

**NATIONAL MEDICAL COMMISSION
ACADEMIC CELL**

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**GUIDELINES FOR COMPETENCY BASED POSTGRADUATE
TRAINING PROGRAMME FOR DM IN MEDICAL GENETICS**

Preamble

There have been significant advances in the field of Medical Genetics in recent years, which are of direct relevance to patient care. Following the completion of the Human Genome Project and the development of advanced molecular genetic testing technologies such as microarray and next generation sequencing, the genetic basis of a large number of chromosomal, monogenic as well as multifactorial disorders has been identified and many novel genetic disorders are being continuously discovered. This will include not only the genes causing monogenic disorders, but also those which predispose to complex multifactorial disorders and those which may be responsible for causing cancer, autoimmunity & aging.

Antenatal diagnosis is currently being offered for chromosomal, monogenic and copy number variations (CNVs) disorders. With wider availability of carrier screening in coming years, primary prevention of genetic disorders will be possible and thus will prevent the birth of a first affected child with genetic disorder. Other important clinical application is pre-symptomatic diagnosis of late onset diseases including cancers.

Advances of genetic technologies have already led to radical changes in the practice of Medical Genetics as well as concepts of genetic counselling. Access to these benefits that aim at birth of healthy children could be of immense importance to our National Family Welfare Program. Besides prevention, better understanding of pathophysiology of genetic diseases shall open new avenues of pharmaco-therapy, and even cure by gene therapy, of these disorders. Availability of treatments using many novel strategies like enzyme replacement therapy, drugs acting on molecular pathways, gene therapy, antisense oligo therapy and drugs for readthrough stop codon mutations has caused paradigm change in the management of many monogenic disorders. This

calls for trained specialized medical professionals to deal with genetic disorders with expertise in diagnosis, treatment and genetic counseling. The proposed course aims to train internists, pediatricians and obstetrician & gynecologists in this super-specialty and provide state of art medical genetics services to the patients and families with genetic disorders.

Eligibility for DM Medical Genetics Course

Post graduate degree in Medicine, Pediatrics or Obstetrics & Gynecology.

SUBJECT SPECIFIC OBJECTIVES

Competency based training for DM in Medical Genetics aims to produce a post graduate student who after undergoing the required training should be able to deal effectively with the needs of the patients, community and should be competent to handle medical problems related to genetic disorders. These include clinical evaluation, investigations, genetic work ups requiring pre-test and post-test counseling, up to-date information and abilities to carry out novel treatments and skills for planning and implementation of population-based prevention programs. Last but not least, be ready for carrying out clinical practice of personalized medicine in the 21st century molecular medicine era. The post graduate student should also acquire skills to teach Medical Genetics to undergraduates and paramedical students as well.

Thus, the objectives of DM in Medical Genetics course are to produce a competent Medical Geneticist who:

1. is a medical doctor who can evaluate a patient with possible genetic disorder, ascertain the risk of a genetic disorders, make a clinical diagnosis, able to decide appropriate test to confirm the diagnosis and provide latest form of treatment. The other goal is genetic counseling to the patient, family and extended family about management, carrier screening, prenatal diagnosis and prevention.
2. is aware of contemporary advances and developments in Medical Genetics, and ways for continued learning for keeping updated about diagnostic investigations, technological developments and treatment.

3. is trained in use of novel clinical ways of phenotyping, use of software for phenotyping and correlation with genotypes, interpretation of genetic variants and CNVs regarding their pathogenicity and causal nature and comfortable with the use of databases of genetic disorders, CNVs and genetic variants.
4. acquires the competencies pertaining to Medical Genetics that are required for practice in the community and at all levels of health system, especially population-based prevention program, screening for late onset disorder and susceptibility to cancer.
5. recognizes the health needs of patients and families with genetic disorders and carries out professional obligations in keeping with principles of the National Health Policy and professional ethics.
6. identify the disorders which are prevalent in Indian populations (identifying pockets).
7. is oriented to principles of research methodology.
8. has acquired skills in educating lay persons, medical and paramedical professionals.
9. has acquired skills in effectively communicating with the person, family and the community.
10. has acquired skills in DNA diagnostic tests including those for screening of carriers and antenatal diagnosis, as well as pre-morbid diagnosis for primary prevention (predictive/preventive medicine), being aware of the PCPNDT 2003 Act and related guidelines as well as processes.
11. has acquired knowledge and skills in genetic counseling and have developed confidence in psychological & social issues involved.

During the course, the student should acquire the following knowledge/skills/expertise:

A. Theoretical Knowledge:

The post graduate student in Medical Genetics must acquire knowledge in all aspects relevant to the practice of medical genetics. This includes clinical presentations of genetic disorders, organ specific genetic disorders, principles of basic genetics, genetic counseling, and knowledge of genes, chromosomes, genome and epigenome including molecular techniques. In whole, this is described as genomic medicine.

B. Clinical/Practical Skills:

The post graduate student must undergo training in all aspects relevant to the practice of Medical Genetics, be able to clinically evaluate, interpret genetic laboratory data and diagnose, investigate and manage patients with gene related conditions. This is similar to any other organ based clinical super-specialty. Here, the disorders for which the expertise in need are not specific to any organ or system; but are those where genetic etiology known. The steps and principles are similar to any clinical situation and involves [1] history taking and examination of a patient to draw up a differential diagnosis, [2] advise the relevant non-genetic and genetic investigations, and [3] treatment of the patient. However, genetic disorder has other implications for the patient and the family due to genetic nature of the etiology and medical geneticist has two more responsibilities, namely counseling the family for prevention and recurrence risks and prenatal diagnosis in appropriate conditions.

C. Teaching Skills:

Should be able to teach relevant aspects of genetic diseases to junior colleagues, nursing and para-medical staff. Population based screening programs like antenatal, neonatal screening and screening for susceptibility to late onset disorders are important components of medical genetics and advocacy, communication with lay persons and educating lay persons, society leaders, policy makers are the responsibilities of medical genetics community and require appropriate skills for teaching in informal ways.

D. Research Methodology:

Should be able to identify and investigate a research problem in Medical Genetics using appropriate methodology and molecular techniques. Most of the genetic disorders being rare, many cases even in clinical settings need research approach. This needs in depth understanding of deep phenotyping, genomic techniques, principles of ethics and guidelines for ethics of research. This needs access to and understanding of rules and regulations of Ethics Committee.

E. Group Approach:

There are some overlap of patients seen by medical geneticists with other organ-based specialists. The organ-based specialists may need expert opinion from medical geneticists about genetic disorders and medical geneticists may need clinical opinion and guidance /

support for management from the specialist like neurologist, ophthalmologist, etc. Hence ability to organize /participate in multi-disciplinary meetings with clinicians, laboratory colleagues and experts from allied clinical disciplines is necessary in the interest of the patients. The medical geneticist when counseling or acting as a consultant, must know the impact of genetic heterogeneity, pleiotropy, variable expression and lack of penetrance, so that the validity and accuracy of the diagnosis is assured. Therefore, the medical geneticist requires a broad exposure to genetic disease as it occurs in the area of many specialties/subspecialties so as to be competent to question, validate, or consider a new diagnosis. This knowledge will include the ability to appropriately use consultations with other specialists who may not be aware of these intricacies of genetic approaches.

The most important related specialties in which the student should gain experience include: Internal Medicine, Pediatrics, Obstetrics and Gynecology, Cardiology, Radiology (skeletal) including prenatal ultrasound, Endocrinology, Hematology, Oncology, Nephrology, Neurology, Physical Medicine & Rehabilitation, Psychiatry, Ophthalmology, Clinical Immunology, Clinical Pharmacology, and Orthopedics. Combined case wise interaction/discussion should be carried out for better understanding and management.

F. Attitudes including Communication Skills:

Should be able to communicate effectively with patients, colleagues and the community about genetic diseases as well as counsel patients and relatives about various decisions taken during management. The compassion, empathy, communication skills are intrinsic but vary in texture and amount amongst different persons. Though all physicians need these skills, their contribution is high for becoming a successful medical geneticist. These attitudes need to be developed and polished by observation, training and practice.

SUBJECT SPECIFIC COMPETENCIES

By the end of the course, the student should have acquired knowledge (cognitive domain), professionalism (affective domain) and skills (psychomotor domain) as given below:

A. KNOWLEDGE/COGNITIVE DOMAIN

By the end of the course, the post graduate student should have acquired knowledge in Core Discipline: Competencies unique to Medical Genetics

1. What genes are, how they are organized and controlled, what they do, and how they segregate, what is the structure and configuration of genome
2. The nature of mutations and pre-mutations and how they contribute to human variability and to disease
3. The patterns of inheritance characteristic of autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive traits
4. Factors that affect development of the phenotype in single-gene disorders, including variable expressivity and incomplete penetrance
5. The basis of mitochondrial diseases and the expected pattern for mitochondrial (maternal) inheritance, and other non-Mendelian form of inheritances like genomic imprinting, isodisomy, unstable/dynamic mutations, copy number variation, etc
6. Organization of genes chromosomes, how chromosomes replicate in mitosis and meiosis, and mode of their transmission from parent to child
7. Clinical manifestations of common numeric, structural and mosaic chromosomal anomalies
8. Concepts and clinical importance of genetic imprinting and uniparental disomy
9. Principles of population genetics and the public health implications of genetic epidemiology
10. Relevance of polymorphisms, gene linkage, and human gene mapping in medicine
11. The basic principles of inborn errors of metabolism and of pharmaco-genetic variations and their general clinical manifestations
12. The multi-factorial nature of human traits, both normal and abnormal, and the principles of multi-factorial inheritance
13. The mechanisms of teratogenesis and the effects of major human teratogens
14. Methods to recognize and classify congenital anomalies and the approach to diagnosis of multiple congenital anomaly syndromes

15. Role of genetics in the pathogenesis of neoplasms and in the predisposition to malignancies
16. Epigenetics
17. Use of evolutionary principles to understand human biology and disease
18. Disease frequency variations in different ethnic groups
19. Conventional as well as advanced molecular genetics, cytogenetics and metabolic genetic diagnostic techniques, and their application to genetic disorders
20. Techniques to study in vitro functional effects of genetic variations
21. Various databases of genes, genetic variations, genetic disorders and tools / software for studying effects of genetic variations, genotype phenotype correlations
22. Clinical presentations of organ specific genetic disorders and approaches to clinical phenotypes of genetic disorders
23. Traditional and novel approaches to the treatments of genetic disorders
24. The procedures available for prenatal & pre-implantation genetic diagnosis and the type of disease that can be detected prenatally/before implantation
25. Appropriate methods of genetic counseling
26. The advantages, limitations, and dangers of predictive testing for genetic disease
27. The existence of and justification for screening programs to prevent genetic disease
28. Approaches to screening for genetic diseases and concepts of personalized medicine
29. Organizational and economic aspects of the health care system with regard to genetics
30. Legal and ethical issues involved in the practice of medical genetics
31. Ability to recognize variations in human form (taking into account the features of the parents)
32. The history of use and misuse of human genetics

Problem Solving

The post graduate student should demonstrate ability to:

1. Take history including collecting complete family history in the form of a pedigree
2. Examine the patient for general findings, dysmorphism, systemic findings, evaluation of growth and development in a systematic way and with great sensitivity (examination of

parents, relatives for subtle findings, dysmorphism is a part of clinical evaluation of the patient. Evaluation of patient for dysmorphism and examination of relatives should be done with an extra sensitivity as this may psychological touchy points/ areas)

3. Interpret family history data
4. Identify and find information relevant to a clinical problem, using consultation, texts, and the archival literature and electronic media
5. Generate an initial list of differential diagnoses (including appropriate non-genetic disorders) given a specific complaint and patient characteristics
6. Re-rank the differential diagnoses based on information gathered from the history, physical, and auxiliary studies
7. Explain a mechanism for each aspect of a patient's problem, including biological, behavioral, and social aspects
8. Plan investigations and provide information about the need and utility of the genetic investigations along with the information about the diagnostic yield and limitations (also described as pretest counseling)
9. Providing appropriate information about phenotype of the patient in the form of Human Phenotype Ontology (HPO) terms to the laboratory while sending samples for genomic tests like cytogenetic microarray (CMA) and next generation sequencing (NGS)
10. Interpret the results of CMA, NGS. Reevaluate them based on various databases of variants, using software for pathogenicity testing and using guidelines and review of latest literature
11. Knowledge in the area of bioinformatics for DNA/ RNA interrogation
12. The results of the tests need clinical correlation and deep / reverse phenotyping after the results are available. Emphasis on Genetic results, as they are irreversible and heritable transmissions are known and are socially stigmatizing (important aspect in marriage counseling).
13. Evaluate scientific/clinical information and critically analyze conflicting data and hypotheses
14. Treat the patients with available modalities of treatment, look for latest drug developments

15. Communicate the diagnosis and uncertainties/limitations in diagnosis

Practice-Based Learning and Improvement

Physician Scholar

1. Demonstrates ability to analyze the quality and implications of medical literature and apply new knowledge in the delivery of health care
2. Demonstrates interest and ability to identify future areas of inquiry in medical genetics research
3. Demonstrates enthusiasm and positive attitude in the educational process and participates fully in educational activities
4. Demonstrates competency in use of genetic databases, software and bioinformatic tools
5. Write case summary and electronic health records for comprehensive data collection
6. Write referral notes to other specialists and primary care physician for continued management, surveillance for complications and sick day management

Health Care Management

1. Demonstrates **practical, efficient and cost-effective approach to screening, diagnosis and treatment** planning and recognizes its social and economic impact
2. Demonstrates ability to engage the patient family in diagnosis and therapeutic treatment planning
3. Demonstrates ability to recognize and outline initial treatment for patient with life threatening emergencies regardless of etiology
4. Consent should be informed written consent from the patient in the language they understand (English and vernacular) and records should be kept as per PCPNDT guidelines

Health Service Delivery

1. Demonstrates knowledge of health-care financing and applies it in assisting patient to access the best possible care
2. Utilizes knowledge of population based and evidence-based medicine in making patient management decisions

3. Works with patient support groups in the area of advocacy and as a communicator with the policy makers
4. Utilizes knowledge of managed care systems in making patient treatment plans and health care maintenance plans

Health Care Team approach to health care delivery

1. Demonstrates understanding of the roles and competencies of other health care providers
2. Demonstrates ability to engage with other health care professionals
3. Demonstrates training, allotment of work and supervision of paramedical workers like genetic counselor, social worker, genetic nurse, medical photographer
4. Demonstrates ability to follow and lead in a team approach to health care delivery

Academic Skills

1. Familiarity with basic research methodology, epidemiology, basic information technology skills, knowledge to access data and information systems, electronic health records
2. Writing a proposal for submission to Ethics Committee
3. Preparing Case Report Form for various presentations
4. Planning the protocol of a thesis, its execution and final report
5. Review of literature
6. Conducting clinical sessions for undergraduate medical students, nurses and paramedical workers
7. Writing and presenting a paper
8. Writing patient information brochures in local languages

B. AFFECTIVE DOMAIN

The post graduate student should:

1. Be able to function as a part of a team, develop an attitude of cooperation with colleagues, and interact with the patient and the clinician or other colleagues to provide the best possible diagnosis or opinion

2. Always adopt ethical principles and maintain proper etiquette in dealings with patients, relatives and other health personnel and to respect the rights of the patient including the right to information and second opinion
3. Develop communication skills to word reports and professional opinion as well as to interact with patients, relatives, peers and paramedical staff, and for effective teaching

Professionalism

1. Accepts personal responsibility for care of patients with genetic disorder, consistent with good work ethics and empathy
2. Demonstrates appropriate truthfulness and honesty with colleagues
3. Recognize personal beliefs, prejudices, and limitations, which should not come in the way of providing service
4. Respect patient confidentiality at all times in verbal and written communication

Interpersonal and Communication Skills

Human Relationships

1. Demonstrate an effective system for identifying and addressing ethical, cultural, and spiritual issues associated with health care delivery
2. Demonstrate knowledge or applies an understanding of psychological, social, and economic factors which are pertinent to the delivery of health care
3. Accurately assess a patient's assumptions in accessing the health care system
4. Effectively engage the patient and/or family in communications
 - a. Non-judgmental and non-coercive
 - b. Non-directive in genetic counselling

Attitudes

1. Appreciate the importance of disease prediction and prevention
2. Respect patients' religious, moral, and ethical beliefs and biases, even if they differ from the student's own beliefs
3. Present all available options accurately and non-directively

4. Be aware of both the importance of confidentiality and the difficulties that confidentiality poses when relatives are found to be at risk for a serious and potentially preventable disease
5. Be aware of the advantages and potential hazards of referring patients and families to community or national resources
6. Recognize the limitations of their own skills and seek consultation when necessary

C. PSYCHOMOTOR DOMAIN

The post graduate student must acquire the following practice-based competencies/skills:

1. Acquire ability to elicit medical history of the patient, including developmental and reproductive history
2. Ability to elicit the family history, including drawing of detailed pedigree chart
3. Ability to conduct physical examination of affected and related individuals (including examination of the affected fetus where required), with special emphasis on morphological features (with sensitivity and subtly, without offending the patient or parents' sensitivity about body image and dysmorphism) and anthropometric measurements and proper documentation of the findings, including photographs
4. Follow a logical approach in syndrome identification including the use of diagnostic aids e.g. computer assisted diagnosis, literature search, image analysis software, etc
5. Use the diagnostic support of the software logically using subjective interpretations and review of latest literature
6. Recognize psycho-social and economic implications of the genetic problem in the family
7. Integrate the clinical & genetic information to exclude the non-genetic causes of the clinical presentation (phenocopy) and formulate an appropriate differential diagnosis of genetic causes related to the case
8. Understand the uses, limitations, interpretation and significance of specialized laboratory procedures and formulate an appropriate plan for medical consultations and investigations, and conduct pre-test counseling with the family
9. Obtain samples for genetic studies e.g. skin biopsy (after obtaining the informed consent), requisition/conduct tests, medical consultations

10. Acquire skills to conduct all required cytogenetic, biochemical and laboratory tests, and discuss the results with clinician-in-charge of the patient to arrive at a logical conclusion on the disease and its management
11. Explain the diagnosis, etiology, natural history, and management of the condition to the patient and the family in a way that is comprehensible to the patient and family
12. Provide general, supportive and specific medical care to the patient including appropriate interventions where necessary
13. Provide client-centered counseling and anticipatory guidance
14. Provide psychosocial support including (a) areas of difficulty and conflict, (b) help families and individuals recognize and cope with their emotional and psychological needs, and (c) recognize situations requiring psychiatric referral
15. Identify and use community resources that provide medical, educational, financial and psychosocial support and advocacy
16. Determine the mode of inheritance and risk of occurrence and recurrence of the genetic condition/birth defect, and give appropriate information on the same to the patient and family, including availability of antenatal diagnosis and other reproductive options
17. Promote informed decision-making about further testing and management of the risk of occurrence/recurrence, including provision of antenatal diagnosis, if possible
18. Provide written documentation of medical, genetic and counseling information for families and other health professionals
19. Communicate to the patient / relatives at a level appropriate to the consultant, information concerning the medical implications and prognosis, the risks that apply, the options available, and to help the individual/ family choose an appropriate course of action for themselves
20. Identify psychosocial issues in the family and if need arises refer to a psychiatrist for evaluation and assistance
21. Acquire necessary management skills to coordinate the care of individuals affected with complex genetic conditions that will require a long-term multidisciplinary approach
22. Demonstrate mastery of adequate medical record keeping.

Syllabus

Course contents:

A. Cognitive domain

History of Medical Genetics

- Foundations of Medical Genetics Before 1956

- Growth and Development of Medical Genetics: 1956 to the Present

- The Future

Genetics in Medicine

- The Principles of Disease, Defining Disease, Prevention and Treatment

Nature and Frequency of Genetic Disease

- Frequency of Genetic Disease

- Single-Gene Disorders

- Multifactorial Disorders

- Somatic Cell Genetic Disorders

Genomics and Proteomics

- Genes and Human Disease

- Genomics

- Mapping the Human Genome

- Sequencing the Human Genome

- Current Approaches to Sequence Human Genome

- Cloning Human Disease Genes

- Sequence-Based Methods for Detecting Chromosomal Abnormalities

- Proteomics

Genome and Gene Structure

- Double Helix Structure, DNA Replication, Transcription, and Meiotic Recombination

- Organization of Genomic DNA

- Gene Structure and the Molecular Pathway of Gene Expression

Epigenetics

- Epigenetic Mechanisms: Chromatin, DNA Methylation and Long Noncoding RNAs

- Epigenetic Reprogramming

- Epigenetic Regulation of X Inactivation

- Genomic Imprinting

- Genetic Disorders Due to Genes Affecting Chromatin Structure

- Methods for Studying Epigenetic Marks

- Cancer Epigenetics

- Environmental Influences on Epigenetic Traits

- Abnormalities in Epigenetic Programming Linked to Infertility and ART

In Utero Epigenetic Programming of Adult Traits and Disease
Genetic–Epigenetic Interactions

Human Gene Mutation in Inherited Disease: Molecular Mechanisms and Clinical Consequences
Molecular Mechanisms of Mutation Causing Human Inherited Disease
Disease-Causing Mutations, Consequences of Mutations
General Principles of Genotype–Phenotype Correlations

Genes in Families

Pedigree Construction
Unifactorial Inheritance/Single-Gene Disorders
Dominance and Recessiveness
Autosomal-Dominant Inheritance
Autosomal Recessive Inheritance
Sex-Linked Inheritance
X-Linked Recessive Inheritance
X-Linked Dominant Inheritance
Y-Linked (Holandric) Inheritance
Partial Sex Linkage

Analysis of Genetic Linkage

Linkage Analysis: Basic Concepts
Extending Parametric Linkage Analysis
Linkage Analysis for Complex and Quantitative Traits

Chromosomal Basis of Inheritance

Chromosome Structure, Chromosomes in Cell Division
Methods for Studying Human Chromosomes
Functional Organization of Chromosomes
Sex Chromosomes and Sex Determination
Uniparental Disomy and Imprinting
Chromosome Abnormalities

Mitochondrial Genetics

Mitochondrial Biology and Genetics
Mitochondrial Etiology for Diseases and cancer
Therapeutic Approaches to Mitochondrial Disease

Multifactorial Inheritance and Complex Diseases

Determining the Genetic Component of a Trait
The International HapMap Project
Genome-Wide Association Studies
Association Methods/Statistical Analysis
Analysis of Rare Variants Using New Technologies
Integration of Genetic, Genomic, and Functional Data for Multifactorial Diseases

Population Genetics

- Hardy–Weinberg Law, Factors that affect Hardy–Weinberg Equilibrium
- Applications in Population Genetics

Pathogenetics of Disease

- The Scope of Abnormal Phenotypes: Disease and Malformation
- Multivariate Normal Distributions and the Threshold Model
- Pathogenetics of Refined Traits
- Molecular Pathogenetics

Human Developmental Genetics

- The Concept of Developmental Fields and Field Defects
- Cellular Signaling in Development
- Steps and Concepts in Embryonic Development
- Regulation of Gene Expression in Development
- Organogenesis

Twins and Twinning

- Determining Zygosity
- Incidence of Twins, Vanishing Twin, Structural Defects in Twins
- Twins in Genetic Studies
- Dizygotic Twins, Monozygotic Twins

The Molecular Biology of Cancer

- Genetic Basis of Cancer
- Viral Oncogenes
- Oncogenic Alleles in Human Cancers
- Tumor Suppressor Genes
- The Role of DNA Damage Repair Genes in Inherited Cancer Syndromes

The Biological Basis of Aging: Implications for Medical Genetics

- Progeroid Syndromes of Humans
- Human Allelic Variants Homologous to Pro-Longevity Genes

Pharmacogenetics and Pharmacogenomics

- Classical Genetics and Pharmacogenetics
- Ethnic Differences in Gene–Drug Interactions
- Pharmacogenomics

Genetic Evaluation for Common Diseases of Adulthood

- The Process of Genetic Evaluation for Common Diseases
- Integrating Genetic Information into Routine Clinical Practice

Genetic Counseling and Clinical Risk Assessment

- Process of Genetic Counseling

Adult-Onset Disorders
Genetic Risk Assessment and Calculation in the Clinical Setting

Cytogenetic Analysis

Milestones in Human Cytogenetics
The Indications for Cytogenetic Analysis
Tissue Samples and Cell Culture, Chromosome Banding
The Normal Human Karyotype
Chromosome Abnormalities
In situ Hybridization

Diagnostic Molecular Genetics

Indications for Molecular Genetic Testing
Technical Approaches to Molecular Genetic Testing
Molecular Genetic Diagnosis of Diseases
Mitochondrial DNA Disorders
Quality Assurance, and Regulatory Issues
Internet Resources for Molecular Genetic Testing

Heterozygote Testing and Carrier Screening

Carrier Screening in Clinical Practice
Carrier Screening in Individuals of Defined Subpopulation Groups
Therapeutic Implications for Heterozygotes
Methods and Tissues used in Carrier Identification
Problems in Heterozygote Detection
Sensitivity and Specificity, Cost and Feasibility
Age for Carrier Testing

Prenatal Screening for Neural Tube Defects and Aneuploidy

Techniques for Prenatal Diagnosis

Amniocentesis, Chorionic Villus Sampling, Fetal Blood Sampling, Fetal Tissue Sampling, Celomocentesis
Embryoscopy
Polar Body Biopsy, Preimplantation Genetic Diagnosis
Ultrasonography
Fetal Cells and Fetal DNA in Maternal Blood

Neonatal Screening

Historical Aspects
Components of Screening Programs
Potential Problems in Newborn Screening
Disorders and Conditions Detected by Newborn Blood Screening
Other Newborn Screening and Issues and Concerns in Screening

Therapies for Lysosomal Storage Diseases

- ERT for Lysosomal Storage Diseases
- Substrate Reduction Therapy
- Pharmacologic Chaperone Therapy

Gene Therapy: From Theoretical Potential to Clinical Implementation

- Genes as Medicines -The Origins of Gene Therapy
- The Basic Science: Gene Transfer
- Developing Cell-Type-Specific and Regulatable Gene Delivery Vectors
- The Clinical Science: Toward Gene Therapy of Human Disease

Ethical and Social Issues in Clinical Genetics

- Genetic Counseling, Testing and Screening
- Goals and Outcomes of Genetic Services
- Non-directiveness in Genetic Counseling
- Diagnostic Genetic Testing, Predictive Genetic Testing
- Confidentiality
- Genetic Testing in Childhood, Population Genetic Screening, Newborn Screening, Antenatal Screening, Carrier Screening
- Genetics, Geneticization and Society
- Reproductive Technologies and Cloning: “Reprogenetics”

Legal Issues in Medical Genetics

- Genetic Malpractice
- Genetic Counseling, Abortion, Adoption, Surrogacy, Embryo cryopreservation
- Newborn Screening
- Prenatal and Carrier Screening
- Genetic Discrimination
- Regulation of Genetic Diagnostic Tests
- Direct to Consumer Genetic Testing
- Regulation of Human Genetic Research
- Regulation of Research with Stem Cells Derived from Human Embryos

Genetics of Male & Female Infertility

- The Hypothalamic – Pituitary - Gonadal Axis
- Hypogonadism: Hypogonadotropic & Hypergonadotropic Hypogonadism
- Eugonadal Infertility
- Chromosome Anomalies and Gene defects

Fetal Loss

- Early Pregnancy Loss, Late Pregnancy Loss
- Evaluation and Management of Recurrent Abortion

Clinical Approach to the Dysmorphic Child

- Prenatal versus Postnatal Onset of Developmental Problems

Clinical Teratology

- Recognized Teratogenic Exposures

- Paternal Exposures and Maternal Exposures

Neurodevelopmental Disabilities: Global Developmental Delay, Intellectual Disability, and Autism

Abnormal Body Size and Proportion

- Pathologic Short Stature

- Pathologic Overgrowth

Susceptibility and Response to Infection

- Genome-Wide Association Studies and Human Infection

- Cell Surface Proteins, Intracellular Proteins, Extracellular Proteins

Transplantation Genetics

- The Physiologic Function of MHC Molecules

- The Structure of Human Histocompatibility Molecules

- Minor Histocompatibility Systems

- Serologic, Cellular and Molecular Methods for HLA Typing

- Clinical Significance of HLA Molecular Typing

- Genetics of Xenotransplantation

- Stem Cells and Transplantation

The Genetics of Disorders Affecting the Premature Newborn

- Respiratory Distress Syndrome, Bronchopulmonary Dysplasia

- Patent Ductus Arteriosus

- Intraventricular Hemorrhage

- Retinopathy of Prematurity

- Necrotizing Enterocolitis

Disorders of DNA Repair and Metabolism

- Disorders of Nucleotide Excision Repair: Xeroderma Pigmentosum and Cockayne Syndrome

- Disorders of Base Excision Repair: MUTYH and Colon Cancer Risk

- Disorders of Mismatch Repair: Lynch Syndrome and Turcot Syndrome

- Disorders Associated with Double Strand Break Recognition and Repair: Ataxia-Telangiectasia and Related Conditions

- Crosslink Repair and Homologous Recombination Defects: Breast-Ovarian Cancer and Fanconi Anemia

- Disorders Associated with Recq Helicase Deficiency: Bloom, Werner, and Rothmund-Thomson Syndromes

- Gene - Environment Interactions: Gorlin - Goltz Syndrome

Autosomal Abnormalities

- Genetic Counseling in the Trisomies

Down Syndrome (Trisomy 21), Trisomy 18, Trisomy 13
Translocations
Uniparental Disomy
Deletion, Duplication

Sex-Chromosome Abnormalities

Turner Syndrome, Klinefelter Syndrome, 47,XXX Syndrome, 47,XYY Karyotype
Sex Chromosome Mosaicism
Sex Chromosome Tetrasomy and Pentasomy (Polysomy)
Structural Abnormalities of the Y Chromosome
Prenatal Diagnosis of Sex Chromosome Abnormalities

Cardio-vascular system: Congenital Heart Defects and Inherited Cardiomyopathies

Specific Syndromes with Congenital Heart Defect
Chromosomal Disorders
Microdeletions/Microduplication Syndromes
Single-Gene Disorders
Holt–Oram syndrome
CHARGE Syndrome
Maternal Diabetes, Drug Ingestion
Folic Acid Supplementation
Risks for Sibs and Offspring of Children with Isolated Heart Defects
Hypertrophic, Dilated and Atypical Cardiomyopathy
Hereditary Hemorrhagic Telangiectasia (Osler–Weber–Rendu Syndrome)
Hereditary Disorders of the Lymphatic System and Venous System (varicose vein)
Capillary Malformation/Arteriovenous Malformation (Capillary Malformation, Sturge–Weber Syndrome, Capillary Malformation–arteriovenous Malformation, Cerebral Cavernous Malformation)
The Genetics of Cardiac Electrophysiology in Humans
Genetics of Blood Pressure Regulation
Preeclampsia
Common Genetic Determinants of Coagulation and Fibrinolysis (Genetic Variants Influencing Components of the Coagulation Cascade, Genetic Variants Influencing Natural Anticoagulants, Genetic Variants Influencing Components of the Fibrinolytic Cascade, Genetic Variants Influencing Platelet Function, Genome-Wide Association Analysis for Thrombosis)
Genetics of Atherosclerotic Cardiovascular Disease (Genetic Studies of CHD, Candidate Gene Studies in Humans, Genome Wide Association Studies, GWAS Findings for CVD Risk Factors, Genetic Risk Scores and Prediction Algorithms for Personalized Medicine)

Respiratory tract disorders

Cystic Fibrosis
Genetic Underpinnings of Asthma and Related Traits
Disorders of ciliary function
Hereditary Pulmonary Emphysema
Interstitial and Restrictive Pulmonary Disorders

Congenital Anomalies of the Kidney and Urinary Tract

- Cystic Diseases of the Kidney
- Nephrotic Disorders
- Renal Tubular Disorders
- Cancer of the Kidney and Urogenital Tract

Gastrointestinal Tract and Hepatobiliary Duct System

- Inflammatory Bowel Disease
- Bile Pigment Metabolism and its Disorders including cholestasis
- Cancer of the Colon and Gastrointestinal Tract

Blood

- Hemoglobinopathies and Thalassemia
- Other Hereditary Red Blood Cell Disorders
- Hemophilia and Other Disorders of Hemostasis
- Rhesus and Other Fetomaternal Incompatibilities
- Disorders of bone marrow aplasia and dyserythropoiesis

Immunologic Disorders: Autoimmunity: Genetics and Immunologic Mechanisms

- Immunodeficiency Disorders
- Inherited Complement Deficiencies
- Disorders of Leukocyte Function
- Genetic Basis of Autoimmune Thyroid Disease

Endocrine

- Abnormalities of growth hormone- pituitary axis
- Monogenic diabetes mellitus
- Susceptibility to type I and type II diabetes
- Genetic Basis of Thyroid Carcinoma
- Familial Hypocalciuric Hypercalcemia
- CASR Mutations in Familial Hypocalciuric Hypercalcemia and Neonatal Severe Hyperparathyroidism
- Neonatal Hyperparathyroidism
- Multiple Endocrine Neoplasia
- Familial Isolated Hypoparathyroidism
- NHERF1 Mutations and Renal Responsiveness to Parathyroid Hormone

Adrenal Gland

- Congenital Adrenal Hyperplasia (21, 11 β , 3 β , 17 α -Hydroxylase Deficiency, 17,20-Lyase Deficiency, Congenital Lipoid Adrenal Hyperplasia, etc): Prenatal Diagnosis and Treatment
- Congenital adrenal hypoplasia

Reproductive system

- Disorders of the Gonads, Genital Tract, and Genitalia
- Disorders of Sexual Development and differentiation

Hereditary Cancers

- Familial Breast Cancers (BRCA1, BRCA2)
- Familial Breast or Ovarian Cancer
- Familial Ovarian Cancer
- Familial Endometrial Cancer
- Hereditary Nonpolyposis Colorectal Cancer Syndrome (or Lynch Syndrome)
- Li Fraumeni syndrome

IEM Amino Acid Metabolism

- Disorders of Phenylalanine Metabolism
- Disorders of Tyrosine Metabolism
- Disorders of Glycine Metabolism
- Disorders of Proline and Hydroxyproline
- Disorders of the Urea Cycle and Ornithine
- Disorders of Serine Metabolism

IEM Disorders of Carbohydrate Metabolism

- Disorders of Galactose Metabolism
- Disorders of Fructose Metabolism
- Disorders of Pentose Metabolism
- Glycogen Storage Diseases
- Gluconeogenic Disorders Associated with Lactic Acidosis

Congenital Disorders of Protein Glycosylation

- Congenital Disorders of Protein N-Glycosylation
- Congenital Disorders of Protein O-Glycosylation
- Congenital Disorders of Protein N- and O-Glycosylation

Purine and Pyrimidine Metabolism

Lipoprotein and Lipid Metabolism

- Monogenic Disorders of Lipoprotein Metabolism
- Disorders with Primarily Elevated LDL Cholesterol
- Disorders with Primarily Depressed LDL Cholesterol
- Disorders with Primarily Elevated HDL Cholesterol
- Disorders with Primarily Depressed HDL Cholesterol
- Disorders with Primarily Elevated Triglycerides
- Disorders with Multiple Lipoprotein Disturbances

Organic Acidemias and Disorders of Fatty Acid Oxidation

Organic Acidemias

Vitamin D Metabolism or Action

- Hereditary Vitamin D Dependency Type 1 (VDDR-1)-1- α -Hydroxylase Deficiency
- Hereditary Vitamin D-Dependent Rickets Type 2 (VDDR-2)
- States Resembling Hereditary Generalized Resistance to 1,25(OH)₂D

Inherited Porphyrias

- Regulation of Heme Biosynthesis
- Classification and Diagnosis of the Porphyrias

Inherited Disorders of Human Copper Metabolism

- Menkes Disease
- Wilson Disease

Iron Metabolism and Related Disorders

- Syndromes of Iron Overload
- Other Disorders Resulting in Derangements of Iron Handling

Mucopolysaccharidoses

- Mucopolysaccharidosis I (IH Hurler, IS Scheie and IH/S Hurler–Scheie Disease)
- Mucopolysaccharidosis II (Hunter Syndrome)
- Mucopolysaccharidosis IIIA (Sanfilippo Syndrome, MPS IIIA)
- Mucopolysaccharidosis IIIB (Sanfilippo Syndrome, MPS IIIB)
- Mucopolysaccharidosis IIIC (Sanfilippo Syndrome, MPS IIIC)
- Mucopolysaccharidosis IIID (Sanfilippo Syndrome, MPS IIID)
- Mucopolysaccharidosis IVA and IVB (Morquio Syndrome, MPS IVA, MPS IVB)
- Mucopolysaccharidosis V (Scheie Syndrome, MPS V)
- Mucopolysaccharidosis VI (Maroteaux–Lamy Syndrome, MPS VI)
- Mucopolysaccharidosis VII (Sly Syndrome, MPS VII)
- Mucopolysaccharidosis VIII
- Mucopolysaccharidosis IX (Natowicz Syndrome, MPS IX)

Oligosaccharidoses: Disorders Allied to the Oligosaccharidoses

Sphingolipid Disorders and the Neuronal Ceroid Lipofuscinoses or Batten Disease (Wolman Disease, Cholesteryl Ester Storage Disease, and Cerebrotendinous Xanthomatosis)

- GM1-Gangliosidosis (β -Galactosidosis)
- GM2-Gangliosidosis
- Loss-of-Function Mutation of GM3-Synthase
- Niemann–Pick Disease
- Niemann–Pick Disease, Types A and B
- Niemann–Pick Disease, Types C and D
- Farber’s Disease
- Acid Lipase Deficiency (Wolman Disease and Cholesteryl Ester Storage Disease)
- Gaucher Disease
- Galactosylceramide Lipidosis, Globoid Cell Leukodystrophy, or Krabbe Disease
- Metachromatic Leukodystrophy
- Fabry Disease
- Neuronal Ceroid Lipofuscinosis or Batten Disease
- Kufs Disease or Adult NCL
- Congenital NCL/CNCL-CLN10/Cathepsin D or CTSD Deficiency

Peroxisomal Disorders

Nervous System (CNS & PNS)

- Fragile X Syndrome and X-linked Intellectual Disability
- Dyslexia and Related Communication Disorders
- Attention-Deficit/Hyperactivity Disorder
- Autism Spectrum Disorders
- Genetics of Alzheimer Disease
- Schizophrenia and Affective Disorders
- Addictive Disorders
- Neural Tube Defects
- Genetic Disorders of Cerebral Cortical Development
- Genetic Aspects of Human Epilepsy
- Basal Ganglia Disorders (Parkinson Disease, Dystonias, Choreic Disorders)
- Hereditary Ataxias (Autosomal-Dominant, Recessive, X-linked ataxias, Intermittent Ataxias, Episodic Ataxias, Progressive Ataxias, Mitochondrial Ataxias)
- Hereditary Spastic Paraplegia (Autosomal Dominant, Autosomal Recessive, X-Linked)
- Autonomic and Sensory Disorders (Familial Dysautonomia, Congenital Sensory Neuropathy with Anhidrosis)
- The Phakomatoses (The Neurofibromatoses)
- Tuberous sclerosis
- Demyelinating Disorders (Krabbe Disease, Metachromatic & X-Linked Adrenoleukodystrophy)
- Hereditary Motor and Sensory Neuropathies (Charcot–Marie–Tooth)
- Spinal Muscular Atrophies
- Motor Neuron Disease: Familial Amyotrophic Lateral Sclerosis

Muscles

- Muscular Dystrophies (Dystrophinopathies, Facioscapulohumeral Muscular Dystrophy Emery–Dreifuss Muscular Dystrophies and Other Contractural Phenotypes, Limb-Girdle Muscular Dystrophies, Myofibrillar Myopathies and Other Distal Phenotypes, Congenital Muscular Dystrophies)
- Congenital (Structural) Myopathies
- Hereditary Muscle Channelopathies
- Myotonic Dystrophies
- Hereditary and Autoimmune Myasthenias

Eye

- Color Vision Defects (Molecular Basis of Variation in Normal Color Vision, Genetics of Red–green Color Vision in Women, Blue–Yellow (Tritan) Color Vision Defects, etc)
- The Achromatopsias
- Optic Atrophy
- Glaucoma
- Congenital Defects of the Cornea
- Congenital Cataracts and Genetic Anomalies of the Lens

Hereditary Retinal and Choroidal Dystrophies: Pigmentary Retinopathies/Retinitis Pigmentosa, Leber's Congenital Amaurosis, The Primary Cone Degenerations
Strabismus
Retinoblastoma and the RB1 Cancer Syndrome
Anophthalmia, Microphthalmia, and Uveal Coloboma

Ear: Hereditary Hearing Impairment

Clefting, Dental, and Craniofacial Syndromes

Craniosynostosis

Skin and Hair

Abnormalities of Pigmentation: Disorders of Melanosome Biogenesis/Transport—Hermansky–Pudlak Syndrome, Chédiak–Higashi Syndrome, and Griscelli Syndrome; Disorders of Melanocyte Survival—Vitiligo
Ichthyosiform Dermatoses
Epidermolysis Bullosa
Ectodermal Dysplasias
Xeroderma Pigmentosum
Epidermodysplasia Verruciformis
Porokeratoses
Muir–Torre Syndrome
Melanoma
Cutaneous Hamartoneoplastic Disorders: Hereditary Leiomyomatosis and Renal-Cell Cancer, Birt–Hogg–Dubé Syndrome
Inherited Disorders of the Hair: Hypotrichosis, Hypertrichosis
Marfan Syndrome and Related Disorders
Ehlers–Danlos Syndrome
Heritable Diseases Affecting the Elastic Fibers: Cutis Laxa, Pseudoxanthoma Elasticum, and Related Disorders

Bone

Osteogenesis Imperfecta (and Other Disorders of Bone Matrix)
Disorders of Bone Density, Volume, and Mineralization: Osteopetrosis Group of Disorders, Raine Dysplasia, Pyknodysostosis, Dysosteosclerosis, Osteopoikilosis, Craniotubular Remodeling Disorders, Hyperphosphatasemia with Osteoectasia, etc
Chondrodysplasias
Abnormalities of Bone Structure (Dysplasia Epiphysealis Hemimelica, Hereditary Multiple Exostoses, Langer–Giedion Syndrome, Enchondromatosis, Maffucci Syndrome, Metachondromatosis, Fibrous Dysplasia of Bone, etc)
Dysostoses
Arthrogyposes (Multiple Congenital Contractures)
Common Skeletal Deformities
Hereditary Noninflammatory Arthropathies
Cohesinopathies

Ciliopathies

B. Psychomotor domain:

The student should be able to understand, learn and/or perform:

1. Elicit a comprehensive medical genetic history and construct an appropriate pedigree
2. Carry out a comprehensive physical examination for major and minor anomalies, with special attention to surface anatomy and anthropometric measurements
3. Understand prenatal diagnostic procedures both invasive (amniocentesis, chorionic villous sampling, cordocentesis, etc) and non-invasive (fetal ultrasonography)
4. Perform fetal autopsy
5. Understand principle of cytogenetic, biochemical, and molecular laboratory methods
6. Perform specialized tests through biochemical, cytogenetic, and molecular genetic laboratories
7. Learn interpretation of cytogenetic, biochemical, and molecular laboratory reports
8. On the basis of results, formulate an appropriate diagnosis
9. Effectively use information systems, including library and electronic resources, in the evaluation and management of patients with genetic diseases, including diagnosis of multiple congenital anomaly syndromes, and the recognition of teratogenic exposures.
10. Ensure that the testing is done in an Accredited Lab Only (Authenticity and legal protection for the referring clinician). Interpretation of the results is an art and science and hence should be included empathetically.
11. Appreciate the role of biomedical research and develop techniques for critical analysis of current scientific developments
12. Able to coordinate information from multiple sources into a coherent and rational plan of management of genetic disorders
13. Able to communicate and counsel patients and families who sometimes may be disturbed and psychologically upset
14. Understand human behavior, maturation, and intelligence, in order to facilitate counseling of varying individuals

Management of genetic diseases

The student should be able to:

1. Provide patients with access to diagnostic and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of such tests.
2. Demonstrate use and correct interpretation of diagnostic procedures and their results
3. Advise appropriate treatments, including dietary, pharmacological, enzyme-replacement, transplantation, and gene therapy.
4. Communicate genetic information in a manner that is suitable for each particular patient and family. Present all available option accurately with consequences explained and non directively.
5. Tolerate and encourage reiteration of information because of patient anxiety or unfamiliarity with the concepts being presented
6. Apply appropriate communication techniques for conveying bad news
7. Recognize patients' defense mechanisms and be able to determine when it is better to leave them intact and when they may need to be breached
8. Cope emotionally with responses of patients
9. Interpret their own attitudes toward ethical, social, cultural, religious, and ethnic issues and develop an ability to individualize each patient or family member.
10. Resolve varying cultural, social, and religious attitudes in relation to issues such as contraception, abortion, parenting, and gender roles
11. Utilize community support services and agencies appropriately
12. Plan and give an appropriate surveillance/ follow-up monitoring plan for anticipated complications and issues
13. Understand all the associated and anticipated multisystem manifestations in order to refer for appropriate multidisciplinary management
14. Participate in treating genetic diseases where applicable; for eg.,Lysosomal Storage Diseases (enzyme replacement, substrate reduction & Pharmacologic Chaperone Therapy), hypophosphatemia (burosumab), Multiple Exostoses (palovarotene), Metaphyseal Chondrodysplasia Schmid type (carbamazepine), Osteoporosis Pseudoglioma (lithium carbonate).

TEACHING AND LEARNING METHODS

Clinical Training

Clinical training must provide students with opportunities to have first-hand experience with individuals and families affected by a broad range of genetic disorders. These clinical experiences must expose post graduate students to the natural history and management of common genetic conditions and birth defects and to relevant psychosocial issues. Students must have the opportunity to develop counseling and psychomotor skills in a variety of clinical settings where genetic services are provided. Such settings must include those where patients are seen for prenatal diagnosis; clinical and laboratory diagnosis of genetic syndromes and mental retardation; multidisciplinary management of chronic genetic disorders and birth defects; newborn, carrier or predisposition screening and evaluation of mutagenic, teratogenic or other risk factors. These clinical experiences must help students to observe and practice skills relating to obtaining medical and family histories; determining risks; performing psychosocial assessments; communicating information about disease characteristics, inheritance and natural history; providing anticipatory guidance and supportive counseling; identifying and using medical and community resources; communicating information to other health-care professionals, case management and follow-up. The students should become proficient in communicating the genetic burden and reproductive options including antenatal diagnosis to the patient and relatives.

Students must be supervised by qualified genetics professionals and given the opportunity for involvement in a variety of cases. Throughout clinical posting, student should maintain clinical/case management records in log book.

Rotation: Postings:

- 24 months of training in a clinical genetic unit where the post graduate student should participate in the care of at least 100 patients with various genetic disorders besides three months posting in fetal medicine, obstetrics & neonatology/pediatrics
- 12 months of posting in Genetics laboratory

Laboratory Training

The students must attain proficiency in carrying out prescribed list of laboratory tests themselves and attain overall ability to organize, supervise and report on results of various specialized genetic

laboratory investigations employed in workup of patients with genetic disorders (this posting may run concurrently with clinical training). For this purpose, laboratory posting should be scheduled as given below:

- a) 2 months of training in Cytogenetics including molecular cytogenetic techniques namely FISH (fluorescence *in situ* hybridization), microarray, QF PCR, MLPA
- b) 2 months of training in biochemical genetics including Newborn Screening
- c) 2 months of training in molecular genetics including PCR, Sanger sequencing, analysis of Next Generation Sequencing data, reverse transcription PCR (RT PCR), Real Time PCR
- d) 6 months Research Project work

These rotations shall focus not only on developing consultative/interpretive skills, but also will provide adequate opportunity for hands-on training. The time spent in the laboratory towards research project should be entered in Log book. During postings, students should be exposed to the following laboratory techniques:

- Cytogenetics including Prenatal Cytogenetic Diagnosis
- Molecular Cytogenetics including Prenatal, Preimplantation and Cancer
- Microarray including molecular karyotyping
- Molecular Techniques including Polymerase Chain Reaction (PCR), Gel Electrophoresis (DNA/RNA i.e., ribonucleic acid, Protein, etc), Southern blotting, Arms technique, Real Time PCR, Quantitative Fluorescent PCR (QF PCR), DNA Sequencing, next generation sequencing (NGS), epigenetics techniques,

Teaching methodology

1. This should include regular case presentations, didactic lectures, seminars, journal clubs, clinical meetings, and combined conferences with allied departments. Hours of lecture must be 6-8 hours a week as doctors undergoing this training are not exposed to basic genetics in their basic graduation
2. The post graduate student should be given the responsibility of managing and caring for patients in a gradual manner under supervision.
3. Department should encourage e-learning activities.
4. Formal teaching sessions

In addition to bedside teaching rounds, at least 5-hr of formal teaching per week are necessary. The departments may select a mix of activities as given under formative assessment. The students should also attend:

- Attend accredited scientific meetings (CME, symposia, and conferences).
 - Additional sessions on basic sciences, biostatistics, research methodology, teaching methodology, hospital waste management, health economics, medical ethics and legal issues related to medical practice.
4. There should be a training program on Research methodology for existing faculty to build capacity to guide research.
 5. The post graduate students shall be required to participate in the teaching and training programme of undergraduate students and interns.
 6. A post graduate student of a post graduate degree course in broad specialities/super specialities would be required to present one poster presentation, to read one paper at a national/state conference and to present one research paper which should be published/accepted for publication/sent for publication during the period of his post graduate studies so as to make him eligible to appear at the post graduate degree examination.
 7. **Log book:** During the training period, the post graduate student should maintain a Log Book indicating the duration of the postings/work done. This should indicate the procedures assisted and performed, and the teaching sessions attended. The Log book shall be checked and assessed periodically by the faculty members imparting the training.

Research work

Research Project: Every post graduate student shall carry out research work on assigned research project under the guidance of a Post Graduate Teacher, the result of which shall be written up and submitted to the institution. Ensure that the topics are selected to focus on genetic disorders in India (common and rare)

During the training programme, patient safety is of paramount importance; therefore, skills are to be learnt initially on the models, later to be performed under supervision followed by performing independently; for this purpose, provision of skills laboratories in medical colleges

is mandatory. Various basic courses organized by the Institute for all round development of the clinician should be attended for increasing knowledge.

ASSESSMENT

FORMATIVE ASSESSMENT, during the training program

Formative assessment should be continual and should assess medical knowledge, patient care, procedural & academic skills, interpersonal skills, professionalism, self-directed learning and ability to practice in the system.

General Principles

Internal Assessment should be frequent, cover all domains of learning and used to provide feedback to improve learning; it should also cover professionalism and communication skills.

The Internal Assessment should be conducted in theory and clinical examination.

Quarterly assessment during the MD training should be based on:

1. Journal based / recent advances learning
2. Patient based /Laboratory or Skill based learning
3. Self-directed learning and teaching
4. Departmental and interdepartmental learning activity
5. External and Outreach Activities / CMEs

The student to be assessed periodically as per categories listed in postgraduate student appraisal form (Annexure I).

SUMMATIVE ASSESSMENT, i.e., assessment at the end of training

The summative examination would be carried out as per the Rules given in POSTGRADUATE MEDICAL EDUCATION REGULATIONS, 2000.

The final examination will be in three parts:

1. Thesis

Every post graduate student shall carry out work on an assigned research project under the guidance of a recognized Post Graduate Teacher, the result of which shall be written up and

submitted in the form of a Thesis. Work for writing the Thesis is aimed at contributing to the development of a spirit of enquiry, besides exposing the post graduate student to the techniques of research, critical analysis, acquaintance with the latest advances in medical science and the manner of identifying and consulting available literature.

Thesis shall be submitted at least six months before the Theory and Clinical / Practical examination. The thesis shall be examined by a minimum of three examiners; one internal and two external examiners, who shall not be the examiners for Theory and Clinical examination. A post graduate student shall be allowed to appear for the Theory and Practical/Clinical examination only after the acceptance of the Thesis by the examiners.

2. Theory Examination:

The examinations shall be organized on the basis of 'Grading' or 'Marking system' to evaluate and to certify post graduate student's level of knowledge, skill and competence at the end of the training. The examination for MD/MS shall be held at the end of third academic year.

There shall be four Theory papers.

Paper I: Basic Sciences as applied to Genetics

Paper II: Clinical Genetics
(e.g., approach to genetic disorder diagnosis, counseling and management)

Paper III: Applied/Laboratory Genetics
(e.g., genetics of various organ systems disorders, pre-implantation genetic screening/diagnosis, non-invasive prenatal screening, prenatal diagnosis, chromosomal microarray in dysmorphology, next generation sequencing, gene panel test, diagnosis of triplet repeat disorder, common and rare genetic disorders in India)

Paper IV: Recent Advances in Genetics

3. Clinical/Practical Examination

Clinical Case Presentation:

Long Case: 1

Short Cases: 3

Genetic Counseling

Laboratory tests/other skills (10 tests/skills): (including OSCE / OSPE stations)

- Cytogenetics: slide reporting
- Molecular Cytogenetics: slide reporting
- DNA electrophoresis
- Primer designing
- Hemoglobin A2 Estimation
- Hemoglobin Electrophoresis
- Other Biochemical Genetics Tests
- Syndrome/disease/findings identification
- (slide/spot/photograph/specimen/radiologic/gel/electropherogram/etc)
- Syndrome diagnosis using the databases and software
- Pedigree Construction (5 generation)
- Analysis of Sanger sequence chromatograms
- Analysis of MLPA electropherogram
- Analysis of TP-PCR and QF-PCR electropherograms
- Analysis of chromosomal microarray results
- Analysis of NGS data
- In silico analysis of sequence variants and CNVs including use of population databases, mutation databases, & mutation prediction software
- Analysis and interpretation of TMS, urine GCMS and plasma amino acid HPLC results

3. Oral/viva voce examination

Oral examination shall be comprehensive enough to test the post graduate student's overall knowledge on the subject

RECOMMENDED READING

Books (latest edition)

1. Emery and Rimoin's Principles and Practice of Medical Genetics and Genomics. Editors: Reed Pyeritz, Bruce Korf, Wayne Grody. Publisher: Elsevier. 6th Edn. 2013. (ISBN:9780123838353).

2. Practical Genetic Counseling. Editors: Peter Harper, Butterworth Heinmann. Publisher: Edward Arnold Ltd. 3rd Edn. 2013. (ISBN: 9781483183664).
3. Smith's Recognizable Pattern of Human malformations. Editor: Kennethlyon Nenes. Publisher: WB Saunders Company. 7th Edn. 2013. (ISBN: 1455738115).
4. Mendelian Inheritance in Man. Editor: Victor A Mckusick. Publisher: The Johns Hopkins University Press. 2014. (ISBN: 9780801820878).
5. Metabolic and Molecular Basis of Inherited Disease. Editors: Charles R. Scriver, Arthur L. Beaudet, William S. Sly, David Valle, Mc Graw-Hill. Publisher: McGraw-Hill. 8th Edn. 2001. (ISBN10 0079130356).
6. The Principles of Clinical Cytogenetics. Editors: SL Gersen, M B Keagle, Publisher: Humana Press. 3rd Edn. 2013. (ISBN 978-1-4419-1688-4).
7. Textbook of Fetal and Perinatal Pathology, Vol. I & II. Editors: Jonathan S Wigglesworth, Don B Singh. Publisher: Wiley–Blackwell. 2nd Edn. 1998. (ISBN-10:0865421188).
8. Catalog of Teratogenic Agents. Editors: TS Shepard. Publisher: John Hopkins University Press. 13th Edn. 2011. (ISBN-13:978-0801897849).
9. Emery's Elements of Medical Genetics. Editors: Peter Turnpenny, Sian Ellard. Publisher: Elsevier Churchill Livingston. 15th Edn. 2017. (ISBN: 9780702066856).
10. The Practical Guide to the Genetic Family History. Editors: Bennett Robin. Publisher: Wiley-Liss. 1st Edn. 2010. (ISBN: 0471251542).
11. Chromosomal abnormalities and genetic counseling. Editors: RjJM Gardner, GR Sutherland. Publisher: Oxford university press. 5th Edn. 2018. (ISBN: 9780199329007).
12. Medical Cytogenetics. Editors: Mark Hon Fong L. Publisher: Marcel Dek. CRC Press. 1st Edn. 2000. (ISBN: 0824719999).
13. Congenital malformation evidence-based evaluation and management. Editors: Pravin Kumar, Barbara K Burton. Publisher: McGraw Hill. 1st Edn. 2007 (ISBN:0071471898).
14. Human Molecular Genetics. Editors: Tom Strachan, Andrew Read. Publisher: Garland Science. 5th Edn. 2018. (ISBN 978-0815345893).
15. Genetic disorders and the foetus: diagnosis, prevention and treatment. Editors: Aubrey Milunsky Publisher: Wiley. 7th Edn. 2015. (ISBN: 978-1-1189-8152-8).
16. Management of genetic syndromes. Editors: Cassidy SB, Allanson JE. Publisher: Wiley liss. 4th Edn. 2020. (ISBN: 978-1-119-43267-8).
17. Essentials of Medical Genomics. Editors: Brown Stuart M. Publisher: Wiley-Liss. 1st Edn. 2008. (ISBN: 047121003X)
18. New Clinical Genetics. Editors: Andrew Read and Dian Donnai. Publisher: Scion publishing. 4th Edn. 2020. (ISBN 9781911510703).
19. Human malformations and related anomalies. Editors: Stevenson RE, Hall JG. Publisher: Oxford university press. 3rd Edn. 2015. (ISBN-13: 9780199386031).
20. Radiology of syndromes, metabolic disorders and skeletal dysplasias. Editors: Taybi H, Lachman RS. Publisher: Elsevier. 5th Edn. 2006. (ISBN-13: 978-0815187097).

Journals

10-15 international Journals and 02-05 national (all indexed) journals

Miscellaneous Resources (including web resources)

1. OMIM - Online Mendelian Inheritance in Man, Catalog of all known human genes and genetic phenotypes (The Johns Hopkins University School of Medicine)
2. LMD (London Medical Databases), POSSUM, etc dysmorphology database
3. DECIPHER, ISCA, DGV, UPD, ICCG, etc database
4. GeneClinics: Medical Genetics Knowledge Base, formerly (Genline), diagnosis, management and counseling for individuals and families with inherited conditions
5. GeneTests (formerly Helix) - DNA diagnostic testing and research information
6. ClinGen: The Clinical Genome Resource
7. The Hereditary Cancer Working Group (centralize and curate genetic knowledge in order to develop guidance for molecular diagnostic germline cancer testing)
8. TERIS (teratogen information system), University of Washington
9. Gene/Disease Specific Information (locus specific databases)
10. GeneCards, database of human genes, products and involvement in diseases
11. Merck Manual (professional version): General Genetics, Inheritance of Single-Gene Defects, Multifactorial Inheritance, Nontraditional Inheritance, Chromosomal Disorders, Mitochondrial DNA Abnormalities, Immunogenetics, Forensic Genetics, Genetic Therapy, Congenital Anomalies, Pedigree symbols, Pharmacogenetics, Cancer Genetics, Prenatal Genetic Evaluation and Counseling, etc
12. World Wide Web Biochemical Genetics Test List, University of California, San Diego, Biochemical Genetics
13. EuroGenTest, includes unites on genetic testing: quality management, information databases, public health, new technologies and education, new 5/07
14. Genetics and Cancer (<https://www.cancer.org/cancer/cancer-causes/genetics.html>)
15. Cytogenetic images and animations, Tokyo Medical College, Hironao Numabe, M.D.
16. Before You are Pregnant, March of Dimes (<https://www.marchofdimes.org/pregnancy/getting-ready-for-pregnancy-preconception-health.aspx>)
17. European Teratology Society (ETS)
18. Folic Acid and Prevention of NTD Educational Materials, by CORN education committee
19. Illinois Teratogen Information Service, Chicago

20. Preconception Screening and Counseling Checklist, March of Dimes
21. Policy statements, American College of Medical Genetics
22. Policy statements, American Academy of Pediatrics
23. Policy statements, American Society of Human Genetics
24. Genetic Screening, American Society for Reproductive Medicine
25. Drugs in Pregnancy and Lactation, Perinatology.com
26. Ethics and genetics in medicine. In: Bioethics for clinicians. Burgess MM, et al. CMAJ. 1998. 158:1309-13.
27. AAFP Core Educational Guidelines in Medical Genetics

Post graduate Students Appraisal Form

Pre / Para /Clinical Disciplines

Name of the Department/Unit :

Name of the PG Student :

Period of Training : FROM.....TO.....

| Sr. No. | PARTICULARS | Not Satisfactory | | | Satisfactory | | | More Than Satisfactory | | | Remarks |
|---------|--|------------------|---|---|--------------|---|---|------------------------|---|---|---------|
| | | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | |
| 1. | Journal based / recent advances learning | | | | | | | | | | |
| 2. | Patient based /Laboratory or Skill based learning | | | | | | | | | | |
| 3. | Self-directed learning and teaching | | | | | | | | | | |
| 4. | Departmental and interdepartmental learning activity | | | | | | | | | | |
| 5. | External and Outreach Activities / CMEs | | | | | | | | | | |
| 6. | Thesis / Research work | | | | | | | | | | |
| 7. | Log Book Maintenance | | | | | | | | | | |

Publications

Yes/ No

Remarks* _____

***REMARKS: Any significant positive or negative attributes of a postgraduate student to be mentioned. For score less than 4 in any category, remediation must be suggested. Individual feedback to postgraduate student is strongly recommended.**

SIGNATURE OF ASSESSEE

SIGNATURE OF CONSULTANT

SIGNATURE OF HOD