

SYLLABUS FOR CREDIT BASED SEMESTER SYSTEM



M.Sc. HUMAN GENETICS – 2021

DEPARTMENT OF ZOOLOGY, BMT, HG AND WLB & C

University School of Sciences
Gujarat University,
Navarangpura,
Ahmedabad -380009
Phone: +91-79-26302362

M.Sc. IN HUMAN GENETICS

HUMAN GENETICS SYLLABUS FOR CREDIT BASED SEMESTER SYSTEM SEMESTER – I SYLLABUS EFFECTIVE FROM JUNE 2021

Sl. No.	Course Code	Name of the Course	Hours per Week	Internal Marks	External Marks	Total Marks	Credits
1.	HG-401	ANIMAL CELL CULTURE AND KARYOLOGY	3+1	30	70	100	04
2.	HG-402	ENZYMOLGY, ENERGY METABOLISM DISORDERS & PHYSIOLOGY	3+1	30	70	100	04
3.	HG-403	BIOLOGICAL TECHNIQUES	3+1	30	70	100	04
4.	HG-404	GENETICS & CYTOGENETICS	3+1	30	70	100	04
5.	HG-405PR	PRACTICAL -1	06	30	70	100	04
6.	HG-406PR	PRACTICAL - 2	06	30	70	100	04
		TOTAL	28	180	420	600	24

M.Sc. In Human Genetics

PROGRAMME OUTCOMES

The programme of master's in **Human Genetics**, imparting theoretical and experimental skills and enabling young minds to pursue higher studies and become teaching faculties in the Universities and Research Centers. This programme, with dissertation projects, imparts competent skills to thrive in research institutions and genetics and molecular Biology laboratories Pharmaceutical industries. Also at the end of course completion the students will be able to identify an area of significance in the form of project development, research and analysis in genetic diagnosis, counseling.

COURSE OUTCOME:

Semester I:

HG 401: The students would be able to learn comprehensive picture of cells, subcellular organelles, cell function, cell division and cell death. They will understand the basics of cell and tissue culture including infrastructure, equipments, materials and sterile technology and different cell based assay protocols.

HG 402: The students would be able to know metabolism and energy generating pathways and metabolism associated diseases in human. They will be also taught about human physiological process and its related diseases.

HG 403: The students would be able to learn various techniques like microscopy, electrophoresis, chromatography, spectrophotometry, centrifugation and different clinical instruments for diagnosis.

HG 404: The students would be able to learn basic understanding related to genetic and cytogenetics. They will also learn about transmission genetics, genetic mutations, chromosomal aberrations and Karyotyping.

HG 405 PR & HG 406PR : The students will attain on hand practical experiences of various topics covered in theory papers.

M.Sc. In HUMAN GENETICS SEMESTER – 1 JUNE 2021 ONWARDS

HG-401: ANIMAL CELL CULTURE AND KARYOLOGY

UNIT I: Cell Organelles:

Membranes of cells and transport across membranes; Energy generation in mitochondria and chloroplast; Cytoskeleton; General organization of protein transport within and outside the cell; Protein sorting and secretion; Mechanism of intracellular digestion; Structure and function of peroxisome, Nucleus, Nucleolus and biogenesis of ribosome.

UNIT II: Cell Junctions, Cell Cycle, Cell Death, Cancer and Stem Cells:

Cell adhesions, adherence, tight and gap junctions; Extracellular matrix and its association with cells; Cell cycle and its regulation; Cell death, apoptosis, necrosis, terminal differentiation and cell aging; Cancer, metastasis and angiogenesis; Stem cells, its types and application.

UNIT III: DNA, Chromatin and Chromosome:

Structure, properties, types of DNA and its isolation; Types and properties of histone and non-histone proteins; Nucleosome, Solenoid structure, Loop and domain, Organization of DNA in the chromosome; Types of chromosomes; Types and properties of heterochromatin; Lyon hypothesis; X-chromosome inactivation; Sex chromatin and its importance.

UNIT IV: Cell, Histotypic and Organotypic Culture of Animal cells:

Basics of cell culture; Culture environment, Sterilization techniques; Factors affecting cell culture, Contamination and its remedies; Serum and serum-free media; Primary and secondary cultures; Cell lines; Cell strains; techniques for creation of cell strains. Monolayer cultures; Suspension cultures; Cell synchronization and characterization; Measurement of cell growth; Scaling-up of animal cells in culture; Cryopreservation, Histotypic and organotypic cultures; 3-D cell culture.

HG-402: ENZYMOLOGY, ENERGY METABOLISM DISORDERS & PHYSIOLOGY

Unit-I: Enzymology:

Significance of Enzymes in Biological systems; Nomenclature and Enzyme Classification; Enzyme structure and function; Properties of Enzymes; Mechanism of Action of Enzymes; Enzyme Kinetics: Unisubstrate enzyme kinetics, K_m , V_{max} ; Linear Plots; Factors affecting Enzyme Activity; Reversible and Irreversible Enzyme Inhibition; Types of Inhibition; Allosteric enzymes; Immobilization and Industrial uses of enzymes; Ribozymes; DNAzymes; Metalloenzymes; Isoenzymes; Clinical Role of Enzymes.

Unit-II: Energy Generation and Associated Diseases:

Reactions, regulation and genetic disorders of glycolysis, Krebs's cycle, Pentose phosphate pathway, Fatty acid oxidation and Glycogen metabolism, Glucose transporters, Diabetes and its effects on the body.

Unit-III: Circulatory, Respiratory and Excretory System:

Cardio-vascular physiology; Functions & Disorders of Circulatory System; Respiratory physiology; Lung volumes and capacities; Control of respiration; disorders of the respiratory system; Role of body fluids; Homeostasis; Structure and function of kidneys; Concentration of urine; Control of urine formation; Disorders of the urinary system.

Unit-IV: Digestive, Nervous and Muscle System:

Physiology of food intake; Digestion and absorption of Carbohydrates; Proteins and Fats. Control and components of secretions in the oral cavity, stomach and intestine; Gastric motility and emptying; Cells and organization of the Nervous system, Nerve impulse transmission; EPSP and IPSP; Neurotransmitters, Cells and organization of the Muscular system; Physiology of Skeletal, Smooth and Cardiac Muscles; Neuromuscular junctions; Muscle contraction and energetic; Neuromuscular Disorders.

HG-403: BIOLOGICAL TECHNIQUES

Unit-I: Microscopy:

Principle, working and types of light microscope: Bright-field, Dark-field, Phase-contrast, Fluorescence, Confocal, Atomic Force Microscopy; Principle and working of electron microscope – SEM and TEM; Freeze-etch and Freeze fracture technique for EM; Image analysis for microscopy.

Unit-II: Electrophoresis and Centrifugation:

Principle of Electrophoresis; Types of electrophoresis: Agarose gel electrophoresis, polyacrylamide gel electrophoresis, Isoelectric focusing and 2D-PAGE, Pulse field gel electrophoresis, Tube gel electrophoresis, Capillary gel electrophoresis; Factors affecting electrophoresis.

Principle of centrifugation; Types of rotors; Types of centrifuges; Types of centrifugation techniques: Differential, Density gradient and Analytical ultracentrifugation.

Unit-III: Chromatography and Photometry

History and introduction of separation techniques; Fundamentals of chromatographic separation, Chromatographic techniques; Methods of separation and detection; Hyphenated techniques - HPLC, HPTLC, FPLC, UPLC, etc.,

Principle and applications of spectroscopic techniques – UV-Vis; IR; NMR; MS; MALDI-ToF etc.

Unit-IV: Clinical Instrumentation

Biomedical recorders (ECG, EEG, EMG); Sphygmomanometer; Pulse Oximeter; Blood Glucose Monitoring apparatus; Imaging Instrumentation for clinical diagnosis; Sonography, Endoscopy, Ventilator, Dialysis machine, Thermometers, Automated blood and cell analysers.

HG-404: GENETICS & CYTOGENETICS

Unit I Mendelian genetics and its extension

Mendel and his experiments; Derived concepts and Laws; Mendelian principles of inheritance: significance and limitations. Chromosome theory of inheritance; Various experimental Crosses: Back cross, Test cross, Reciprocal cross, Hybrid cross.

Extensions of Mendelian principles; Allelic variation and multiple alleles; pseudo-alleles; test of allelism; non-allelic interaction of genes; Pleiotropy and Pleiotropic genes and their relevance; Linkage & crossing over.

Unit II Inheritance and Pharmacogenetics

Pedigrees, Inheritance – autosomal, dominant, recessive, Sex-linked, Sex-limited and sex-influenced traits; mechanism of sex determination; Mitochondrial inheritance; Polygenic inheritance; Genetic components and susceptibility in multifactorial disorders; Pharmacogenetics and ecogenetics.

Unit III Basic Cytogenetics

History and development of human cytogenetics; Centromere and Kinetochore; Telomere and its maintenance; Banding techniques for pericentric heterochromatin, intercalary heterochromatin, telomeric heterochromatin and nucleolar heterochromatin; FISH; Array CGH.

Unit IV Human Cytogenetics

Idiogram; Karyotype; Cytogenetic nomenclature; Numerical and Structural aberrations; Chromosomal disorders: Structural and numerical; Autosomal/sex chromosomal/sex reversal; Mechanisms – mitotic/meiotic non-disjunction/ chromosomal rearrangements; Chromosome instability and associated syndromes; genetics of fetal loss.

HG – 405 PR:

Practicals based on the theory papers HG-401 and HG-402 and Industrial/ Laboratories visit.

HG – 406 PR:

Practicals based on the theory papers HG-403 and HG-404 and Industrial/ Laboratories visit.

LIST OF REFERENCE BOOKS

1. Becker's World of the cell.- Jeff Hardin and Gregory Bertoni.
2. Karp's Cell and Molecular Biology Concepts and Experiments.- Gerald Karp, Janet Iwasa and Wallace Marshall.
3. Cell and Molecular Biology.- E.D.P. DeRobertis and EMP De Robertis Geoffrey Cooper.
4. Molecular Biology of the Cell.- Bruce Alberts et al.,
5. Animal cell Culture:Essential methods.- John M. Davis(Ed).
6. Culture of Animal cells:A manual of basic technique and specialized applications.- R. Freshney.
7. Animal cell culture and Technology.- Michael Butler.
8. Animal cell culture.- Mohd. Al-Rubeai(Ed).
9. Animal cell culture.- John R.W. Masters(Ed).
10. Textbook of Physiology.- George Howard Bell, Donald Emslie-Smith, Colin Ralston Paterson.(Eds).
11. Human Physiology, Biochemistry and Basic Medicine.- Laurence cole , Kramer, Peter.
12. Physiology and Biochemistry in modern medicine.- John Macleod.
13. Principles of Biochemistry.- Albert L. Lehninger, David L. Nelson and Michael M. Cox.
14. Textbook of Medical Physiology.- John E. Hall and C. Guyton.
15. Tortora's Principles of Anatomy and Physiology.- Gerard J Tortora.
16. Fundamentals of Enzyme kinetics.
17. Understanding the control of metabolism.- David Fell.
18. Fundamentals of Enzymology.- Nicholas C. Price and Lewis Stevens.
19. Wilson & Walker's Principles and Technique of Biochemistry and Molecular Biology.
20. Instrumental Analysis in the Biological Sciences.- Gordon M.H.
21. Human Cytogenetics.- John L. Humerton.
22. Human Cytogenetics: A Practical approach.- D.E. Rooney, B.H. Czepulkowski.
23. Fundamentals of Enzymology-The Cell and Molecular Biology of Catalytic Proteins.- Price, N. C. and Stevens, L.

24. Enzymes: Biochemistry, Biotechnology and Clinical Chemistry.-Bonner, P.L. and Palmer, T.
25. Enzymes: Catalysis, Kinetics and Mechanisms.- Punekar, N. S.
26. Harper's Illustrated Biochemistry- Rodwell, V.W., Bender, D. Botham, K.M. et al.
27. Biochemistry.- Satyanarayana, U. and Chakrapani, U.
28. Biochemistry & Metabolism.- Davison, A., Milan, A. Phillips, S. and Ranganath, L.
29. Molecular cell biology.- Harvey Lodish, etal.
30. Techniques in microscopy and cell biology.- Sharma, V. K.
31. A manual of laboratory experiences in cell biology.- Gasque, C.E.
32. Electrophoresis in practice: A guide to methods and applications of DNA and protein separations.- Westermeier, R.
33. Electrophoresis: theory, methods, and applications.- Bier, M. (Ed.).
34. Centrifugal separations in biotechnology.- Leung, W. W. F.
35. Centrifugal Separations in Molecular and Cell Biology.- Birnie, G. D. and Rickwood, D. (Eds.).
36. Encyclopaedia of Separation Science.- Smith, R. M.

SYLLABUS FOR CREDIT BASED SEMESTER SYSTEM



M.Sc. in HUMAN GENETICS – 2021

DEPARTMENT OF ZOOLOGY, BMT, HG AND WLB & C

University School of Sciences,
Gujarat University,
Navarangpura,
Ahmedabad -380009
Phone: +91-79-26302362

M.Sc. IN HUMAN GENETICS

SYLLABUS FOR CREDIT BASED SEMESTER SYSTEM

SEMESTER – II

EFFECTIVE FROM JUNE 2021

Sl. No.	Course Code	Name of the Course	Hours per Week	Internal Marks	External Marks	Total Marks	Credits
1.	HG-407	GENETICS AND IMMUNOLOGY	3+1	30	70	100	04
2.	HG-408	GENETIC ENGINEERING and ADVANCED MOLECULAR TECHNIQUES	3+1	30	70	100	04
3.	HG-409	CLINICAL and MOLECULAR GENETICS	3+1	30	70	100	04
4.	HG-410	GENETIC COUNSELING and RADIATION BIOLOGY	3+1	30	70	100	04
5.	HG- 411PR	PRACTICAL -1	06	30	70	100	04
6.	HG- 412PR	PRACTICAL - 2	06	30	70	100	04
		TOTAL	28	180	420	600	24

M.Sc. In Human Genetics

PROGRAMME OUTCOME:

The curriculum and syllabus outlined for the Masters degree in Human Genetics has been designed with emphasis on elucidating the structure and complexity of the human genome and the variations that result in genetic disorders. This syllabus is thus aimed at guiding a post graduate student in Human Genetics towards a career as a geneticist in clinical genetic diagnosis and genomic analysis. The focus is to develop a student's ability to carry out chromosomal/molecular analysis of various genetic anomalies, with an in-depth knowledge of genetics and a good practical grasp of the state-of-the-art technologies. A student of this course would therefore be prepared and equipped for the analytical thinking requisite for a career in human clinical, molecular genetics and genomic analysis.

COURSE OUTCOME: Semester II

HG 407:

The module includes topics in genetics and immunology. The student would gain information on the molecular organization of the human genome, gene complexities and mutations leading to disorders. The course would also cover molecular processes, gene regulation mechanisms, and epigenetics- topics which are vital to enhance the understanding of gene expression and its variation. Topics under this course would consequently give the student a comprehensive understanding of the basic molecular mechanisms that manifest diverse control and regulation.

Topics in Immunology would help the student to learn about the details of the immune system, its function, immune responses and immune related disorders which have an immense bearing on the diseases that affect human beings. An in-depth working knowledge and laboratory experience in the various Immunotechnologies would fortify the student's opportunity for research and laboratory analysis, which is a vastly expanding area of medicine, since innumerable clinical conditions and diseases are based on immune attack and dysfunction.

HG 408:

In an era driven by DNA analysis, genetic engineering and advanced molecular technologies is a cornerstone for reinforcing a student's knowledge in human genetics, in the context of current human need in medical genetic diagnosis. A postgraduate student of Human Genetics would therefore develop a deeper understanding of the principles, methodology, practical applications and trouble-shooting modalities in several molecular analytical technologies, such as genetic engineering, PCR, gene sequencing methods, etc.. This training is extremely pertinent for a student who wishes to pursue a career in human genetics and molecular analysis.

HG 409

This module provides the student of Human Genetics, composite and complete detailed knowledge of various human genetic disorders, systematically organized in relation to the organ/system. In addition to learning the technologies to diagnose a disorder as mentioned in the earlier course paper, a student would also gain complete, comprehensive details of several commonly occurring genetic disorders. It is imperative for a student to understand the underlying gene/chromosomal/epigenetic causes of a genetic anomaly in order to develop novel strategies for the management of such disorders in the future.

HG 410

The course curriculum in Human Genetics has specifically included genetic counseling as a vital part of the syllabus as it is a major clinical complement to the analytical and diagnostic work-up for a patient with a genetic abnormality. A student in this field requires firm grounding in the intricacies of genetic counseling in order to manage genetic disorders in a professional and appropriate manner. This would mold the student in the right direction for a successful career as a Genetic Analyst or a Geneticist.

The course also includes important topics in Radiation Biology, since the use of radioactive isotopes in diagnosis and therapy are pivotal elements in medical diagnosis and therapy. A student of Human Genetics should consequently be aware of the technical uses, handling, precautions and exposure effects of radiation based analyticals. In addition a student should also be aware of radiation induced mutations and the implications of radiation genetics.

HG 405 PR & HG 406PR : The students will attain practical experiences of various topics covered in theory papers.

M.Sc. In HUMAN GENETICS SEMESTER – II JUNE 2021 ONWARDS

HG-407: GENETICS AND IMMUNOLOGY

Unit-I: Genetics-I

Mutation and mutagenesis; DNA repair mechanisms; Hereditary diseases caused by defective DNA repair; C value; Complexity of genomes; Split genes; non-coding genes; Overlapping genes; Multi-gene families; Pseudogenes; Overlapping sequences; Unique sequences, Repetitive sequences - satellite DNAs and interspersed repeated DNAs; LINES; SINES; Alu family; Transposons

Unit-II: Genetics-II

Population genetics; DNA replication; Transcription and Splicing; Regulation of gene expression in prokaryotes (negative and positive control of *lac* operon, *trp* operon and its attenuation) and in eukaryotes; Epigenetics

Unit-III: Immunology-I

An Introduction to the Immune System; Immune response; Antigens; Antibodies; Major histocompatibility complex; Primary and secondary immunodeficiencies; Allergy and hypersensitivity; Autoimmunity; Immunosenescence; Cancer immunology

Unit-IV: Immunology-II

Immunological diagnostic techniques/Immunochemical techniques; Immunoassays; Immunoelectrophoresis; Future of Diagnostic Immunology; Immunocapture polymerase chain reaction; Immunotherapy; Immunosuppressive therapy; Transplantation; Environmental allergy and allergotoxicology; Immunotoxicity testing

HG-408: Genetic Engineering and Advanced Molecular Techniques

Unit-I: Genetic engineering - I

Enzymes used in DNA technology – nucleases, polymerases, ligase, kinases and phosphatases; Vectors – plasmids, phages, cosmids, artificial chromosomes, shuttle vectors and expression vectors; Construction of genomic and cDNA libraries; Applications of recombinant DNA technology; GMOs

Unit-II: Genetic engineering - II

Expression and interaction-based screening of colonies; Principles of hybridization and hybridization-based techniques; CRISPR and Cas9 based gene edition; Recombination-based cloning; Molecular interaction assays including two hybrid assays; Transformation and transfection methods; Promoter characterization and mutagenesis

Unit-III: Advance molecular biology techniques - I

Isolation of DNA, RNA and plasmid; Polymerase chain reaction and its general applications; Dye and probe based detection in real time PCR; Gene expression study and data analysis in qPCR; DNA fingerprinting; DNA foot-printing

Unit-IV: Advance molecular biology techniques – II

RFLP and restriction mapping; Southern blotting; Northern blotting; Western blotting; Dot Blot technique; DNA sequencing by chain termination method and chemical degradation method; Capillary sequencing technique; Gene transfer techniques

HG-409: Clinical and Molecular Genetics

Unit-I: Single genetic disorders

General and monogenic (Single gene) disorders; Prevalence estimations for Single gene disorders; Autosomal dominant disorders; Autosomal recessive disorders; X-linked dominant and recessive disorders; Rare monogenic disorders; Complexity factors influencing monogenic inheritance; Gene penetrance and expressivity, Genetic heterogeneity, Phenocopies, Mechanisms and disorders due to uniparental disomy; Genomic imprinting; Modifier genes and monogenic disorders; Identification of genetic modulators (GENDULF algorithm); OMIM database

Unit-II: Disorders of eye, mitochondria and multifactorial disorders

Mitochondrial disorders; Ocular disorders such as cataract, glaucoma, macular dystrophy, color blindness, etc; Multifactorial disorders such as diabetes, obesity, atherosclerosis and hyperlipidemia

Unit-III: Neuromuscular disorders

Neurogenetic Disorders; Muscle Disorders; Genetics of psychiatric diseases (Schizophrenia, mood disorders); Disorders of childhood; Concept of biochemical and molecular tests; Clinical importance of biochemical and molecular tests; Understanding qualitative and quantitative indices; Qualitative and quantitative estimation of various diagnostic markers

Unit-IV: Haemopoietic, skin, skeleton and reproductive disorders

Molecular genetics of hematopoiesis and its disruption; Blood disorders such as thalassemia, sickle cell anemia, hemophilia; Genetic diseases of skin and skeleton; Pseudohermaphroditism; True hermaphroditism; Gonadal dysgenesis; Anomalies of genital ducts; Genetic basis of male and female infertility; Disease etiology – non-genetic determinants

HG-410: Genetic Counseling and Radiation Biology

Unit-I: Genetic counseling-I

Patterns of inheritance: classical and non-classical; Overview of genetic counseling; Components of genetic counseling; Context and situations in genetic counseling; Information gathering; Construction of pedigrees and their interpretation

Unit-II: Genetic counseling-II

Ethics of genetic counseling; Psychosocial counseling – psychosocial assessment, structure of session, working alliance; Patient education – information giving and barriers in communication; Risk communication and decision making; Medical documentation

Unit-III: Radiation biology - I

Radioactive isotopes, types, properties and application in nuclear medicine and research; Radioactivity, specific activity, radioactive decay and half-life; Modes of radioactive decay; Sources of Exposure; Types and properties - electromagnetic and particulate forms (α , β , η , protons, positrons); Radiation dosimetry, Measurement modalities and Units of Radiation; Ionizing radiation: types, uses and properties of X-rays; Varied diagnostic applications of X-rays; Properties of gamma rays. Interaction of radiation with matter; Non-Ionizing radiation: Types, sources, uses, properties and biological effects of ultraviolet radiation; Infrared radiation, microwaves and radiofrequency radiation

Unit-IV: Radiation biology - II

Law of radiosensitivity; Differential radiosensitivity; By-stander effect; Effects of ionizing radiation: Effects on biomolecules, cell and cell organelles; Effects on various organs and organ systems; Radiation genetics; Radiation syndromes; Radiation and cancer; Radiotherapy: external beam radiotherapy and brachytherapy; Radioprotective agents; Radiosensitizing agents; Radiation hormesis; Precautions and ICRP safety rules in handling radiation/radioactive isotopes

HG – 411 PR:

Practicals based on the theory papers HG-407 and HG-408 and Industrial/ Laboratories visit.

HG – 412 PR:

Practicals based on the theory papers HG-409 and HG-410 and Industrial/ Laboratories visit.

REFERENCE BOOKS

- Lewin's Genes XII by Krebs, Goldstein and Kilpatrick by Jones and Bartlett Learning
Synthetic Biology by Meyers, Wiley – Blackwell
Mishra and Alok, A handbook of research methodology by Educreation Publication
Arumugam, Research Methodology for life Sciences by Saras publication
Punt, J.; Jones, P.; Owen, J. A. & Stranford, S. (2018). Kuby Immunology. United Kingdom: Macmillan Learning.
Wilson, K.; Hofmann, A.; Walker, J.; Clokie, S. (2018). Wilson and Walker's Principles and Techniques of Biochemistry and Molecular Biology. United Kingdom: Cambridge University Press.
Paul, W. E. (2013). Fundamental immunology. Philadelphia: Lippincott Williams et Wilkins.
Murphy, K. M. & Weaver, C. (2017). Janeway's Immunobiology. United Kingdom: Garland Science.
Latha, P. M. (2012). A Textbook of Immunology. India: S. Chand & Company.
Wilson, K.; Hofmann, A.; Walker, J. & Clokie, S. (2018). Wilson and Walker's Principles and Techniques of Biochemistry and Molecular Biology. United Kingdom: Cambridge University Press.
Tortora, G. J. & Grabowski, S. R. (2017). Principles of anatomy and physiology. New York, NY: HarperCollins College.
Praful B. Godkar; Darshan P. Godkar. (2021). Textbook of Medical Laboratory Technology - Vol 1 and 2. Bhalani Publishing House.
Uhlmann, W.; Schuette, J. & Yashar, B. (2011). A Guide to Genetic Counseling. Somerset: Wiley-Blackwell.
Veach, P.; LeRoy, B. & Bartels, D. (2006). Facilitating the genetic counseling process. Springer
Usha, D. & Dhanlaxmi, S. (2022). Genetic Counseling Clinical and Laboratory Approach. Atithibooks
Sambrook and Russell. 2001. Molecular Cloning. Coldspring Harbour
Pasternak. 2000. An introduction to molecular human genetics. Fitzgerald.
Wilson and Walker. 2000. Practical biochemistry. Principle and techniques. Cambridge.
Griffith's et al., 2004. An introduction to genetic analysis. Freeman.
Brown. 2001. Essential Molecular biology (Volume I & II). Oxford University Press.
Glick and Pasternak. 1990. Molecular biotechnology. ASM press.
Miesfield. 1999. Applied Molecular Genetics. Willey.
Griffith et al. An introduction to Genetic analysis. Published by Freeman
Jocelyn E. Krebs, Elliott S. Goldstein, Stephen T. Kilpatrick. Lewin's GENES XII. Published by Jones and Bartlett learning.
D. Peter Snustad, Michael J. Simmons. Principles of Genetics.
W S Klug & M R Cummings. Concepts of Genetics.
R E Pyeritz, B R Korf, W W Grody (Ed.). Emery & Rimoin's Principles & practice of medical genetics & Genomics. Academic Press.
R F Mueller & I D Young. Elements of Medical Genetics. Published by Churchill Livingstone
J M Connor & M A Ferguson-Smith. Essential Medical Genetics. Blackwell publishers
W. R. Uhlmann, J. L. Schuette, B. M. Yashar. (Ed.) A Guide to Genetic counseling. Wiley-Blackwell publication.
P S Harper. Practical genetic counseling. Published by Butterworth Heinemann

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

**M.Sc. Human Genetics, Semester-III
June, 2022 Onwards**

SR. No.	Course Code	Name of the Course	Hours per Week	Internal Marks	External Marks	Total Marks	Credits
1	HG – 501	BIostatistics, Bioinformatics AND RESEARCH METHODOLOGY	3+1	30	70	100	04
2	HG - 502	DEVELOPMENTAL BIOLOGY, CANCER AND MOLECULAR MEDICINE	3+1	30	70	100	04
3	HG – 503EA	CELL BIOLOGY, CYTOGENETICS AND MOLECULAR BIOLOGY-I	3+1	30	70	100	04
4	HG – 503EB	ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY – I					
5	HG – 503EC	GENETIC COUNSELING – I					
6	HG – 503ED	TOXICOLOGY-I					
7	HG – 504EA	CELL BIOLOGY, CYTOGENETICS AND MOLECULAR BIOLOGY-II	3+1	30	70	100	04
8	HG – 504EB	ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY – II					
9	HG – 504EC	GENETIC COUNSELING – II					
10	HG – 504ED	TOXICOLOGY-II					
11	HG - 505PR	PRACTICAL - I	6	30	70	100	04
12	HG - 506PR	PRACTICAL - II	6	30	70	100	04
		Total	28	180	420	600	24

PROGRAMME OUTCOME

The program and syllabus designed for the third semester for the Master's degree in Human Genetics has been framed to provide the student with the essential pre-requisites for the next step in research, for carrying out a dissertation or for pursuing Doctoral research. This would equip a student with the required knowledge of all aspects related to carrying out intrinsic experimental research and analyze and process the data as per current standards. The syllabus designed for the third semester for the Master's degree has been formulated to offer the student an elective paper with promise for future career opportunities. This would enable a student to obtain theoretical and practical knowledge in an area which would have wide applicability in securing a good career option and help the student fulfill their career goals.

HUMAN GENETICS COURSE OUTCOME

Human Genetics – 501 & 502

This Unit includes topics in Biostatistics. It is well-accepted that no research is accepted or can be validated without substantiation by correctly used Statistical methods. A student equipped with the theory and practical aspects of this curriculum would have a good foundation in precise Sampling and Data Collection, have the ability to represent data effectively and carry out the requisite Descriptive as well as Analytical Statistics to ensure accurate Scientific standards. A student would be able to carry out the Statistical Analysis for both parametric and non-parametric data. A firm foundation in Biostatistics would thus prepare a student for research associated with Industry or Medical or Biological fields.

This Paper also provides a student with the basic concepts in Bioinformatics. basic concepts of biological databases, Resources and Tools. A student would thus be familiar with the use of related software, various Tools for Bioinformatics, Sequence alignment, exploring Sequence & Structure Databases and the applications of Bioinformatics. This knowledge would extremely useful vocations in any Organization or Institution dealing with Molecular Analysis, which is currently ubiquitous in Medical, Forensic Sciences, Pharmaceutical, Biotechnological and several other fields.

The inclusion of Research Methodology is specifically with the forethought of moulding a student for future research. With the discontinuation of the M.Phil. degree, it is now imperative to add this module in the Master's degree syllabus. With this unit a student will comprehend and accurately carry out Experimental design in Research, Measurements and Experimentation with Scientific logic, setting hypothesis with proper rationale and data interpretation with analytical reasoning. This curriculum will also guide a student in the workings for development of pre-proposal and full proposal, Scientific writing and documentation, Ethics in Science and all regulatory processes for Laboratory accreditation. Students will learn about different aspects of Developmental Biology such as gene expression during development, stem cells in development as well as cytological, genetical and chemical basis of development. The students would be able to learn cancer biology as the comparative study of normal versus cancerous cells and tissues. The discipline includes the study of normal and tumor cell biology, molecular biology of cancer, rationale approach for cancer diagnosis, prevention and treatment using new technologies with translational research. Students will also have great exposure on topic such as Molecular Medicine. Understanding of fundamentals, approaches and future of molecular medicine will help students advancing their career in research field.

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

CELL BIOLOGY, CYTOGENETICS AND MOLECULAR BIOLOGY 503 & 504

There have been unprecedented technical advancement in genetics and molecular biology which is continuing and holds a lot of promise. Hence, this course has been designed to include all the basic as well as new emerging techniques in this field. Conventional tools such as pedigree analysis, chromosome identification, Banding techniques etc. have been included which still remain a powerful starting tool. Emphasis has also been given to Cancer Biology and toxicology. Furthermore, students will be taught other important topics of genetics including Human Genome Project, DNA repair, Toxicity and toxicity testing, cell signalling and communication etc. Students will also become proficient in new areas such as Nanotechnology and stem cell biology. In this module, the students will be familiar with the science of toxicology, including hazard identification, exposure assessment, dose-response assessment, and an understanding of the mechanisms of action and effects of toxic chemicals at multiple levels of biological organization. They will also learn technical aspects and experimental approaches in toxicological research, testing, and risk assessment. Students will also learn about molecular techniques such as PCR, sequencing, gene silencing, gene expression analysis using microarray. Our goal is to provide students a broad understanding of all these recent techniques which are important for advanced biological research. We will also be giving students the comprehensive idea of gene therapy to cure the disease which will help them in future career development.

ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY 503 & 504

The course covers details of embryonic development of the Endocrine and Reproductive system and its regulation, as well as anatomical, biochemical, molecular and functional details of the Endocrine organs, male and female reproductive systems. These topics give a student the background and foundation needed for pursuing a research or investigation (diagnostic) career in Endocrinology, Fertility regulation, control and management. The curriculum of the specialization on Endocrinology and Reproductive Technology is designed to provide students with in-depth knowledge of various aspects of Endocrinology and Reproductive Technology. This paper also confers knowledge and practical skill related to Endocrine and Reproductive Technologies, which includes Molecular technologies, Bioassays, Immunoassays and specific laboratory based *in-vitro* technologies. With the growing focus on fertility regulation and management, this is a major thrust area for research and diagnosis.

Genetic Counselling 503 & 504

Genetic counselling is progressing as a subject of great significance today due to rapid and revolutionary advancements in Human Genetics. Hence, these two papers have been designed in a manner to develop a comprehensive knowledge and proficiency apt for Genetic Counselling. Also, care is taken to further hone the practice-based competencies in the students to manage a genetic counselling case before, during, and after the clinic visit. The syllabus includes detailed aspects of pedigree analysis, risk assessment, prenatal diagnosis, genetic testing and interpretation etc. The ethical, legal and social issues arising from developments in Human genetics and the influences on Genetic counselling are also included.

TOXICOLOGY 503&504

In this module, the students will be familiar with the science of toxicology, including hazard identification, exposure assessment, dose-response assessment, and an understanding of the mechanisms of action and effects of toxic chemicals at multiple levels of biological organization. They will also learn technical aspects and experimental approaches in toxicological research, testing, and risk assessment. They will also get the understanding of how the toxic chemicals harms the specific organs, the kinetics of those hazardous toxicants and how they create certain diseases after getting digested in the body. This course will also provide the platform for those students who want to build their future in pharmaceuticals by understanding the basics of guidelines for animal house facilities and its formation. In this module, the students will be familiar with the science of toxicology, including hazard identification, exposure assessment, dose-response assessment, and an understanding of the mechanisms of action and effects of toxic chemicals at multiple levels of biological organization. They will also learn technical aspects and experimental approaches in toxicological research, testing, and risk assessment. They will get to learn the important toxicological aspect which is ecological or environmental toxicology in which they will understand how the aquatic and terrestrial nature is getting harmed by toxicants and the tests to assess it. This course will also provide the platform for those students who want to build their future in pharmaceuticals by understanding the basics of guidelines and quality standards for animal house facilities and animal experimentation along with applications of Toxicology.

HG– 501: Biostatistics, Bioinformatics and Research Methodology

UNIT-I: Biostatistics-I

Data Collection, Representation and Sampling:

Types of Data: Qualitative, Quantitative Data: Discrete and Continuous. Understanding of Population, Sampling and Sampling methods, Methods Data Collection, Grouping of data, Frequencies: Frequency Distributions, Relative Frequencies. Tabulation, Graphical Presentation

Descriptive Statistics:

Measures of Central Tendency-concept; Mean, mode, median for ungrouped and grouped data, Measures of Dispersion: Absolute and Relative measures; Range, Interquartile Range, Variance, Standard Deviation, Standard Error and Coefficient of Variation, Probability: Basic concepts of Probability, Empirical, Theoretical Probability. Addition Rule, Product law. Applications of Probability, Probability Distributions: Binomial, Normal Distribution, Measures of Skewness and Kurtosis

UNIT-II: Biostatistics-II

Tests of Hypotheses:

Parametric, Non-Parametric data, Statistical hypotheses: Null and Alternative hypotheses. Simple and Composite hypotheses. Statistical Tests: Acceptance and Rejection Norms. Sampling Distributions - t, chi-square and F distributions. Significance Tests for Normal Distribution: One sample test (unmatched) – z test and t-test and scores; Confidence Intervals. Bivariate and Multivariate Analysis: Correlation: Methods for Correlation analysis, Pearson's Correlation Coefficient 'r', Rank Correlation, Regression: Linear regressions. Chi Square Analysis, two sample tests for normal distributions: Tests for Means when variances are known or unknown. Paired t-test for equality of means. Student's t test; Analysis of Variance: One-way, Two-way (parametric) ANOVA. Tukey's test for two-way ANOVA, Mann-Whitney U test, Wilcoxon matched pairs tests

UNIT- III: Bioinformatics

Introduction to Bioinformatics, Basic concepts of biological databases, Gene and Protein Databases and Resources (NCBI, EBI, ExPasy, Entrez, RCSB) - NCBI Databases and Tools for Bioinformatics, Sequence alignment - BLAST, FASTA, PSI-BLAST algorithms, Primary & Secondary Sequence & Structure Databases (Genbank, SwissPort/Uniport, EMBL, MMDB & KEGG) and its application, Primer designing.

UNIT -IV: Research Methodology and regulation

Scientific Logic, Biological Thoughts, Methods of Reasoning in Research, Principles of measurements and experiment, Impact assessment and Management, Role of criticism in sciences, Conceptualization and development of research plan, Development of pre-proposal and full proposal, Writing of scientific and popular articles, Case studies, Ethics in Science, Good laboratory techniques, Intellectual property rights, Laboratory accreditations

HG-502: Developmental Biology, Cancer and Molecular Medicine

UNIT – I: Developmental Biology - I

Patterns and process of development (Fate maps and Gradients)
Differential gene expression during development
Major model organisms
Stem cells in development

UNIT – II: Developmental Biology - II

Building with Ectoderm, Mesoderm and Endoderm
Cytological, genetical and chemical basis of development
Cell-cell communication during development

Unit-III: Advances In Cancer Biology

Cell transformation and tumorigenesis, Familial cancers, Chromosomal aberrations in cancer, Genetic, Epigenetic modifications, telomerase activity, Microsatellite instability in cancer, Genetic predisposition to sporadic cancer syndrome, Tumor specific markers, Cancer Genome Atlas Project, Mitelman Database of chromosome, Advancement in Genomic landscaping of cancer.

UNIT – IV: Molecular medicine

Introduction to molecular medicine, Molecular pathological epidemiology, Molecular pathology, Approaches and methods in molecular medicine, Future of molecular medicine.

ELECTIVE PAPER - I
CELL BIOLOGY, CYTOGENETICS AND MOLECULAR BIOLOGY

HG-503EA - Cell Biology, Cytogenetics And Molecular Biology-I

Unit-I: Human cytogenetics

Pedigree construction - inheritance patterns and risk assessment, Types and structure of chromosomes, Chromosome identification, ISCN, Banding techniques and karyotyping, FISH and mFISH, Spectral karyotyping, CGH, Chromosomes & diseases, chromosomes and cancer, Chromosomes and evolution, Prenatal diagnosis

Unit-II: Gene Expressions & Regulations

DNA modification & restriction, DNA repair mechanism, DNA repair genes and genetic instability, hereditary diseases caused by defective DNA repair, Regulation of gene expression in eukaryotes, Isolation of gene, Understanding Human Genome Project – organization, gene families

Unit-III: Cancer Biology

Hallmarks of cancer, Mutagens, Carcinogens, Oncogenes and Tumor suppressor genes, Cell transformation and tumorigenesis, Cell cycle checkpoint and cancer, Telomerase and cancer, Angiogenesis and Metastasis, Familial cancers, Chromosomal aberrations in cancer, Genetic predisposition to sporadic cancer, Tumor specific markers, Applications of new technologies in diagnostics, treatment and prevention of cancer.

Unit-IV: General Toxicology and Toxicity Testing:

General principles and terminology; Types of toxicity; Factors affecting toxicity; Acute and Chronic toxicity; LD₅₀, LC₅₀, IC₅₀, EC₅₀; Route of administration; Dose response relationship and its evaluation, Risk assessment Introduction and Principle of Free radical toxicity testing methods, Cytotoxicity testing methods and Genotoxicity testing methods.

ELECTIVE PAPER - I
CELL BIOLOGY, CYTOGENETICS AND MOLECULAR BIOLOGY

HG-504EA - Cell Biology, Cytogenetics And Molecular Biology-II

Unit-I: Cell Signaling and communication

Cell signaling, Types of signal molecules, Secondary messengers, Types of receptors, Signal transduction, Component of signaling system, Hormone mechanism of action, KEGG pathway, Scatchard plot, Cell signaling technology

Unit-II: Nanotechnology

Introduction to nanotechnology, Nanotechnology: origin, history and principles, Approaches of nanotechnology in biology, Nanoscale advances, Nanoparticles, Nanodevices, Nanomedicine

Unit-III: Stem Cell Biology

Molecular Basis of stem cells; Different types of stem cells; Stem cells isolation and characterization, Recent advances in stem cell research and regenerative medicine; Application of stem cell therapy in diabetes, skin, dental, bone, heart, reproductive, muscles, neuronal disease.

Unit-IV: Molecular Biology

Variants in PCR and its specific applications; Second and third generation sequencing; Chromosome walking and jumping; Somatic and germ line gene therapy, Criteria for effective gene therapy, General gene therapy strategies; Gene silencing; Transgenic animals; DNA vaccines; Microarray types and applications; Flow cytometry; Recent advances in molecular biology.

ELECTIVE PAPER - II
ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY

HG--503EB: ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY - I

UNIT – I: General Endocrinology

Origin, Evolution and Classification of Endocrine system. Feedback Mechanisms, Regulation by Signal transduction and Hormones, Nerves' metabolites, Hormones Mechanism of action, Hormone receptors mechanism and metabolism

UNIT – II: Endocrine Glands

Endocrine glands – Hormone synthesis, release, regulation and its pathology. Hormones of extra endocrine sites – GI tract, Kidney, Liver, Heart, Lungs

UNIT – III: Hormonal Control

Hormonal control of carbohydrate, lipid, protein and nucleic acid and mineral metabolism. Hormonal control of growth and development, Endocrine role of adipose Tissues, Hormonal control of Reproductive behavior, Endocrine basis of communication in reproduction and aggression Pheromones, Vitamin D as a Hormone

UNIT – IV: General Toxicology and Toxicity Testing

General principles and terminology; Types of toxicity; Factors affecting toxicity; Acute and Chronic toxicity; LD₅₀, LC₅₀, IC₅₀, EC₅₀; Route of administration; Dose response relationship and its evaluation, Risk assessment Introduction and Principle of Free radical toxicity testing methods, Cytotoxicity testing methods and Genotoxicity testing methods.

ELECTIVE PAPER - II
ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY

HG-504-EB: ENDOCRINOLOGY AND REPRODUCTIVE TECHNOLOGY - II

UNIT – I: Gonadal Development

Development of Gonads, Accessory sex organ and external genitalia, Factors affecting sex determination, Genetic Control of sex determination, Disorders of gonadal differentiation, Endocrinology of the foetal gonads, Prenatal diagnostic techniques

UNIT – II: Female Reproductive Organs

Structure and functions of Female reproductive organs. Folliculogenesis, Ovulation, Corpus luteum, atrecia, Implantation, Placenta, Pregnancy and its control Foetoplacental unit as an endocrine entity. Menopause and hormone replacement therapy, reproductive cycles, non-steroidal regulators of ovarian function. Gamete and zygote transport. Mammary glands, Lactation and its hormonal control

UNIT – III: Male Reproductive Organs

Structure and function of the Male Reproductive Organs. Spermatogenesis and its hormonal control. Ultrastructure functions of the Sertoli cells and Leydig Cells. Semen formation and its biochemistry. Sperm structure function and sperm function tests. Testis Anomalies, Senescence, Puberty

UNIT – IV: IVF and General Techniques.

Principles of Fertility Regulation in males and females, *In-Vitro* Fertilization, ET and AR technologies, PGS and PGD, Mitochondrial Replacement therapy, Collection and preservation of Gametes, Cryopreservation assay, Research methodologies; RIA, PCR, EIA, IRMA, Radioreceptors, Sequencing methods, Chemiluminescence, ECL, Polyclonal and Monoclonal antibodies, Hybridoma technology.

ELECTIVE PAPER - III GENETIC COUNSELING

HG-503EC: Genetic Counseling – I

Unit-I: Practice of Genetic counseling & Pedigrees

Concept and purpose, Historical overview of genetic counseling, goals, Philosophy and ethos of genetic services and counseling, Providers of genetic counseling, Practice based competencies for genetic counselors, Indications and purpose, Past medical, social & family history, construction of pedigrees & Patterns of inheritance, Incidental detection of other genetic disorders on pedigree evaluation, establishing carriers.

Unit-II: Interviewing, Case preparation & Management

Basic communication skills, interviewing techniques, verbal & non-verbal communication, inter cultural communication, case preparation & management, Psychotherapeutic counseling & decision-making concepts, Medical Genetic evaluation, Physical examination, Documentation.

Unit-III: Risk assessment

Risk assessment and counseling in: common Variants, Mendelian and multifactorial Disorders, genetic case pathway and preventive management guidelines, bioinformatics resources and their use in informing genetic testing methodologies and reporting.

Unit-IV: Principles of Genetic Toxicology & Toxicity Testing

General principles and terminology; Types of toxicity; Factors affecting toxicity; Acute and Chronic toxicity; LD₅₀, LC₅₀, IC₅₀, EC₅₀; Route of administration; Dose response relationship and its evaluation, Risk assessment Introduction and Principle of Free radical toxicity testing methods, Cytotoxicity testing methods and Genotoxicity testing methods.

ELECTIVE PAPER - III GENETIC COUNSELING

HG-504EC: Genetic Counseling – II

Unit-I: Prenatal diagnosis & Newborn screening

Ethical and legal issues in Prenatal diagnosis, Indications of prenatal diagnosis, prenatal diagnosis in late onset disorders, Techniques: invasive, non-invasive methods – Amniocentesis, Chorion Villus sampling, NIPT, ultrasound, maternal blood screening, PGD, Screening of genetic disorders in newborn.

Unit-II: Genetic testing & interpretation

understanding genetic testing: Interpretation of new genome testing techniques, DNA testing – Direct testing (Known and unknown mutations) and Indirect testing (gene tracking), DNA profiling: establishing identity and relationships Population screening - ethics, organization and advantages. Pre-symptomatic testing for late onset diseases.

Unit-III: Contexts of Genetic counseling

Reproductive issues: Technologies in reproductive assistance, pre-conceptional counseling, Medico-legal Issues: Surrogacy, Paternity testing. Congenital anomalies and rare medical disorders: risk factors, Rare disorders – why not treatable? pre-pregnancy evaluation and intervention strategies, Social and cultural issues, support groups for rare medical disorders. Impact of disorders of sexual development (DSDs) and genetic counselling. Genetic counseling for late onset diseases (example neuro-genetic disorders, Myotonic dystrophy and Inherited cancers, breast and ovarian cancer): Predictive counseling & Principles, management in follow-up sessions. Testing of vulnerable populations, ethical issues in testing of minors & incapacitated individuals.

Unit-IV: Ethical, legal and social issues in Human genetics

Influences on Genetic counseling, Bioethics in research & experimentation, Implications of New Technologies: embryonic stem cells, animal cloning, genetic engineering, gene therapy & clinical trials, Supervision and strategies for providing support & guidance to new counselor, Dilemmas faced by counselors, Medical ethics in India, Informed consent and confidentiality, Right of choice, Impact of genetic disease on patients and families, social practices and health cost issues. Case studies, interaction with parents & families to increase empathy.

ELECTIVE PAPER - IV
TOXICOLOGY

HG-503ED - TOXICOLOGY-I

Unit-I Animal House and Maintenance:

Design and construction of animal housing facilities; Caging and housing system; Important factors in animal housing: Illumination, Noise, Ventilation, Temperature, Humidity, Bedding, Water, Food; Health monitoring; Sanitation; Waste disposal; IAC and CPCSEA

Unit-II: Target Organ Toxicity:

Toxic responses of Blood, Liver, Kidney, Skin, Immune system, Respiratory system, Nervous system, Ocular and visual system, Heart and vascular system, Reproductive system, Endocrine system

Unit-III: Toxic Agents and Toxicokinetics:

Classification of toxicants; Metals; Pesticides; Xenobiotics; Teratogens; Food additives and contaminants; Toxins of animal and plant origin; Radiation types, detection and effects. Absorption; Digestion; Metabolism; Excretion; Mutagenicity; Carcinogenicity; Teratogenicity; Biotransformation; Bioactivation; Mechanism of Toxicity

Unit-IV: Principle of Toxicology and Basics of Toxicity Testing Methods:

General principles and terminology; Types of toxicity; Factors affecting toxicity; Acute and Chronic toxicity; LD₅₀, LC₅₀, IC₅₀, EC₅₀; Route of administration; Dose response relationship and its evaluation, Risk assessment, Introduction and Principle of Free radical toxicity, Cytotoxicity and Genotoxicity testing methods

ELECTIVE PAPER - IV TOXICOLOGY

HG-504ED- TOXICOLOGY-II

Unit-I: Cytotoxicity, Genotoxicity and Free Radical Toxicity Testing:

Cell viability and cytotoxicity: Trypan blue dye exclusion assay, MTT assay, LDH and Neutral red uptake assay, WST assay, SRB assay; Genotoxicity: Ames test, TK, HPRT and XPRT mutation test, SCE, Chromosomal aberration assay, Comet assay, Micronucleus assay; Free radical toxicity: Total ROS, LPO, SOD, Catalase, GSH, GPx, Grd, GST; Carcinogenicity testing; Teratogenicity testing, (OECD 473 and 490) Cell lines for toxicity testing: L5178Y, CHO, AS52, V79, TK6, CACO II

Unit-II: Ecotoxicology:

Principle of ecotoxicology; Terrestrial model and ecotoxicity testing: Earthworm, Honey bee, Birds, Plants; Aquatic models and ecotoxicity testing: Algae, Daphnia, Brine shrimp, Fish. Route of exposure: Water, Soil, Food; Ecotoxicogenomics: Estrogen receptor, Aryl hydrocarbon receptor, Protein damage, DNA damage

Unit-III: Environmental Toxicology:

Environmental risk assessment; Environmental health and hazards; Air pollutants: Past disasters and current condition, Outdoor pollutants, Photochemical oxidants, Indoor air pollutants; Water and soil pollutants: Synthetic persistent chemicals, Inorganic ions, Cyanotoxins

Unit-IV: Guidelines, Quality Standards and Applications of Toxicology:

General principles and terminology; Types of toxicity; Factors affecting toxicity; Acute and Chronic toxicity; LD₅₀, LC₅₀, IC₅₀, EC₅₀; Route of administration; Dose response relationship and its evaluation, Risk assessment Introduction and Principle of Free radical toxicity testing methods, Cytotoxicity testing methods and Genotoxicity testing methods.

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

HG – 505 PR:

Practicals based on the theory papers HG - 501 and HG - 502. Field/Industrial/ Laboratory visits.

HG – 506 PR:

Practicals based on the theory papers HG - 503 and HG - 504. Field/Industrial/ Laboratory visits.

Reference Books

1. David W. Mount (2001). Bioinformatics: Sequence and Genome Analysis (1st edition). Cold Spring Harbor Laboratory Press, USA.
2. Hooman H. Rashidi; Lukas K. Buehler (1999). Bioinformatics Basics: Applications in Biological Science and Medicine. CRC press
3. S. C. Rastogi, Namita Mendiratta, Parag Rastogi (2006). Bioinformatics, Concepts Skills and Applications. CBS Publishers & Distributors.
4. Arthur M. Lesk (2019). Introduction to Bioinformatics (5th edition). OUP Oxford
5. Misner & Krawetz (2000). Bioinformatics. Methods and protocols. Humana press
6. Khan & Kanum (2001). Recent Advances in Bioinformatics. Ukraaz Publication
7. Jeremy Ramsden (2016). Bioinformatics: An Introduction. Springer
8. David Edward; Jason Stajich; David Hansen (2014). Bioinformatics: Tools and Applications. Springer
9. Robert A. Weinberg (2012). The Biology of Cancer. Garland Science
10. Vincent T. DeVita, Jr., Theodore S.; Lawrence, Steven; A. Rosenberg (2011). Cancer: Principles and Practice of Oncology, 9th Edition. Lippincott Williams and Wilkins
11. Fred Bung (2008). Principles of cancer Genetics. Springer
12. Lauren Pecorino (2008). Molecular Biology of Cancer. Oxford Uni. Press
13. D.E. Rooney (2001). Human Cytogenetics: malignancy and acquired abnormalities. Oxford
a. University Press
14. D.E. Rooney (2001). 9. Human Cytogenetics: constitutional analysis. Oxford University Press
15. Margaret J. Barch (1997). The AGT Cytogenetics Laboratory Manual. Lippincott-Raven
16. Yao-Shan Fan (2003). Molecular Cytogenetics - Protocols and Applications. Methods in Molecular Biology (MIMB-204); Humana Press
17. Sverre Heim; Felix Mitelman (2011). Cancer Cytogenetics 3rd Edition. Willy-Blackwell
18. Hema Purandhare; Amit Chakravarty (2000). Human Cytogenetic Techniques and clinical Applications. Bhalani Publishing House
19. Benjamin M. Lewin (2000). Genes VII. Oxford University Press
20. Benjamin A. pierce (2002). Genetics: A Conceptual Approach. W. H. Freeman
21. Berwick, S.H. and Saharia, V.B. 1995. *Wildlife Research and Management*. OUP, New Delhi. 481pp.
22. A handbook of research methodology by Mishra and Alok, Educreation Publication
23. Research Methodology for life Sciences by Arumugam, Saras publication
24. Graduate research - A guide for students in Life Sciences. 2nd edition. Robert V. Smith
25. Research methods for Biosciences. 3rd edition. Debbie Holmes, Peter Moody, Diana Dine, Laurence Trueman
26. Experimental designs for Life Sciences. 4th edition. Graeme D., Ruxton and Nick Colegrave
27. Research methodology - Step by Step Guide for Beginners. 3rd edition. Ranjit Kumar
28. Research methodology - Tools and techniques, Bridge Centre 2015
29. Thesis and Dissertation Writing in a Second Language a handbook for supervisors - Brian Paltridge and Sue Starfield, Routledge Publishers
30. The process of research writing - Steven Krause – Eastern Michigan University

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

31. Animal - Use Research IAEC Project Proposal & animal imports Guidelines – NCBS Animal Care and Resource Center
32. Torrey, T. W. Morphogenesis of vertebrates. John Wiley and Sons Inc., New York
33. Comparative embryology of the vertebrates 1 – 2 (1953) McGraw – Hill Book company, New York
34. Developmental Biology. Eleventh Edition. By Scott F. Gilbert and Michael J. F. Barresi. Sunderland (Massachusetts): Sinauer Associates.
35. Jens, K. & Cy Aaron, S. (2016). An Introduction to Molecular Medicine. Germany, Wiley-VCH.
36. Dennis, W. R. (2013). Introduction to molecular medicine. Germany, Springer Science and Business Media.
37. William, B. C. & Gregory, J. T. (2016). Diagnostic Molecular Pathology. United States.
38. Robert A. Weinberg (2012). The Biology of Cancer. Garland Science
39. Vincent T. DeVita, Jr., Theodore S.; Lawrence, Steven; A. Rosenberg (2011). Cancer: Principles and Practice of Oncology, 9th Edition. Lippincott Williams and Wilkins
40. Lauren Pecorino (2008). Molecular Biology of Cancer. Oxford Uni. Press
41. Yao-Shan Fan (2003). Molecular Cytogenetics - Protocols and Applications. Methods in Molecular Biology (MIMB-204); Humana Press
42. Lauren Pecorino (2008). Molecular Biology of Cancer: Mechanisms, Targets and Therapeutics. Oxford University Press
43. Paul B. Fisher (2007). Cancer Genomics and Proteomics: Methods and Protocols (Methods in Molecular Biology, Vol. 383); Humana Press
44. Brian C.-S. Liu; Joshua R. Ehrlich (2008). Tissue Proteomics: Pathways, Biomarkers, and Drug Discovery (Methods in Molecular Biology, Vol. 441); Humana Press
45. Lawrence S.B. Goldstein; Meg Schneider (2010). Stem Cells For Dummies (1st Edition). For Dummies
46. Dov Zipori (2012). Biology of Stem Cells and the Molecular Basis of the Stem State (Stem Cell Biology and Regenerative Medicine). Humana press
47. Stem Cell Technology by Meghna Razdan & P.C. Trivedi (2009). Stem Cell Technology. Pointer Publishers
48. Paul Knoepfler (2013). Stem Cells: An Insider's Guide. World Scientific Publishing Company
49. Mary Clarke; Jonathan Frampton (2020). Stem Cells Biology and Application. Garland Science
50. Mikkel L. Sorensen (2008). Stem Cells Applications in Diseases. Nova Science Publishers Inc
51. Gabriela Rodrigues; Bernard A. J. Roelen (2020). Concepts and Applications of Stem Cell Biology - A Guide for Students. Springer Cham
52. Firdos Alam Khan (2021). Advances in Application of Stem Cells: From Bench to Clinics. Springer Nature Switzerland AG
53. Casarett and Doull's Toxicology The Basic Science of Poisons. Curtis D. Klaassen. Ed. McGraw Hill.
54. Lu's Basic Toxicology Fundamentals, Target Organs, and Risk Assessment. Frank C. Lu and Sam Kacew. Informa Healthcare USA.
55. Principles and Methods of Toxicology by A. Wallace Hayes. Taylor and Francis.
56. Griffith et al. An introduction to Genetic analysis. Published by Freeman

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

57. Jocelyn E. Krebs, Elliott S. Goldstein, Stephen T. Kilpatrick. Lewin's GENES XII. Published by Jones and Bartlett learning.
58. D. Peter Snustad, Michael J. Simmons. Principles of Genetics.
59. W S Klug & M R Cummings. Concepts of Genetics.
60. R E Pyeritz, B R Korf, W W Grody (Ed.). Emery & Rimoin Principles & practice of medical genetics & Genomics. Academic Press.
61. R F Mueller & I D Young. Elements of Medical Genetics. Published by Churchill Livingstone
62. J M Connor & M A Ferguson-Smith. Essential Medical Genetics. Blackwell publishers
63. Prakash S Lohar. 2021. Endocrinology Hormones and Human Health. MJP Publishers.
64. C. O'Donnell Turner and Joseph Bagnara. 2012. General Endocrinology. East West Press Ltd. New Delhi
65. Melmed, Polonsky, Kronenberg and Larsen. 2011. Williams Textbook of Endocrinology. 12th Edition. Elsevier Science.
66. Francis S. Greenspan and David G. Gardner. 2003. Basic & Clinical Endocrinology. Lange Medical Publishers
67. Jerome F. Strauss and Robert L. Barbieri. Yen & Jaffe's Reproductive Endocrinology Physiology, Pathophysiology, and Clinical Management. 2018. Elsevier Science.
68. E.S.E. Hafeez. 1973. Human Reproduction. Lippincott Williams & Wilkins.
69. Roy O. Greep, Maurice B. Visscher, John R. Pappenheimer. 1973. Handbook of Physiology: A Critical, Comprehensive Review. Oxford University Press.
70. E.S.E Hafeez. 1976 Human Semen and Fertility Regulation. Lippincott Williams & Wilkins.
71. Martin H. Johnson and Barry J. Everitt. 1988. Essential Reproduction. A Blackwell Scientific Publications.
72. E.S.E. Hafez 1975. Human reproduction: Conception and contraception, Lippincott Williams & Wilkins.
73. Casarett and Doull's Toxicology The Basic Science of Poisons. Curtis D. Klaassen. Ed. McGraw Hill.
74. Lu's Basic Toxicology Fundamentals, Target Organs, and Risk Assessment. Frank C. Lu and Sam Kacew. Informa Healthcare USA.
75. Principles and Methods of Toxicology by A. Wallace Hayes. Taylor and Francis.
76. Casarett and Doull's Toxicology The Basic Science of Poisons. Curtis D. Klaassen. Ed. McGraw Hill.
77. Lu's Basic Toxicology Fundamentals, Target Organs, and Risk Assessment. Frank C. Lu and Sam Kacew. Informa Healthcare USA.
78. Principles and Methods of Toxicology by A. Wallace Hayes. Taylor and Francis.
79. W. R. Uhlmann, J. L. Schuette, B. M. Yashar (Ed.) A Guide to Genetic counseling. Wiley-Blackwell publication.
80. R F Mueller & I D Young. Elements of Medical Genetics. Published by Churchill Livingstone
81. J M Connor & M A Ferguson-Smith. Essential Medical Genetics. Blackwell publishers
82. T Strachan & A P Read. Human Molecular Genetics. Bios Scientific publishers.
83. R J M Gardner, G R Sutherland. Chromosome abnormalities & genetic counseling. Oxford University Press.
84. J de Grouchy, J Turleau. Clinical atlas of Human chromosomes. Published by John Wiley.

**HUMAN GENETICS - SYLLABUS-SEMESTER III - JUNE 2022 ONWARDS,
GUJARAT UNIVERSITY, AHMEDABAD – 380 009, GUJARAT, INDIA.**

85. P S Harper. Practical genetic counseling. Published by Butterworth Heinemann
86. D L Rimoin, J M Connor & R S Pyeritz (eds). Principles & practice of medical genetics. Published by Churchill Livingstone
87. F Vogel & A G Motulsky. Human Genetics: problems & approaches. Published by Springer.
88. S V Hodgson & E R Maher. A practical guide to human cancer genetics. Published by Cambridge University Press.
89. Casarett and Doull's Toxicology The Basic Science of Poisons. Curtis D. Klaassen. Ed. McGraw Hill.
90. Lu's Basic Toxicology Fundamentals, Target Organs, and Risk Assessment. Frank C. Lu and Sam Kacew. Informa Healthcare USA.
91. Principles and Methods of Toxicology by A. Wallace Hayes. Taylor and Francis.
92. Principle of Biochemical Toxicology by John Timbrell.
93. Presenting Toxicology results by Gerhard J. N.
94. Emergency Toxicology. Bania, Brent et. Al.
95. Reproductive and developmental Toxicology. Korach.
96. Medical Toxicology. Schonwald.
97. Toxicology and Pathology. Haschek R.
98. Experimental Toxicology. Aldridge et al.
99. A guide to practical toxicology. Evaluation, Prediction & Risk. Adam Woolley.
100. Food & Nutritional Toxicology. Stanley T. Omaye.
101. Experimental Toxicology. The Basic Issues. D. Anderson and D. M. Conning.
102. Environmental Toxicology. David A. Wright and Pamela Welbourn.
103. Environmental Toxicology. Sigmund F. Zakrzewski
104. Predictive Toxicology. Christoph Helma.
105. Principles of Toxicology. Environmental & Industrial Applications.
106. Hodgson, A Textbook of Modern Toxicology. J Wiley and Sons.
107. Molecular and Biochemical Toxicology. Smart and Hodgson, eds. J Wiley and Sons.
108. Frank and Ottoboni, The Dose makes the Poison: A Plain-language Guide to Toxicology. J Wiley and Sons.
109. Ottoboni, The Dose makes the Poison: A Plain-language Guide to Toxicology. J Wiley and Sons.
110. A-Z Guide to Drug-Herb-Vitamin Interactions. Gaby, Batz, Chester and Constantine, eds. Three Rivers Press.
111. Gilbert, A Small Dose of Toxicology: The health effects of common chemicals. CRC Press.
112. Gibson, Multiple Chemical Sensitivities: A survival guide. New Harbinger Publications.
113. Toxicology and Clinical Pharmacology of Herbal Products, Cupp and Karch, eds. Springer-Verlag.
114. Lawson, Staying Well in a Toxic World. Lynwood Press.
115. Guide for the Care and Use of Laboratory Animals. The National Academies Press
116. Compendium of CPCSEA. Ministry of Environment, Forest and Climate Change, Government of India.

M.Sc. Human Genetics, Semester – IV

SR. No.	Course Code	Name of the Course	Hours per Week	Internal Marks	External Marks	Total Marks	Credits
1	HG – 507PT	Dissertation and Viva-voce	20	120	280	400	16
2	HG – 508S	Assignments / group discussions.	4	30	70	100	04
3	HG – 509M	Seminars and Industrial Visits during the Semester.	4	30	70	100	04
		Total	28	180	420	600	24

HUMAN GENETICS SEMESTER – IV

HG-507PT : Dissertation and Viva

HG-508S : Seminar /Industrial Visits

HG-509M : Assignment/Group Discussions.