-structures (PDB), gene ontologies (GO), protein-interactions (STRING) and pathways (KEGG and reactome).
8. Demonstration of data analysis in genomics, transcriptomics (mRNA and miRNA analysis), and metagenomics

****

SEMESTER 3

Course code and title		Type	Core	Credits
HDG12	Human Molecular Genetics	T	H	4
HDG13	Biochemical genetics	T	H	4
HDG14	Essentials in Immunology	T	H	2
HDG15	Genetic counselling and ethics	T	H	2
HDG16	Use of model organisms to study human disease	T	S	2
HDGP8	Research Methodology	L	H	2
HDGP9	Lab Module IX - Molecular Diagnostics	L	H	4
HDGP10	Lab module X – Genetic counselling and ethics	L	H	2
HDG17	Course will be held in other suitable departments	T	OE	4

Abbreviations: T- Theory, L- Lab, H- Hard core, S- Soft core, OE- Open elective

HDG12. HUMAN MOLECULAR GENETICS TH4

Unit I - Introduction to Human Genetics - Genetic Disorders and Single Gene Inheritance (6 Hrs): Overview of genetic disorders: consequences and mechanisms; nomenclature of mutations, importance of the position of a base, databases of known mutations; from genotype to phenotype: loss of function mutations, gain of function mutations, expanding repeats.

Unit I – Complexities of basic-Mendelian pedigree patterns (2 Hrs): Inbreeding and pseudo-dominant inheritance, locus heterogeneity, incomplete and age-related penetrance, phenocopies, *de-novo* mutations and mosaicism

Unit III - Molecular methodologies (6Hrs): DNA polymorphisms, molecular markers and genotyping: restriction site polymorphisms (RSP), minisatellites, microsatellites, single nucleotide polymorphisms (SNP), heterozygosity of a marker, DNA-fingerprinting; polymerase chain reaction (PCR); Sanger and next-generation sequencing (NGS).

Unit IV - Genetic Mapping of Monogenic Traits (12 Hrs): Parametric linkage analysis: markers in mapping, recombinants and non-recombinants, phase-known and phase-unknown pedigree, informative and uninformative meiosis, two-point mapping, multipoint mapping: calculation of logarithm of odds (LOD) scores, haplotype analysis and defining critical linked interval in large pedigrees; autozygosity mapping; principles and strategies of identifying disease genes: positional cloning and position independent approaches, prioritization of candidates, whole-exome and whole genome sequencing in gene-hunting, assigning pathogenicity to sequence variants: genetic and bioinformatic criteria, functional validation of variants; computation exercises for two-point LOD scores for a Mendelian disorder using Mlink program; linkage, manual haplotype construction for large family Mendelian segregating phenotype

Unit V - Multifactorial Traits or Complex Disorders (12 Hrs): Polygenic theory of quantitative traits**;** partitioning of variance, heritability; polygenic theory of discontinuous characters; genetic component assessment in families: risk-ratio, twin and adoption studies; mapping of complex traits by parametric linkage analysis in near-Mendelian families and affected sib-pair analysis; non-parametric linkage analysis: association-mapping studies: role of linkage disequilibrium in association studies, odds ratio and chi^2 test, tag-SNPs, genome-wide association studies (GWAS), Transmission disequilibrium test (TDT); identifying susceptibility variation through association studies, common disease-common variant hypothesis and mutation selection hypothesis.

Unit VI - Sex Linked Disorders, Sex Limited, Sex Influenced Traits, Genomic Imprinting (7 Hrs)

Unit VII - Gene Action _ Tracing Defects in Gene Function (7 Hrs): (a) haemoglobinopathies-, beta-thalassemia and sickle cell anaemia (b)fragile X syndrome (c) hearing impairment (d) epilepsy (e) Gauchers Disease and Glycogen Storage disorders (f) Noonan syndrome and Rasopathies.

HDG13. BIOCHEMICAL GENETICS TH4

The course shall cover various aspects of inborn errors of metabolism (IEM), a large class of genetic diseases in humans. Through the study of IEM, the students can broaden their understanding of the biology of human genetic disorders. The course is divided into nine units.

Unit I - Inborn errors of metabolism (IEM) (6 hours):

***Basic concepts/ general features* (2 Hrs):** Basic concepts; history; inheritance patterns; incidence of IEM; challenges in the management of IEM.

Classes of IEM **(2 Hr):** Classes of IEM — an overview. Classification of IEM based on organelle involved, metabolite/ pathway affected, organs affected, etc. Major classes of IEM.

General clinical features **(1 Hr):** Symptoms; age of onset; and clinical heterogeneity.

Diagnosis **(1 Hr):** Clinical suspicion, biochemical methods (examples: tests for the metabolites and the activity of specific enzymes), molecular genetics methods, and newborn screening; recent advances in the methods for diagnosis of IEM.

Unit II - Phenylketonuria (PKU) (10 hours):

PKU as a model to learn about IEM in general, and about disorders of amino acid metabolism.

Biology of PKU **(5 Hrs):** Discovery of PKU; incidence; causes — (1) mutations in the phenyl alanine hydroxylase (PAH) gene and (2) deficiency of tetrahydrobiopterin; symptoms; "maternal" PKU; diagnosis; the catabolism of phenylalanine in individuals with untreated PKU; how the symptoms manifested in patients may be explained on basis of the various biochemical aberrations; molecular genetics of PKU; structure and regulation of PAH.

Treatment options **(4 Hrs):** Treatment options available and those under development: Dietary management; intake of large neutral amino acids; enzyme replacement/ substitution therapy (with PAH and phenylalanine ammonia lyase); co-factor therapy; advantages and disadvantages of each of these therapies.

Group activity **(1 Hr)**: Screening of the documentary film *The forgotten children*, followed by a discussion.

Unit III - Glycans in mammalian systems (6 hours):

Study of certain aspects of glycobiology as background for the IEM covered in later sections.

Glycosylation in mammalian systems **(1 Hr):** Salient points; protein glycosylation — the biochemistry of N- and O-linked glycosylation and cell organelles where the reactions occur. Functions of glycans — an overview.

Glycosaminoglycans and proteoglycans **(5 Hrs):** General properties and functions of glycosaminoglycans; chemical structures of common glycosaminoglycans (heparan sulfate, dermatan sulfate, keratan sulfate, chondroitin sulfate and hyaluronic acid); an overview of proteoglycans; sources of chemical diversity among glycosaminoglycans and proteoglycans; the general scheme of biosynthesis and breakdown of glycosaminoglycans; the general composition, properties, and functions of proteoglycans; common cellular and anatomical locations of glycosaminoglycans and proteoglycans.

Unit IV - Lysosomes (2 hours): Discovery; properties; functions; trafficking of proteins into lysosomes.

Unit V - Lysosomal storage disorders (LSD) (7 hours):

Biology **(2 Hrs):** Types of LSD; milestones in research on LSD; modes of inheritance; diagnosis of LSD, challenges in the management of LSD; symptoms; biomarkers.

Treatment options **(3 Hrs):** Treatments available and those under development: Dietary management; treatment of specific symptoms; enzyme replacement therapy; substrate reduction therapy; hematopoietic stem cell transplantation; pharmacological chaperone therapy (a.k.a. enzyme enhancement therapy); gene therapy; other treatments suitable for specific LSD.

Group activity **(2 Hrs):** Screening of a popular movie pertaining to treatment of LSDs, followed by discussion.

Unit VI – Gaucher and MPS group of diseases (7 hours):

- Study of Gaucher disease as a typical lipid storage disorder.
- Study of the MPS group as a model for disorders of carbohydrate metabolism.

Gaucher disease **(1 Hr):** Causes, classification, molecular genetics, diagnosis, symptoms, and treatment; the connection between Gaucher disease and Parkinson's disease.

***Mucopolysaccharidoses (MPS*) (6 Hrs):** Definition and types of MPS; biochemical basis, molecular genetic basis, diagnosis, symptoms and treatment of the MPS disorders; salient features of each type of MPS, namely, MPS I (Hurler syndrome, Scheie syndrome and Hurler-Scheie syndrome), MPS II (Hunter syndrome), MPS III (Sanfilippo syndrome) subtypes A–D, MPS IV (Morquio syndrome) subtypes A and B, MPS VI (Maroteaux-Lamy syndrome), MPS VII (Sly syndrome) and MPS IX; basic properties of the enzymes each of whose deficient activity causes an MPS disorder; animal models specifically designed to monitor the efficacy of enzyme replacement therapy.

Unit VII - IEM due to defects post-translational modifications (1 Hr): The study of the biochemical basis of multiple sulfatase deficiency as a model to learn about this group of disorders. Study of mucolipidosis II and mucolipidosis III (Unit VIII) would serve as additional models for diseases due to defects in post-translational modifications.

Unit VIII - IEM due to defects in intracellular trafficking (1 Hr): Study of mucolipidosis II (I-cell disease) and mucolipidosis III (pseudo-Hurler dystrophy) as models for this group of diseases. Study of the biochemical basis of aberrant targeting of proteins into lysosomes in patients with these diseases.

Unit IX - Classroom presentations by students (12 Hrs)

HDG14. ESSENTIALS IN IMMUNOLOGY TH2

Unit I - Organs and cells of the Immune system (1 Hr): Primary lymphoid organs, Secondary lymphoid organs, B lymphocytes, T lymphocytes, mononuclear phagocytes, granulocytes, mast cells, dendritic cells, natural killer cells.

Unit II - Innate immunity (2 Hrs): Anatomical barriers, inflammation, anti-microbial peptides, acute phase proteins, toll like receptors

Unit III - B cell immunity (4 Hrs): Development of B cells, B cell activation, proliferation & differentiation, structure and Functions of Immunoglobulins

Unit IV - Complement system (2 Hrs): Components of the classical, alternate, lectin pathways, complement cascade, biological consequences of complement cascade, regulation of the complement system

Unit V - Antibody diversity (2 Hrs): Organization of immunoglobulin genes, mechanism of gene rearrangements, class switching, regulation of immunoglobulin genes

Unit VI - The major histocompatibility complex and antigen presentation (2 Hrs): General organization of the MHC genes, cellular expression of MHC genes, MHC and immune responsiveness, MHC antigens and antigen processing

Unit VII - T cell immunity (4 Hrs): Development of T cells and their education, T cell activation, proliferation & differentiation, T cell types, T cell receptor and co receptors, signaling in T cells, helper and cytotoxic T cells, cytokines

Unit VIII - Hypersensitivity (2 Hrs): IgE antibody-mediated hypersensitivity, antibody-mediated cytotoxicity, immune complex mediated hypersensitivity, delayed type hypersensitivity

Unit IX - Autoimmunity (2 Hrs): Organ specific autoimmune diseases, systemic autoimmune diseases, mechanisms of autoimmunity, treatment of autoimmune diseases

Unit X - Autoimmunity (2 Hrs): Organ specific autoimmune diseases, systemic autoimmune diseases, mechanisms of autoimmunity, treatment of autoimmune diseases

Unit XI - Immunodeficiency disorders (2 Hrs): Primary immunodeficiency diseases of lymphoid cells, myeloid cells and granulocytes, complement components, acquired immunodeficiency diseases

Unit XII - Cancer and immune system (1 Hr): Tumor evasion of the immune system, cancer immunotherapy

Unit XIII - Vaccines (2 Hrs): Active and passive immunization, vaccine designs, subunit vaccines, DNA vaccines

HDG15. GENETIC COUNSELLING AND ETHICS TH2

PART 1-GENETIC COUNSELLING

Unit I - Introduction to genetic counselling (2Hrs)

Unit II - Impact of illness on patients and families (4Hrs): Morbidity and support services, access to education and economic support, social practices and health cost issues. Incidental detection of other genetic disorders on pedigree evaluation and NGS testing, manifesting carriers, later onset genetic disorders including neuro-genetic disorders, e.g., Huntington's disease, Myotonic dystrophy and Inherited cancers

Unit III - Congenital anomalies and rare medical disorders, its impact on community health and health priorities (4Hrs): Incidence of common congenital anomalies and risk factors, pre-pregnancy evaluation and intervention strategies, new-born screening for rare metabolic disorders, Consanguinity and its impact on genetic disorders, registries and support groups for rare medical disorders

Unit IV - Social and cultural issues in rare medical disorders (3Hrs): Evaluation of social attitudes and customs, caste, consanguinity, economic status and cultural perceptions in the evaluation of genetic disorders. Impact of disorders of sexual development (DSDs) and genetic counselling in these disorders, eg. sex reversal, androgen insensitivity disorder and Klinefelter and Turner syndrome. Impact of rare disorders on decisions about marriage, reproduction, prenatal diagnosis and integration in mainstream education and society.

Unit V - Predictive counselling for late onset disorders, e.g., Huntington's disease, breast and ovarian cancer (3Hrs): Principles about predictive counselling and testing in late onset disorders, imparting results of predictive testing, counselling and management in follow up sessions, ethical issues in testing of minors, prenatal diagnosis in late onset disorders, ethical and social issues

PART 2-ETHICS

Unit I - Clinical Ethics (1 Hr)

Unit II - Ethical issues in prenatal diagnosis and newer reproductive technologies (1 Hr)

Unit III - Genetic testing in adult onset disorders (1 Hr)

Unit IV - Testing of vulnerable populations (1 Hr): for example, children of intellectually incapacitated individuals, consent and confidentiality

Unit V - Research Ethics (1 Hr): the use of placebos, conflicts of interest and clinical trials, research on animals and vulnerable populations, research in developing countries

Unit VI –Role and Scope of Institutional review board in scientific research (1 Hr)

Unit VII - Global & Population Ethics (1 Hr): global disparities in health and public health, global pandemics, population growth, human rights to health and health care, role of foreign aid.

Unit VIII - Ethics of New Technologies (1 Hr): embryonic stem cells, animal cloning, genetic engineering, synthetic biology.

Unit IX - Environmental & Animal Ethics (1 Hr): species preservation, biodiversity loss, ecosystem services, the use and misuse of animals, ethics

Unit X - Medico-legal Issues (1 Hr): Surrogacy, Organ donation, Paternity testing.

HDG16. USE OF MODEL ORGANISMS TO STUDY HUMAN DISEASE TS2, Hours:39

This course is aimed at an interactive learning experience for the students and will consist of seminars and discussions led by invited speakers who use model organisms in their research. Students will also make presentations and write essays defending a particular choice of model organism.

The following topics will be covered:

Unit I - *Planaria*: Using a simple micro-organism to study the cell biology of regeneration.

Unit II - *C. elegans*: Overall advantages of *C. elegans* as a system to study human diseases, ease of culture, ease of performing cellular and genetic studies, conservation in signalling pathways, use of the system to study the biology of metabolic disorders such as diabetes and ageing; use of the system in drug screening.

Unit III - *D. melanogaster*: Overall advantages and disadvantages of *D. melanogaster* as a system to study human diseases, ease of culture, short generation time, ease of performing cellular and genetic studies, formidable range of genetic tools available, conservation of signalling pathways and cellular mechanisms; limitations of the system (for example, innate immunity versus adaptive immunity, specialized organ diseases such as those affecting vision and hearing; disadvantages of using invertebrate models) use of the system to study the biology of diseases such as Alzheimer's disease, neurodegenerative disorders, Parkinson's disease, triplet repeat expansion diseases, Fragile X syndrome, metabolic disorders and diabetes, tumour formation; use of the system in drug screening.

Unit IV - Zebrafish: Overall advantages and disadvantages of the zebra fish *D. rerio* as a system to study human diseases, economy and ease of culture, ease of performing cellular and genetic studies, formidable range of genetic tools available, conservation of signalling pathways and cellular mechanisms; advantages of vertebrate models, similarity in organ systems across different vertebrates; using vertebrate orthologous genetic, mutant models to study haematological diseases such as *sideroblastic anaemia*, polycythaemia, and porphyria; T-cell leukaemia models, Melanomas, heart defects resembling human dilated cardiomyopathies (DCMs), modelling *Duchenne muscular dystrophy*, Polycystic kidney disease (PKD), etc. ; use in drug screening.

Unit V - Mouse and other mammals: Overall advantages and disadvantages of using mammalian systems, similarity in physiology and organ function with humans, similarity in organization and function of the brain and sense organs between mammals and humans, use as models to study behaviour and diseases that affect behaviour, learning and memory; high genetic conservation; ethical issues concerning use of mice, primates and dogs. Studying humanised mouse models containing transplanted human cells or the human orthologues of specific genes; modelling cancers; genetic disorders such as hearing-loss disorders; uses in drug testing and treatment of early onset cancers such as acute promyelocytic leukaemia (APL), role of the protein Leptin in controlling obesity.

Unit VI - Human Induced Pluripotent Stem Cells: as tools in drug development and modelling diseases; the ability to create patient and disease specific stem cells.

HDGP8. LAB MODULE VIII- RESEARCH METHODOLOGY LH2

This course is designed to teach students how to address a research problem of interest. It is an opportunity to explore plausible problems and then define more specifically the project that will be undertaken during the fourth semester. This exercise will be carried out in one of the research laboratories in CHG. The students will be expected to conduct a literature survey, define project objectives, collect material required for the project and conduct preliminary investigations. They will then present their results through a seminar and written report.

HDGP9. LAB MODULE IX- MOLECULAR DIAGNOSTICS LH4

1. **Polymerase chain reaction (PCR):** Types of PCRs and its multiple applications; Gradient PCR, Multiplex PCR, GC rich PCR, ARMS PCR
2. **Primer design:** Guidelines for primer designing; tools for designing primers- NCBI primer BLAST and Oligocalc; Primer design for genes to be used for PCR and sequencing.
3. DNA gel electrophoresis of PCR amplified products
4. Purification of PCR amplified products.
5. Sanger sequencing of gene exons for mutation detection.
6. Analysis of Sequence-electropherograms and analysis of identified sequence variations. Sequencher software for data analysis; Use of NCBI database and mutation Database (HGMD) for analysis of variants; Online prediction tools –SIFT, PROVEAN, PolyPhen and mutation Taster.
7. **Screening for Y microdeletion**: polymorphic markers in AZF region; multiplex PCR
8. **Restriction Fragment length Polymorphism**: SMN exon 7 and exon 8 deletion
9. **Quantitative PCR (qPCR) for estimation of gene copy or transcript expression level:** Detection of SMN gene copy number – to identify heterozygous or homozygous status. Analysis of qPCR data, calculation of double delta Ct value.

10. Genotyping with PCR for deletion screening.
11. STR-based Human Identification; GeneMarker software.
12. Detection of Fragile X mutation though capillary electrophoresis; Calculation of CGG repeat size
13. Multiplex Ligation Dependent Probe Amplification (MLPA). Analysis of MLPA data with Coffalyser software.

HDGP10. LAB MODULE X - GENETIC COUNSELLING AND ETHICS LH2

Time spent in clinics (clinical rotations)

This course consists of visits to the clinic, where students obtain first-hand experience of the symptoms presented and patients' concerns. These rotations will provide an opportunity for students to learn directly about medical genetic conditions and their impact on individuals and families and interpretation of medical diagnosis in real life clinical sessions based in a hospital outpatient setting. They will gain practical insight in pedigree evaluation, recent advances in medical genetics, therapy options, prenatal diagnosis and practical genetic counselling.

The students then participate in the discussions that ensue, that helps in the diagnosis of the disease and decisions on possible courses of action.

****

SEMESTER 4

Course code and title		Type	Core	Credits
HDG18	Cancer biology	T	H	2
HDG19	Human genome organization	T	H	2
HDG20	Genetics of infertility and assisted reproduction technology	T	H	2
HDG21	Introduction to disease biology	T	H	2
HDGP11	Scientific writing and presentation – term paper submission	L	H	4
HDGP12	Dissertation work	L	H	10

Abbreviations: T- Theory, L- Lab, H- Hard core

HDG18. CANCER BIOLOGY TH2

Unit I - Basics about Cancer (1 Hr): Introduction to cancer, origin, types (types of tumours, stages of malignancy), basic terminology.

Unit II - Hallmarks of Cancer (5 Hrs): Basic mechanisms regulating normal tissue homeostasis: regulation of cell-proliferation, growth, differentiation and apoptosis; aberrations in regulatory mechanisms that result in cancer.

Unit III - Genetic and Epigenetic Alterations in Cancer (13 Hrs): The nature of commonly occurring mutations in cancerous tissue: gain of function, loss of function, copy number variation (CNV), chromosomal-translocations etc.; signalling pathways commonly affected in cancers; oncogenes: mechanisms of activation and action, different functions of oncogenes, rationale for therapeutic targeting.
Tumour suppressor genes: mechanisms of loss of function, loss of heterozygosity, Knudsen's two hit hypothesis, different functions of tumour suppressor genes.
Caretaker and gatekeeper genes.
Epigenetic alterations: role of the Polycomb group (PcG) and Trithorax (Trx) proteins in carcinogenesis; basis of epigenetic therapy.
Methods of detecting genetic alterations and their use as diagnostic/prognostic tools; targeting genetic alterations for therapy; mouse models for understanding the role of these gene products in the development of cancer.

Unit IV - Viral causes of cancer (1 Hr): History of discovery, mechanism of action of commonly seen viruses such as HPV, EBV, HBV, HCV.

Unit V - Familial cancer syndromes (1 Hr): NF1, FAP, VHL, etc.

Unit VI - Molecular mechanisms of metastasis (1 Hr): Different steps and cellular state transitions in metastases; genes responsible for metastases; organ specific metastases.

Unit VII - Tumour microenvironment (1 Hr): Composition of the tumour microenvironment; mechanisms of tumour angiogenesis, targeting angiogenesis for therapy; mechanisms of immune evasion, targeting immune evasion for therapy.

Unit VIII - Metabolic reprogramming in cancer (1 Hr): The phenomenon known as Warburg effect.

Unit IX - Challenges in treatment of cancer (2 Hrs): The development of therapeutic resistance, the occurrence of a relapse; current developments in treating cancers.

HDG19. HUMAN GENOME ORGANIZATION TH2

Unit I - Mapping the Human Genome (techniques used and historical perspective) (6 Hrs): DNA markers in human genetic mapping, RFLP, occurrence of single nucleotide polymorphisms; current data from understanding human genome libraries, expression libraries, DNA microarray and CHIP technology for genome wide analysis; linkage analysis and genetic mapping; an over view of genome sequencing technology, assembling genomic DNA sequences, the human genome draft sequence.

Unit II - Organization of the human genome (an overview) (6 Hrs): Complexity, size, heterochromatin vs euchromatin; comparisons with genomes of model organisms; case study annotation of human genes and visualization of gene tracks using UCSC genome browser; examples: non coding RNA Xist locus, Myc locus, Promoters (Ink4a) , Enhancer elements (P300)

Unit III - Understanding genome function (4Hrs): Studying the human transcriptome and proteome **Coding vs non-coding sequences in humans:** Regulatory sequences, open reading frame (ORF) organization; microRNA and other non-coding RNAs; mobile genetic elements and repeats.

Unit IV - Comparison of the Genetic features of Human Nuclear Genomes and organelle Genomes (5 Hrs).

Unit V - Chromatin modification and gene expression in humans (5 Hrs): Epigenetic landscapes of human genome; use of ENCODE and modENCODE to understand regulatory epigenetic landscape dynamics during development and disease; DNA modification and gene expression, analysis of DNA elements in human genome, DNA binding sites of proteins and their signature in the genome; examples of current CHIP-seq analyses in humans; transcriptional initiation through a genomic perspective, enhancer-promoter interactions, chromatin contact mapping in 3D as visualized in hi-seq/5C or FISH (3D architecture) taking examples of human studies.

HDG20. GENETICS OF INFERTILITY AND ASSISTED REPRODUCTION TECHNOLOGY TH2, Hours - 26

Unit I - Oogenesis and Folliculogeneis: Oocyte retrieval and selection. Preparation and evaluation of oocytes for ICSI. Hyaluronic acid binding-mediated sperm selection for ICSI.

Unit II - Spermatogenesis and Andrology: Evaluation of sperm. Sperm preparation techniques. Sperm chromatin assessment.

Unit III - Fertilization and Embryos in assisted reproduction technology (ART): Embryology, In Vitro Fertilization (IVF), Analysis of Fertilization, Morphological Assessment of Embryos and Oocytes, Embryo Transfer Techniques, Cryopreservation and Vitrification, Time Lapse Videos.

Unit IV - Endometrial Receptivity and Female Factor Sterility: Window of implantation, endometrial cycle, Assessment of Receptivity, Impact of Ovarian Stimulation.

Unit V - Male Infertility: Etiology and Pathophysiology, Clinical and Endocrinal Evaluation.

Unit VI - Genetics of Infertility: Cytogenetic Abnormalities, Genetics of y chromosome-Derived Infertility, Molecular Genetic Testing.

Unit VII - Female Factor Infertility: Uterine, Cervical, Tubal and Fallopian tube factors.

Unit VIII - Infertility and Molecular Genetics in Females.

Unit IX - Advances and Dilemmas in Assisted Reproductive Technologies.

Unit X - Preimplantation Genetic Screening of Embryos.

Unit XI - Preimplantation Genetic Diagnosis.

Unit XII - Preimplantation Genetic Diagnosis and HLA typing.

Unit XIII - OMICS in Infertility.

HDG21. INTRODUCTION TO DISEASE BIOLOGY — TH2, Hours - 26

Unit I – Cancer Biology: What is disease biology, Etiology and pathogenesis, Hallmarks of cancer, Cancer as a disease and theories of origin- Mathematical models, Clonal evolution theory, Cancer Stem cell theory, tumour measurement, tumour diagnosis using Histochemistry, tumour gradation and growth; role of mutations in cancer progression; resistance to radiotherapy- need to discover new radio-sensitizers

Unit II - Hematological Disease/disorders- Etiology and Pathogenesis of Hemophilia-A, microparticles and their role in hematological disorders.

Unit II - Infectious Disease- Etiology, Epidemiology of Fungal infections and their significance in population health, Diagnosis using PCR

Unit IV – Flow cytometry - Theory sessions on principles of flow cytometry and applications of flow-cytometry. Hands on training in handling samples for flow-cytometry and data analysis.

Unit V – Biology of Kidney diseases: Introduction to Kidney diseases (special emphasis on Nephrotic syndrome); SNP and population genetics- genotype and allele frequency calculation; mutation nomenclature and classification; high resolution melting analysis with qPCR – analysis and applications; a practical guide to the use of various SNP databases; Vitamin A pathway gene polymorphism and kidney size.

HDGP11. SCIENTIFIC WRITING AND PRESENTATION - TERM PAPER SUBMISSION — LH4

The student will write and present an up to date review of literature on a selected topic or alternately, a project proposal. This would serve as a platform to enhance the student's skills in reading scientific literature, critical analysis of the literature and communication skills. Special attention will be given to discouraging plagiarism.

HDGP12. DISSERTATION — LH10

This course consists of an individual research project to be carried out in one of the research laboratories in CHG. The students will be expected to complete the project and present their results through a seminar and written dissertation.

****

References

SEMESTER1

HDG01. Cell Biology

1. JD Watson (2013) **Molecular Biology of the Gene,** Pearson Publication, 7th Edition
2. B Alberts, A Johnson, J Lewis, M Raff, K Roberts and P Walter (2014) **Molecular Biology of the Cell**, Taylor and Francis Publication, 6th Edition
3. H Lodish, A Berk, SL Zipursky, P Matsudaira, D Baltimore and J Darnell (2012) **Molecular Cell Biology**, W. H. Freeman and Company, 7th Edition
4. JE Krebs, ES Goldstein and ST Kilpatrick (2012) **Lewin's Gene XI**, Jones and Bartlett Learning Publication, 11th Edition

HDG02. Fundamentals of Genetics

1. Choi, Jung H. (2017) **Genetics: a conceptual approach.** New York: W.H. Freeman/Macmillan Learning.

2. Elrod, Susan L. (2010) **Schaum's outlines: genetics**. New York: McGraw-Hill.
3. Falconer, D. S., and Trudy F. Mackay (1996) **Introduction to quantitative genetics,** Essex, England: Longman.
4. Fogiel, M. (1985) **The Genetics Problem Solver.** New York, N.Y: The Association.
5. Knowles, Richard V. (2001) **Solving problems in genetics,** New York: Springer.
6. Griffiths, Anthony J., et al. (2015) **Introduction to genetic analysis**, New York, NY: W.H. Freeman & Company.
7. Hartl, Daniel L., and Elizabeth W. Jones (1998) **Genetics: principles and analysis,** Sudbury, Mass: Jones and Bartlett Publishers.
8. Pierce, Benjamin A., and Benjamin A. Pierce (2014) **Genetics: a conceptual approach,** New York: W.H. Freeman and Company.
9. Snustad, D P., and Michael J. Simmons. (2012) **Principles of genetics**. Hoboken, NJ: Wiley.
10. David T. Suzuki, Anthony J.F. Griffiths**,** et al, (1996) **An Introduction** to **Genetic Analysis,** 6th Revised edition New York: W H Freeman & Company
11. T Strachan and AP Read (2011), **Human Molecular Genetics**, Garland Science/Taylor and Francis Group Publication, 4th Edition.
12. Robert L. Nussbaum, Roderick R, McInnes C M, Huntington F Willard (2015), **Thompson & Thompson Genetics in Medicine**, 8th edition, Elsevier.

HDG03. Human cytogenetics

1. Steven L. Gersen and Martha B. Keagle (1999) **The Principles of Clinical Cytogenetics** Humana Press
2. Orlando J. Miller (2000) **Human Chromosomes** Springer-Verlag New York
3. Morgan Key (2015) **Cytogenetics: Techniques and Applications** CALLISTO REFERENCE
4. Swansbury (2003) **Cancer Cytogenetics** Humana Press
5. Wan**,** Thomas (2017) **Cancer Cytogenetics** Humana Press
6. RL Nussbaum, RR. McInnes and HF Willard (2007) **Thomson and Thomson Genetics in Medicine**, Saunders, Elsevier Publication
7. JL Hamerton (2013) **Human Cytogenetics: Clinical Cytogenetics**, Academic Press
8. S Gersen and MB Keagel (2013) **The Principles of Clinical Cytogenetics**, Springer Science and Business Media Publication
9. LB Jorde, JC Carey and MJ Bamshad (2009) **Medical Genetics**, Elsevier Publication
10. S Heim, F Mitelman (2011) **Cancer Cytogenetics: Chromosomal and Molecular Genetic Aberrations of Tumor Cells**, John Willey and Sons Publications

HDG04 and HDGP4: Biostatistics and R programming

1. JH Zar (2010) **Biostatistical Analysis**, Prentice Hall Publication, 5th Edition
2. W. W. Daniel (2013) **Biostatistics - Basic concepts and methodology for the health sciences**, Wiley Student edition
3. R. B. D'Agostino Sr., L. M Sullivan and A. S Beiser (2006) **Introductory Applied Biostatistics**, Thomson Brooch.
4. M. R. Spiegel, J. J Schiller and R. A. Srinivasan (2001) **Probability and Statistics**; Schaum's outline Series, McGraw-Hill Companies Inc., 3rd Indian edition.
5. R. V. Hogg and E. Tanis (2001) **Probability and Statistical Inference** Pearson Education Inc. Asia.

HDG05. Human Embryology, Anatomy and Physiology

1. TW Sadler (2011) **Langman's Medical Embryology**, Lippincott Williams and Willkins Publication
2. S Standring (2008) **Gray's Anatomy**, Churchill Livingstone Publication, 40th Edition
3. E Hall (2010) **Guyton and Hall, Text book of Medical Physiology,** Saunders Publication, 12th Edition

HDG06. Science writing and presentation

1. N. D. V. Prasada Rao (1995) **Wren & Martin's High School English Grammar and Composition Book** S. Chand & Co. Ltd, Revised edition.
2. T. L. H. Smith Pearse (1997) **English Errors of Indian Students**, Oxford, 5th revised edition
3. J. Butcher and C Drake (2006) **Butcher's Copy-editing: The Cambridge Handbook for Editors, Copy- editors and Proofreaders**, Cambridge University Press; 4th edition.
4. P. Sebranek, V. Meyer and D. Kemper (1995) **Writers INC: A Student Handbook for Writing & Learning,** Great Source Education Group Inc.
5. R. A. Day (2011) **Scientific English: A Guide for Scientists and other Professionals,** Greenwood Press; 3rd revised edition.
6. M. Raman and S. Sharma (2012) **Technical Communication; Principles and Practice**, Oxford University Press, New Delhi; 2nd edition.
7. D. Carnegie (1980) **The Quick and Easy Way to Effective Speaking**, Simon & Schuster; Reissue edition.
8. J. V. Vilanilam (2000) **More effective communication; A manual for professionals**, Response Books (SAGE Publications); 1st edition.
9. Y.N. Bui (2013) **How to Write a Master's Thesis** Paperback, 2nd edition.
10. John Swales (1990) **Genre Analysis: English in Academic and Research Settings (Cambridge Applied Linguistics)** Series editors: Carol A. Chapelle and Susan Hunston, Cambridge University Press, 1st edition.
11. ONLINE SOFTWARE: https://www.rationaleonline.com

HDGP1. Lab Module I-Genetics

1. RJ Greenspan (2004) **Fly Pushing: The Theory and Practice of Drosophila Genetics,** Cold Spring Harbor Laboratory Press
2. S Chyb and N. Gompel (2013) **Atlas of Drosophila Morphology: Wild Type and Classical Mutants**, Academic Press, Elsevier
3. DS Henderson (2004) **Drosophila Cytogenetics Protocols**, Humana Press Inc.
4. PA Lawrence (1992) **The Making of a Fly: the Genetics of Animal Design,** Wiley Publications
5. C Dahmann (2010) **Drosophila: Methods and Protocols (Methods in Molecular Biology),** Humana Press Inc.
6. M Ashburner (2011) **Drosophila A Laboratory Handbook,** Cold Spring Harbor Laboratory Press
7. W Sullivan, M Ashburner, RS Hawley (2000) **Drosophila Protocols,** Cold Spring Harbor Laboratory Press

HDGP2. Lab Module II-Basic cell immmortalization techniques

1. Bird, A.G., Britton, S., Ernberg, I., *et al*. (1981) Characteristics of Epstein-Barr virus activation of human B lymphocytes. *J. Exp.Med.* **154**: 832-839.
2. Anderson, M.A., Gusella, J.F. (1984) Use of cyclosporin A in establishing Epstein-Barr virus-transformed human lymphoblastoid cell lines. *In Vitro*. **20**:856-858.
3. Neitzel H. (1986) A routine method for the establishment of permanent growing lymphoblastoid cell lines. *Hum Genet*.**73**:320-326.
4. Caputo, J.L., Thompson, A., McClintok, P., *et al*. (1991) An effective method for establishing human B lymphoblastic cell lines using Epstein-Barr virus. *J. Tissue Cult. Methods*. **13**: 39-44.
5. Wall, F.E., Henkel, R.D., Stern, M. P., Jenson, H.B., Moyer, M. P. (1995) An efficient method for routine Epstein-Barr virus immortalization of human B lymphocytes. *In Vitro Cell Dev Biol Anim*. **31**:156-159

HDGP3. Lab Module III-Human cytogenetics

1. JL Hamerton (2013) **Human Cytogenetics: Clinical Cytogenetics,** Academic Press
2. S Gersen and MB Keagel (2013) **The Principles of Clinical Cytogenetics,** Springer Science and Business Media Publication
3. Fan, Yao-Shan (2003) **Molecular Cytogenetics Protocols and Applications,** Humana Press
4. **Gersen**, Steven L., **Keagle**, Martha B. (2013) **The Principles of Clinical Cytogenetics,** Springer-Verlag New York
5. Marilyn S. Arsham (2017) **The AGT Cytogenetics Laboratory Manual**, Wiley-Blackwell
6. ISCN 2016: An International System for Human Cytogenomic Nomenclature (2016) Reprint of: Cytogenetic and Genome Research 2016 Karger Publishers
7. Susan Mahler Zneimer (2014) **Cytogenetic Abnormalities: Chromosomal, FISH, and Microarray**, Wiley-Blackwell
8. Faramarz Naeim MD (2013) **Atlas of Hematopathology: Morphology, Immunophenotype, Cytogenetics, and Molecular Approaches** Academic Press

****

SEMESTER2

HDG07. Biochemistry

1. DL Nelson and MM Cox (2013) **Lehninger, Principles of Biochemistry**, WH Freeman Publication, 6th Edition
2. D Voet and JG Voet (2010) **Biochemistry**, John Willey & Sons Publication, 4th Edition
3. T. Palmer (2004) **Enzymes: Biochemistry, biotechnology, clinical chemistry**; Affiliated East West Press Private Limited.
4. L. Stryer (2002) **Biochemistry**; W H Freeman & Co., 5th edition.
5. Nussey S, Whitehead S. **Endocrinology: An Integrated Approach**. Oxford: BIOS Scientific Publishers; 2001. Chapter 1, **Principles of endocrinology**. Available from: http://www.ncbi.nlm.nih.gov/books/NBK20/
6. Robert A. Weinberg, The Biology of Cancer, Garland Science; 2nd edition, 2013

HDG08. Molecular Biology

1. JD Watson (2013) **Molecular Biology of the Gene**, Pearson Publication, 7th Edition
2. B Alberts, A Johnson, J Lewis, M Raff, K Roberts and P Walter (2014) **Molecular Biology of the Cell**, Taylor and Francis Publication, 6th Edition
3. H Lodish, A Berk, SL Zipursky, P Matsudaira, D Baltimore and J Darnell (2012) **Molecular Cell Biology**, W. H. Freeman and Company, 7th Edition
4. JE Krebs, ES Goldstein and ST Kilpatrick (2012) **Lewin's Gene XI**, Jones and Bartlett Learning Publication, 11th Edition

HDG09. Principles of Development

1. SF Gilbert (2013) **Developmental Biology**, Sinauer Publication, 10th Edition
2. L Wolpert, C Tickle and AM Arias (2015) **Principles of Development**, Oxford University Press, 5th Edition

3. Jonathan M W Slack (2012), **Essential Developmental Biology,** Wiley-Blackwell, 3rd Edition.

HDG10. Bioinformatics

1. N. Gautham (2006) **Bioinformatics: Databases and Algorithms**; Alpha Science.
2. J. Bedell, I. Korf and M. Yandell (2003) **BLAST**; O'Reilly Press.
3. J. M. Keith (2008) **Bioinformatics Vol. 1, Data, sequence analysis & evolution**; Humana Press.
4. R. Durbin (1998) **Biological sequence analysis**; Cambridge University Press, 1998.
5. ONLINE COURSE: **ExPASy**: **http://www.expasy.org/**
6. web portal of multiple sources: **wws.startbioinfo.com**

Databases:

1. NCBI Genome Browser and databases: **http://www.ncbi.nlm.nih.gov/**
2. UCSC Genome Browser: **http://genome.ucsc.edu/**
3. Ensemble Genome Browser: **http://www.ncbi.nlm.nih.gov/**
4. Protein Catalogue ExPASy: **http://www.expasy.org/**
5. Protein Catalogue Uniprot: **http://www.uniprot.org/**

HDG11. Principles of Evolution and Population genetics

1. DL Hartl and AG Clark (2006) **Principles of Population Genetics,** Sinauer Associates Publication, 4th Edition
2. LL Cavalli-Sforza and WF Bodmer (2013) **The Genetics of Human Population,** Dover Publication
3. M Jobling, E Hollox, M Hurles, T Kivisild and C Tyler-Smith (2013) **Human Evolutionary Genetics**, Garland Science/Taylor and Francis Group Publication, 2nd Edition

HDGP5. Lab Module V-Biochemistry and Molecular Biology

1. K Wilson and J Walker (2010) **Principles and Techniques of Practical Biochemistry,** Cambridge University Press, 7th Edition
2. Protocols Online: **http://www.protocol-online.org/**
3. Sambrook and DM Russell (2001) **Molecular Cloning: a Laboratory Manual**. Cold Spring Laboratory Press Publication, 6th Edition
4. FM Ausubel (1990) **Current Protocols in Molecular Biology,** John Wiley and Sons Publication
5. Protocols Online: **http://www.protocol-online.org/**

HDGP6. Lab Module VI-Cell Biology

1. JS Bonifacino (2003) **Current Protocols in Cell Biology,** John Willey & Sons Publication
2. Protocols Online: **http://www.protocol-online.org/**
3. PA Lawrence (1992) **The Making of a Fly: the Genetics of Animal Design,** Wiley Publications
4. C Dahmann (2010) **Drosophila: Methods and Protocols (Methods in Molecular Biology),** Humana Press Inc.
5. M Ashburner(2011) **Drosophila A Laboratory Handbook,** Cold Spring Harbor Laboratory Press
6. W Sullivan, M Ashburner, RS Hawley (2000) **Drosophila Protocols,** Cold Spring Harbor Laboratory Press

HDGP7. LAB MODULE VII – BIOINFORMATICS

1. NCBI: **http://www.ncbi.nlm.nih.gov/**
2. EMBL: **www.embl.de/**
3. UCSC: **genome.ucsc.edu/**
4. collection of case studies and reviews at **wws.startbioinfo.com**
5. R. M. Holmes (2007) **A cell biologists' guide to modelling and bioinformatics**; Wiley Interscience.

****

SEMESTER 3

HDG12. Human Molecular Genetics

1. T Strachan and AP Read (2011), **Human Molecular Genetics**, Garland Science/Taylor and Francis Group Publication, 4th Edition.
2. For information on Mendelian phenotypes: **http://www.omim.org**
3. Access to Biomedical Literature: **http://www.ncbi.nlm.gov/entrez**

HDG13. Biochemical Genetics

1. CR Scriver, A Beaudet, WS Sly, D Valle, B Childs, K W Kinzler and B Vogelstein. **The Metabolic and Molecular Basis of Inherited Disease**, McGraw-Hill Publication, 8th edition, 2000.
2. Maureen E. Taylor and Kurt Drickamer. **Introduction to Glycobiology**, Oxford University Press, 3rd edition, 2011.
3. Varki A, Cummings RD, Esko JD, et al., editors. **Essentials of Glycobiology**. Cold Spring Harbor (NY): Cold Spring Harbor Laboratory Press; 3rd edition, 2017. Available from: http://www.ncbi.nlm.nih.gov/books/NBK310274/
4. Review articles published in journals such as *Nature Reviews Genetics*, *Journal of Biological Chemistry*, *Cell Metabolism*, *Nature Metabolism*, *Trends in Endocrinology and Metabolism*, *Journal of Inherited Metabolic Disease*, *Molecular Genetics and Metabolism*.
5. ***Book review***: Finger and Christ (2014) **Pearl Buck and phenylketonuria (PKU)**. *Journal of the History of the Neurosciences Basic and Clinical Perspectives*, Vol. 13, No. 1, pp. 44–57.
6. ***General book***: *The child who never grew*. Pearl S. Buck. Woodbine House; Reprint edition, 1992. ISBN-10: 0933149492, ISBN-13: 978-0933149496 (Original edition: 1950.)
7. ***Documentary film***: *The Forgotten Children*. https://www.espku.org/2016/06/28/the-forgotten-children-untreated-pku/ A documentary by the German Association for Phenylketonuria and allied inherited disorders (DIG PKU).
8. ***IEM in commercial cinema***: *Extraordinary measures* (2010). Directed by Tom Vaughan. CBS Films.

HDG14. Essentials in Immunology

1. T J Kindt, B A Osborne, R Goldsby (2006) **Kuby Immunology**, W H Freeman publication, 6th Edition
2. W E Paul (2003) **Fundamental Immunology**, Lipincott Williams and Wilkins publication, 4th Edition
3. P J Delves, S J Martin, D R Burton, I M Roitt (2017) **Roitt's Essential Immunology,** Wiley Blackwell publication, 13th edition.

HDG16. Use of Model Organisms to study human disease

1. SF Gilbert, AM Raunio, NJ Haver (1997) **Embryology: Constructing the Organism,** Sinauer Associates Inc. Publication
2. T Strachan and AP Read (2011) **Human Molecular Genetics**, Garland Science, Taylor and Francis Group Publication, 4th Edition
3. A Spradling, B Ganetsky, P Hieter, M Johnston, M Olson, T Orr-Weaver, J Rossant, A Sanchez, R Waterston (2006) **New roles for model genetic organisms in understanding and treating human disease: report from the 2006 Genetics Society of America meeting. *Genetics*. 172: 2025-2032.**
4. Online resource -Using Model organisms to study Health and Disease: **http://www.nigms.nih.gov/Education/Pages/modelorg_factsheet.aspx**
5. Online resource-Model organisms for Biomedical research: **http://www.nih.gov/science/models/**

HDGP9. Lab Module IX - Molecular Diagnostics

1. JL Hamerton (2013) **Human Cytogenetics: Clinical Cytogenetics,** Academic Press
2. S Gersen and MB Keagel (2013) **The Principles of Clinical Cytogenetics,** Springer Science and Business Media Publication
3. RL Nussbaum, RR. McInnes and HF Willard (2007) **Thomson and Thomson Genetics in Medicine,** Saunders, Elsevier Publication
4. LB Jorde, JC Carey and MJ Bamshad (2009) **Medical Genetics,** Elsevier Publication
5. S Heim, F Mitelman (2011) **Cancer Cytogenetics: Chromosomal and Molecular Genetic Aberrations of Tumor Cells,** John Willey and Sons Publications
6. L Buckingham (2011) **Molecular Diagnostics: Fundamentals, Methods and Clinical Applications**, FA Davis Company Publication, 2nd Edition
7. CA Burtis, D Bruns and ER Ashwood (2007) **Fundamentals of Molecular Diagnostics,** Saunders, Elsevier Publication
8. P George, GP Patrinos and WJ Ansorge (2010) **Molecular Diagnostics**, Academic Press, Elsevier Publication, 2nd Edition
9. NCBI Genome Browser and databases: **http://www.ncbi.nlm.nih.gov/**

HDG15 and HDGP10. Lab Module X - Genetic Counselling and Ethics

1. K Park (2011) **Park's Textbook of Preventive and Social Medicine,** Banarsidas Bhanot Publication, 21st Edition.
2. J Owen, J Punt and S Stranford (2013) **Kuby Immunology**, WH Freeman Publication, 7th Edition.
3. T Strachan and A Read (2011), **Human Molecular Genetics**, Garland
4. Science/Taylor and Francis Group Publication, 4th Edition.
5. Peter Turnpenny and Sian Ellard (Eds) (2012) **Emery's Elements of Medical Genetics**, Elsevier, 14th Edition.
6. R.J. McKinlay Gardner, Grant R Sutherland, and Lisa G. Shaffer (2011), **Chromosome abnormalities and Genetic counselling**, Oxford University Press, 4th Edition.
7. David L. Rimoin, Reed E. Pyeritz and Bruce Korf. (Eds.) (2013) **Emery and Rimoin's Principles and Practice of Medical Genetics**, Elsevier, 6th Edition.
8. Peter S Harper (2010), **Practical Genetic Counselling** Elsevier, 7th Edition.
9. Jean-Marie Saudubray, Georges van den Berghe, John H. Walter, (Eds.) (2012), **Inborn Metabolic Diseases: Diagnosis and Treatment,** Springer, 5th Edition.
10. T Smith (1999) **Ethics in Medical Research: A Handbook of Good Practice,** Cambridge University Press

11. I S Shergill, A Thompson and N Temple (2011) **Ethics, Medical Research, and Medicine: Commercialism versus Environmentalism and Social Justice,** Springer Science and Business Media Publication

****

SEMESTER4

HDG18. Cancer Biology

1. RA Weinberg (2012) **Biology of Cancer,** Garland, Taylor and Francis Group Publication, 2nd Edition
2. B Alberts, A Johnson, J Lewis, M Raff, K Roberts and P Walter (2008) **Molecular Biology of Cell**, Taylor and Francis group Publication, 5th Edition
3. V T DeVita, TS Lawrence and SA Rosenberg (2015) **DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology**, Wolters Kluwer Publication, 10th Edition
4. Relevant research articles, reviews and online resources

HDG19. Human Genome Organization

1. Alberts, Bruce. (2015) **Molecular biology of the cell**. New York, NY: Garland Science, Taylor and Francis Group.
2. Allis, C D., et al. (2015) **Epigenetics**. Cold Spring Harbor, New York: CSH Press, Cold Spring Harbor Laboratory Press.
3. Giardine, B. et al., (2005) 2005 Galaxy: A platform for interactive large-scale genome analysis Genome Res. 15(10): 1451–1455.
4. Brown, T. A. (2018) **Genomes**, New York, NY: Garland Science.
5. Krebs, Jocelyn E., Elliott S. Goldstein, and Stephen T. Kilpatrick. (2018) **Lewin's genes XII.** Burlington, MA: Jones & Bartlett Learning.
6. Mount, David W (2004) **Bioinformatics: sequence and genome analysis**. Cold Spring Harbor, N.Y: Cold Spring Harbor Laboratory Press.
7. Strachan, T, and Andrew P. Read. (2011) **Human molecular genetics**. New York: Garland Science.
8. Gibson, G and Spencer V. Muse (2009), A **Primer** of **Genome Science**, 2nd edition, Sinauer Associates Inc.

HDG21. Introduction to Disease Biology

1. DL Rimoin, RE Pyeritz, B Korf (2013), **Emery and Rimoin's Principles and Practice of Medical Genetics**, Elsevier Science Publication, 6th Edition.
2. Genes and Diseases (NCBI Bookshelf): **http://www.ncbi.nlm.nih.gov/books/NBK22185/**
3. L Buckingham (2011) **Molecular Diagnostics: Fundamentals, Methods and Clinical Applications**, FA Davis Company Publication, 2nd Edition
4. CA Burtis, D Bruns and ER Ashwood (2007) **Fundamentals of Molecular Diagnostics,** Saunders, Elsevier Publication
5. P George, GP Patrinos and WJ Ansorge (2010) **Molecular Diagnostics**, Academic Press, Elsevier Publication, 2nd Edition
6. NCBI Genome Browser and databases: **http://www.ncbi.nlm.nih.gov/**

****