M.Sc. in Human Genetics 2 Years (4 Semesters)

Overview: The M.Sc. in Human Genetics is a specialized postgraduate program designed to provide students with a deep understanding of the genetic basis of human health, disease, and evolution. This field focuses on studying the inheritance of genetic traits, the molecular mechanisms underlying genetic diseases, and how genetic information is passed down through generations.

Students in this program will gain an in-depth knowledge of genetics at both the molecular and population levels, covering areas such as genomics, gene therapy, genetic counseling, and bioinformatics. The program combines theoretical learning with practical laboratory training, equipping students with the skills necessary for research and clinical applications in genetics.

Human genetics is a rapidly evolving field with significant implications for personalized medicine, genetic testing, and disease prevention, making it an exciting area for those interested in healthcare, biotechnology, and research.

Affiliated Institution: School of Medical Sciences and Technology, Malla Reddy Vishwavidyapeeth (Deemed to be University) ****** The minimum eligibility for M.Sc. in Human Genetics is a pass in B.Sc with at least 50% marks in qualifying exam.

Key Highlights:

- **Advanced Genetics Knowledge**: Students will develop a deep understanding of human genetics, focusing on both the basic principles and modern advancements in genomics and genetic technologies.
- **Genetic Disease Mechanisms**: Study the genetic causes of inherited and complex diseases, including single-gene disorders, multifactorial diseases, and cancer genetics.
- **Cutting-Edge Technologies**: Exposure to advanced technologies such as next-generation sequencing (NGS), CRISPR-Cas9 gene editing, gene therapy, and bioinformatics tools for analyzing genetic data.
- **Clinical Applications**: Focus on the clinical aspects of human genetics, including genetic counseling, prenatal testing, genetic screening, and the application of genetic information in personalized medicine.
- **Hands-on Laboratory Training**: Practical training in laboratory techniques for DNA/RNA extraction, PCR, gel electrophoresis, genome sequencing, and genetic data analysis.
- **Ethics and Legal Issues in Genetics**: Study the ethical, legal, and social implications of genetic research and testing, especially in areas like gene editing and genetic privacy.

Course Curriculum:

The M.Sc. in Human Genetics is typically a two-year program, combining coursework, laboratory training, and research projects. Below is a general outline of the course structure:

Year 1:

Core Modules:

- **Molecular Genetics**: The study of genes at the molecular level, including DNA structure, gene expression, mutations, and genetic variation.
- **Genomic Technologies**: Introduction to the technologies used to study the genome, including PCR, microarray analysis, next-generation sequencing (NGS), and gene editing tools like CRISPR-Cas9.
- **Human Genetics and Inheritance**: The principles of inheritance, Mendelian genetics, genetic mapping, and the application of genetic principles to human traits and diseases.
- **Genetics of Disease**: Study the genetic basis of both monogenic (single-gene) and complex (multifactorial) diseases, with examples such as cystic fibrosis, sickle cell anemia, and diabetes.
- **Population Genetics**: Focus on genetic variation within populations, genetic drift, selection, migration, and gene flow, and how these factors contribute to human evolution and disease susceptibility.
- **Bioinformatics in Genetics**: Introduction to computational tools and software used to analyze genetic data, including genome-wide association studies (GWAS), sequence alignment, and variant calling.
- **Genetic Counseling**: Principles of genetic counseling, including how genetic information is communicated to patients and families, and the ethical issues involved in genetic testing.

Practical Training:

- Laboratory-based training in techniques such as PCR, gel electrophoresis, restriction enzyme analysis, and DNA sequencing.
- Hands-on use of bioinformatics tools to analyze genetic data, including mutation identification and variant interpretation.
- Practical experience in case studies related to genetic counseling and clinical genetics.

Year 2:

Advanced Modules:

- **Cancer Genetics**: Explore the genetic basis of cancer, including the role of oncogenes, tumor suppressor genes, and the impact of mutations on cancer progression and treatment.
- **Genomic Medicine**: The application of genomic information in the clinic, including pharmacogenomics, genetic testing for disease prevention, and personalized medicine strategies.
- **Gene Therapy and Editing**: Study the principles of gene therapy and the use of genetic engineering tools, such as CRISPR, to treat genetic disorders.
- **Ethics and Policy in Human Genetics**: Examination of ethical, legal, and social issues in genetics, including genetic privacy, discrimination, and the implications of gene editing technologies.

- **Molecular Diagnostics**: Learn about diagnostic techniques that use genetic information to diagnose diseases, including genetic screening, carrier testing, and prenatal diagnostics.
- **Clinical Genetics and Genomics**: Explore the role of geneticists in diagnosing and managing inherited diseases, using case studies to understand the application of genetics in a clinical setting.

Research Project/Dissertation:

- The second year culminates in a research project or dissertation, where students conduct original research in a laboratory or clinical setting. Students will apply their knowledge of human genetics to investigate specific research questions, such as the identification of genetic markers for disease, the effects of gene therapy, or the use of genomics in personalized medicine.
- Students are encouraged to present their findings in written and oral formats at academic conferences and journals.

Career and Academic Opportunities:

Career Opportunities:

Graduates of the M.Sc. in Human Genetics have a wide range of career options in healthcare, research, biotechnology, and education. Potential career paths include:

- **Genetic Counselor**: Providing counseling to individuals and families regarding genetic testing, inheritance patterns, and the risks of genetic disorders.
- **Genetics Research Scientist**: Conducting research in academic or corporate laboratories to investigate genetic diseases, develop new diagnostic methods, or improve gene therapies.
- **Clinical Geneticist**: Working in clinical settings to diagnose genetic disorders, interpret genetic test results, and advise patients on genetic health risks.
- **Laboratory Technician**: Managing genetic testing labs, performing genetic analyses, and processing patient samples.
- **Bioinformatician**: Analyzing genetic data using computational tools and bioinformatics techniques, often in collaboration with researchers in genomics, epidemiology, and cancer genetics.
- **Pharmaceutical/Biotech Industry Professional**: Developing or testing genetic-based therapies, such as gene editing or gene therapies, for genetic diseases.
- **Public Health Geneticist**: Conducting population-based studies on the genetic epidemiology of diseases and developing public health strategies based on genetic findings.
- **Genomics Specialist in Personalized Medicine**: Working in hospitals or research centers to develop personalized treatment plans based on a patient's genetic profile.

Academic Opportunities:

Graduates can pursue further academic qualifications, including:

- **Ph.D. in Human Genetics or Genomics**: Conducting advanced research in genetics, often focusing on specific diseases, gene therapy, or personalized medicine.
- **Postdoctoral Research**: Engaging in specialized research on topics such as cancer genetics, genetic epidemiology, or gene editing.
- **Medical Degrees (MD)**: Some graduates choose to pursue an MD in genetics or other related fields to work directly with patients.

Research Prospects:

- **Gene Editing and Therapy**: Researching advanced gene editing technologies like CRISPR-Cas9 and their potential for treating genetic disorders.
- **Cancer Genomics**: Investigating the genetic alterations that drive cancer and developing genetic-based therapies or diagnostic tools.
- **Genetic Disease Mechanisms**: Understanding the molecular basis of inherited diseases and exploring gene-targeted treatments.
- **Genetic Epidemiology**: Studying the distribution of genetic diseases within populations and identifying genetic risk factors for complex diseases such as diabetes and heart disease.
- **Pharmacogenomics**: Research into how genetic variation affects an individual's response to medications, with the goal of personalizing drug prescriptions.

Professional Opportunities:

- **Certified Genetic Counselor**: After completing the M.Sc. program, students can become certified genetic counselors by passing certification exams in countries like the US (ABGC) or the UK (GCRB).
- **Clinical Research Associate (CRA)**: Overseeing clinical trials for genetic therapies and ensuring compliance with ethical standards and regulations.
- **Regulatory Affairs Specialist**: Ensuring compliance with regulations and ethical guidelines in genetic testing, gene therapy, and genomic medicine.

Higher Education and Research Prospects:

- **Ph.D. in Human Genetics**: Graduates may pursue a Ph.D. in genetics or related fields to explore specialized research in areas such as gene therapy, cancer genomics, or rare genetic diseases.
- **Postdoctoral Research**: Engaging in advanced research at universities or research institutions, focusing on cutting-edge developments in genomics, gene editing, or personalized medicine.

Conclusion:

The **M.Sc. in Human Genetics** is an ideal program for students passionate about understanding the genetic basis of health and disease. It offers comprehensive training in both theoretical knowledge and practical skills, making graduates well-equipped to contribute to the rapidly growing fields of genetic research, diagnostics, and therapy.

With the increasing importance of genomics in personalized medicine and disease prevention, graduates of this program will be in high demand in academic, healthcare, and biotechnology sectors. As the field of genetics continues to evolve, the opportunities for research and clinical applications of human genetics are vast and varied, offering exciting career and academic prospects.

Labs

1. Molecular Genetics & DNA Analysis Lab

- **DNA & RNA Extraction** (from blood, saliva, tissue)
- **Polymerase Chain Reaction (PCR) & Quantitative PCR (qPCR)**
  - ✓ SNP genotyping
  - ✓ Gene expression studies
- **Next-Generation Sequencing (NGS)**
  - ✓ Whole Genome Sequencing (WGS)
  - ✓ Whole Exome Sequencing (WES)
  - ✓ Targeted gene panels for disease-specific analysis
- **Mitochondrial DNA analysis** for inherited disorders

2. Cytogenetics & Chromosomal Analysis Lab

- **Karyotyping & G-banding** for chromosomal abnormalities (Down syndrome, Turner syndrome, etc.)
- **Fluorescence In Situ Hybridization (FISH)** for chromosomal deletions & duplications
- **Comparative Genomic Hybridization (CGH)** for genome-wide CNV detection
- **Spectral Karyotyping (SKY) & Multi-color FISH (mFISH)** for complex chromosomal rearrangements

3. Genomics & Bioinformatics Lab

- **Genome Annotation & Comparative Genomics**
  - ✓ Use of databases (NCBI, UCSC Genome Browser, Ensembl)
  - ✓ Gene and variant annotation tools (ANNOVAR, SnpEff)
- **Variant Calling & Functional Interpretation**
  - ✓ GATK, FreeBayes for SNP & INDEL detection
  - ✓ PolyPhen, SIFT for predicting variant pathogenicity
- **Transcriptomics & Epigenetics Analysis**

- ✓ RNA-Seq data analysis (STAR, HTSeq, DESeq2)
- ✓ DNA Methylation Studies (Bisulfite Sequencing)

4. Genetic Disease & Clinical Diagnostics Lab

- **Hereditary Disease Testing**
  - ✓ Carrier screening for autosomal recessive diseases (e.g., Cystic Fibrosis, Thalassemia)
  - ✓ Genetic risk assessment for cancer (BRCA1/BRCA2 mutations)
- **Pharmacogenomics**
  - ✓ CYP450 enzyme genotyping for drug metabolism
  - ✓ Warfarin, Clopidogrel, Statins gene-drug interactions
- **Forensic & Population Genetics**
  - ✓ DNA fingerprinting using STR analysis
  - ✓ Y-chromosome & mitochondrial haplogroup analysis

5. Functional Genomics & Gene Editing Lab

- **CRISPR-Cas9 & RNAi Technology**
  - ✓ Gene knockout & gene correction studies
- **Gene Therapy Research**
  - ✓ Viral & non-viral gene delivery systems
- **Protein Expression & Functional Studies**
  - ✓ Western blotting, ELISA, Immunocytochemistry

6. Stem Cell & Developmental Genetics Lab *(optional but useful)*

- Differentiation of iPSCs into disease models
- Organoid models for studying genetic disorders

7. Ethical, Legal & Social Implications (ELSI) Lab

- Genetic counseling & ethical considerations in genetic testing
- Bioethics in genome editing (CRISPR) and gene therapy
- Clinical genetics regulations (FDA, EMA, WHO guidelines)

PROGRAM OUTCOMES (POs)

PO	Program Outcomes
PO-1	Understand the principles of genetics and genomic medicine.
PO-2	Apply genetic testing and counseling in clinical practice.
PO-3	Conduct research on genetic disorders and personalized medicine.
PO-4	Integrate bioinformatics for genomic data interpretation.
PO-5	Adhere to ethical and legal considerations in genetic studies.
PO-6	Develop precision medicine approaches for genetic diseases.

COURSE STRUCTURE – M.Sc. Human Genetics

SEMESTER – I

Sl. No.	Broad Category	Course Code	Name of the Subject/Practical	Contact hours/week			Credits
				L	T	P	
1.	Major (Core)	MSHG101	Cell Biology	2	1	0	3
2.		MSHG102	Basics of Human Embryology, Anatomy and Physiology	2	1	0	3
3.		MSHG103	Fundamentals of Human Genetics	2	1	0	3
4.		MSHG104	Human Molecular Biology	2	0	2	3
5.	**Minor** Select any two minor courses, each worth 3 credits, for a maximum of 6 credits per semester	MSHG105	1. Epigenetics & Gene Regulation 2. Population Genetics & Evolutionary Biology 3. Pharmacogenomics & Precision Medicine 4. Genetic Analysis & Cytogenetics Laboratory 5. Computational Genomics & Bioinformatics 6. Research Methodology & Biostatistics	2 2	0 0	2 2	6
6.	**Skill Enhancement Courses**	MSHG106	1. Molecular Techniques in Genetic Testing 2. Interpretation of genetic test results	0 0	0 0	2 2	2
			Total	12	3	10	**20**
Total Contact Hours				**25**			

Course outcomes for M.Sc. Human Genetics MAJOR

Course Name	Course Outcomes
Cell Biology	- Understand the structure and function of prokaryotic and eukaryotic cells.- Explain cellular organelles, their functions, and interactions in maintaining homeostasis.- Analyze cell cycle regulation, apoptosis, and cell signaling mechanisms.- Evaluate the role of cellular processes in health, disease, and therapeutic interventions.- Apply microscopy and molecular techniques to study cellular structures and functions.
Basics of Human Embryology, Anatomy, and Physiology	- Understand the key stages of human embryonic development and organogenesis.- Explain the anatomical structure and physiological functions of major organ systems.- Analyze the relationship between structure and function in different tissues and organs.- Evaluate physiological homeostasis and its dysregulation in diseases.- Apply knowledge of human anatomy and physiology in clinical and research settings.
Fundamentals of Human Genetics	- Understand the principles of Mendelian and non-Mendelian inheritance.- Explain chromosomal structure, genetic variation, and patterns of inheritance.- Analyze the role of genes in health, disease, and genetic disorders.- Evaluate genetic counseling, screening, and ethical considerations in genomics.- Apply molecular and cytogenetic techniques in genetic diagnostics and research.
Human Molecular Biology	- Understand the molecular mechanisms of DNA replication, transcription, and translation.- Explain gene expression regulation, epigenetics, and RNA biology.- Analyze the impact of mutations, gene editing, and genetic engineering technologies.- Evaluate molecular approaches in personalized medicine and gene therapy.- Apply molecular biology techniques (PCR, sequencing, CRISPR) in biomedical research.

Course outcomes for M.Sc. Human Genetics MINOR

Course Name	Course Outcomes
Epigenetics & Gene Regulation	- Understand the mechanisms of epigenetic modifications, including DNA methylation, histone modifications, and non-coding RNAs.- Explain the role of epigenetic regulation in gene expression, development, and disease.- Analyze the impact of environmental factors on epigenetic changes and transgenerational inheritance.- Evaluate epigenetic therapies and their applications in cancer and neurological disorders.- Apply laboratory techniques for studying epigenetic modifications, such as ChIP-seq and bisulfite sequencing.
Population Genetics & Evolutionary Biology	- Understand the principles of population genetics, including allele frequency, genetic drift, and natural selection.- Explain the genetic basis of evolution, speciation, and adaptation.- Analyze

Course Name	Course Outcomes
	phylogenetic relationships and molecular evolution using genetic markers.- Evaluate the role of human genetic diversity in disease susceptibility and population health.- Apply statistical and computational tools to study genetic variation in populations.
Pharmacogenomics & Precision Medicine	- Understand the genetic basis of drug metabolism, response, and adverse effects.- Explain the role of genetic polymorphisms in personalized medicine and targeted therapies.- Analyze case studies on pharmacogenomics applications in oncology, cardiology, and psychiatry.- Evaluate ethical, legal, and social implications of genetic-based treatments.- Apply pharmacogenomic data analysis techniques for optimizing drug efficacy and safety.
Genetic Analysis & Cytogenetics Laboratory	- Understand the principles of genetic and chromosomal analysis techniques.- Explain karyotyping, fluorescence in situ hybridization (FISH), and chromosomal microarray analysis.- Perform laboratory-based cytogenetic techniques for diagnosing genetic disorders.- Analyze chromosomal abnormalities and their clinical significance.- Apply cytogenetic tools for prenatal screening, cancer genetics, and reproductive medicine.
Computational Genomics & Bioinformatics	- Understand the fundamentals of computational biology, genome sequencing, and annotation.- Explain data analysis techniques for next-generation sequencing (NGS) and genome-wide association studies (GWAS).- Analyze genomic datasets using bioinformatics tools such as BLAST, FASTA, and phylogenetic analysis.- Evaluate the role of artificial intelligence and machine learning in genomic research.- Apply computational methods for variant analysis, functional genomics, and systems biology.
Research Methodology & Biostatistics	- Understand the principles of scientific research design and hypothesis testing.- Explain data collection methods, sampling techniques, and statistical study designs.- Analyze statistical methods used in genetic and biomedical research.- Evaluate data interpretation, statistical significance, and error analysis.- Apply biostatistical software (SPSS, R, Python) for data analysis and visualization.

M.Sc. in Human Genetics – Course Structure & Syllabus

Course Duration: 2 Years (4 Semesters)

Total Credits: 80–100

Total Teaching & Training Hours: ~3,600

Total Teaching Hours Distribution

1. **Theory Classes:** ~1,200–1,500 hours
2. **Practical & Laboratory Training:** ~800–1,000 hours
3. **Clinical Internship & Hands-on Training:** ~800–1,000 hours
4. **Research Project & Dissertation:** ~300–500 hours

Assessment Methods

Assessment Component	Weightage (%)	Details
Continuous Internal Assessment (CIA)	**40%**	Includes internal exams, assignments, presentations, case studies, and practical performance
End-Semester Examination (ESE)	**60%**	Divided into theory (40%) and practical (20%)
Mid-Semester Exams	**20%** (Part of CIA)	Two internal tests per semester
Assignments & Case Studies	**5%** (Part of CIA)	Research-based assignments, literature reviews, clinical case reports
Seminars & Presentations	**5%** (Part of CIA)	Oral/poster presentations on diabetes management
Practical Performance & Clinical Evaluation	**5%** (Part of CIA)	Skill-based assessments in labs/hospitals
Attendance & Participation	**5%** (Part of CIA)	Regularity in theory & practical sessions
Theory Examination (Final)	**40%** (Part of ESE)	Structured written paper covering subject knowledge
Practical Examination (Final)	**20%** (Part of ESE)	Includes viva, skill demonstration, case handling
Dissertation/Research Project	**Mandatory**	Evaluated in the final year by internal & external examiners
Clinical Internship/Training	**Pass/Fail**	Logbook-based evaluation with hospital mentor review

Marking System & Grading

Marks (%)	Grade	Grade Point (GPA/CGPA Equivalent)	Classification
90 - 100	O (Outstanding)	10	First Class with Distinction
80 - 89	A+ (Excellent)	9	First Class with Distinction
70 - 79	A (Very Good)	8	First Class
60 - 69	B+ (Good)	7	First Class
50 - 59	B (Satisfactory)	6	Second Class
<50 (Fail)	F (Fail)	0	Fail (Re-exam Required)

Pass Criteria:

- **Minimum 50% marks in each subject** (Theory & Practical separately).
- **Aggregate of 55% required for progression** to the next semester.
- **No more than two backlogs** allowed for promotion to the final year.

Exam Pattern for Theory & Practical

A. Theory Examination Pattern

Total Marks: 100 (Converted to 40% for End-Semester Assessment)
Duration: 3 Hours

Section	Question Type	No. of Questions	Marks per Question	Total Marks
Section A	Short Answer Type (SAQ)	10 (Attempt all)	2	20
Section B	Long Answer Type (LAQ)	5 (Attempt any 4)	10	40
Section C	Case-Based/Clinical Scenario	3 (Attempt any 2)	15	30
Section D	MCQs/Objective Type	10 (Compulsory)	1	10
Total				**100**

Weightage:

- Molecular & Cytogenetics – 40%
- Genetic Disorders & Clinical Applications – 30%
- Research & Case Studies in Human Genetics – 20%

- Ethical, Legal & Social Aspects of Genetics – 10%

Passing Criteria: Minimum **50% (50/100 marks)**

B. Practical Examination Pattern

Total Marks: 100 (Converted to 20% for End-Semester Assessment)
Duration: 4–6 Hours

Component	Marks Distribution
Clinical Case Presentation & Genetic Disorder Assessment	30
OSCE (Objective Structured Clinical Examination) – Skill Demonstration	25
Molecular & Cytogenetic Techniques	20
Lab-Based Examination (DNA Extraction, PCR, Karyotyping, Genetic Counseling)	15
Record Work (Logbook & Assignments)	10
Total	**100**

OSCE (Skill-based Assessment) includes stations on:

- DNA Isolation & Quantification
- PCR & Gel Electrophoresis for Genetic Analysis
- Karyotyping & Chromosomal Abnormality Detection
- Genetic Counseling & Interpretation of Family Pedigrees

Passing Criteria: Minimum 50% (50/100 marks) in practicals.

Recommended Books & E-Resources

Textbooks

- **"Human Molecular Genetics"** – Tom Strachan & Andrew Read
- **"Principles of Genetics"** – Snustad & Simmons
- **"Medical Genetics"** – Lynn B. Jorde, John C. Carey, Michael J. Bamshad
- **"Thompson & Thompson Genetics in Medicine"** – Robert L. Nussbaum

E-Resources & Journals

- **Human Molecular Genetics (Oxford University Press)**
- **American Journal of Human Genetics (AJHG)**
- **National Center for Biotechnology Information (NCBI)**

- **Online Mendelian Inheritance in Man (OMIM)**

Career Opportunities after M.Sc. in Human Genetics

- **Genetic Counselor** in Hospitals & Clinics
- **Molecular Geneticist** in Research Labs
- **Clinical Geneticist** in Healthcare & Pharma
- **Pharmacogenomics Specialist** in Personalized Medicine
- **Bioinformatics & Genomics Analyst**