

Guidelines
for
Competency Based Training Programme
in
DNB-MEDICAL GENETICS



NATIONAL BOARD OF EXAMINATIONS

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INTRODUCTION

The National Board of Examinations was established in 1975 by the Government of India with the prime objective of improving the quality of the Medical Education by establishing high and uniform standards of postgraduate examinations in modern medicine on an all India basis.

Currently the DNB in Medical Genetics will be run as a **National Course**, funded by the Department of Biotechnology. Three institutions are participating in the course: 1). Center of Medical Genetics, Sir Ganga Ram Hospital, New Delhi, 2). Nizam's Institute of Medical Sciences, and Centre of DNA Fingerprinting and Diagnosis, Hyderabad , and 3). National Institute of Biomedical Genetics, and Institute of Postgraduate Medical Education and Research and SSKM Hospitals. The candidates will rotate through the various institutions during the course, as outlined below under curriculum

Medical Genetics is the speciality of medicine that involves the diagnosis and management of genetic disorders. With the recent dramatic advances in genomics and genetics, Medical Genetics has emerged as an important clinical speciality worldwide. It is true to say that all diseases have a genetic component, which varies with the disorder. All families with genetic disorders can be helped by making a precise diagnosis, appropriate management and proper counseling. In a resource poor country like India, there is a paramount need for preventing the birth of affected children, and reduce the socio-economic burden of genetic disorders through community control programs comprising genetic screening and prenatal diagnosis. To achieve these objectives a cadre of physicians and scientists trained in genetic medicine is required. This is especially important as Human Genome Project has generated lot of new information that can be applied to help patients and be the basis of personalized medicine. This National DNB Program in Medical Genetics, therefore, aims to fulfill the pressing need to train physicians in medical genetics, so that they can bring the benefits of modern biology and genomics to the people of India.

PROGRAMME GOAL

The National DNB Program in Medical Genetics will be designed to give physicians a thorough knowledge of the principles and practice of medical genetics, and prepare them for a leadership role in training other medical geneticists for service and research.

The candidates will be provided hands-on training in modern genetic technologies such as PCR, Sanger sequencing, massively parallel sequencing, micro-arrays, enzyme assays, FISH and Luminex multiplexing etc. They will be imparted knowledge of the principles of epidemiology and statistics as applied to genetic and genomic research. They will be given the opportunity to carry out research on a chosen topic. They will learn ethical principles as applied to genetic services and practice.

The main goals of the National DNB Program in Medical Genetics are:

- *Impart* training that will enable the trainees to evaluate patients with genetic disease, order appropriate tests, interpret them for the patient, make a precise diagnosis and provide genetic counselling.
- *Inform* about screening pregnant women for genetic disease and take appropriate action to prevent birth of children with malformations and genetic disorders.
- *Provide* skills in laboratory genetics to enable them to establish genetic tests using chromosomal studies, biochemical assays and molecular techniques.
- *Ensure* that the students acquire necessary knowledge and skills to plan and carry out research and perform statistical analysis of the data generated.
- *Acquaint* candidates with the principles of ethics as applied to genetic services and research.
- *Train* the candidates to think independently and become leaders in setting up genetic services and carry out research.

PROGRAMME OBJECTIVES

In this course main emphasis is laid on practical training in genetic counseling of diverse conditions like mental retardation, metabolic disorders, neurological disorders etc.; as well as basic and advanced genetic and genomic techniques through rotation in different institutions.

The core objective is to inspire in the students a curiosity that prompts them to explore the new world of genetics; to instil in them intellectual and technical tools for a career in clinical genetics; and to emphasize learning of genetics is a lifelong process.

The objectives of the course are summarized below divided into three domains:

- (I) Specific knowledge,
- (II) Specific skills, and
- (III) Attitudes

I. SPECIFIC KNOWLEDGE

At the end of the course the students should have a basic understanding of the following:

- 1.1) The Role of Genetics in Medicine
- 1.2) Biology of the cell
- 1.3) Structure, Function and Behaviour of Genes, and Chromosomes
- 1.4) Anatomy of the human genome – Gene mapping, linkage and positional Cloning
- 1.5) Genetic variation - its origin and detection
- 1.6) Family history and type of inheritance: Mendelian (autosomal dominant, autosomal recessive, x-linked)
- 1.7) Non-Traditional inheritance – mitochondrial, uniparental disomy, genomic imprinting
- 1.8) Multifactorial Inheritance and common diseases
- 1.9) Population genetics
- 1.10) Statistics as applied to Genetic epidemiology and Research

- 1.11) Consanguinity and its Consequences
- 1.12) Chromosomes and their disorders
- 1.13) Biochemical genetics and inborn errors of metabolism
- 1.14) Molecular genetics of human diseases - Hemoglobinopathies, Duchenne Muscular Dystrophy (DMD), Spinal Muscular Atrophy, Triplet repeat expansion etc.
- 1.15) Immunogenetics and HLA
- 1.16) Developmental genetics - Malformations and Teratology
- 1.17) Cancer Genetics
- 1.18) Clinical genetics: Evaluation of a dysmorphic child, Fragile X syndrome Cystic fibrosis Micro-deletion syndromes, Myotonic dystrophy, Muscular atrophies, Hereditary peripheral neuropathies, Diabetes mellitus, Stroke, Coronary artery disease, Hypertension, Huntington disease and Spinocerebellar ataxias, Neurofibromatosis, hemophilias, Autosomal and Sex chromosome abnormalities, Neural tube defects etc.
- 1.19) Genetic testing and Genetic screening
- 1.20) Genetic counselling & Prenatal diagnosis
- 1.21) Reproductive genetics, Pre-pregnancy and pregnancy counselling, screening in 1^s and 2nd trimester, obtaining and analysing fetal tissues for genetic disease, counselling
- 1.22) Therapy of genetic disorders
- 1.23) Human genome project and after, Genome wide association studies
- 1.24) Sequencing, Deep sequencing, Exome sequencing
- 1.25) Pharmacogenetics & Personalised medicine
- 1.26) Ethico-legal and social issues in Medical Genetics

II. SPECIFIC SKILLS

(a) Clinical

At the end of the course students should be able to:

- 1) *Elicit* a comprehensive family medical history, construct an appropriate pedigree and recognize patterns of inheritance
- 2) *Evaluate* cases of mental retardation, dysmorphology, disorders of sexual development, short stature and other common genetic problems
- 3) *Learn* to record anthropometric measurements used in clinical genetics

- 4) *Conduct* a general and systemic physical examination for proper evaluation of patients with genetic disorders
- 5) *Interview* pregnant women to identify those at risk for abnormalities in the fetus and learn their management
- 6) *Conduct* genetic counseling sessions
- 7) *Carry out*. Sweat chloride test, Muscle biopsy, Nerve biopsy, and Skin biopsy. And other common procedures required for practice of medical genetics
- 8) *Infer from* Dysmorphology databases

b) Laboratory techniques

1. Molecular

- ✓ DNA isolation and quantification
- ✓ Probe and primer designing
- ✓ PCR - standard and various modifications
- ✓ Real time PCR
- ✓ MLPA analysis
- ✓ Southern blotting – isotopic and nonisotopic methods
- ✓ Western blotting
- ✓ DNA Sequencing, including massively parallel sequencing
- ✓ Use of microarrays
- ✓ Bioinformatics as applied to sequencing and microarrays

2. Cytogenetics

- ✓ Karyotype analysis, blood , bone marrow, amniotic fluid, chorionic villus samples, products of conception
- ✓ Fluorescent in situ hybridization
- ✓ Cytogenetic studies using microarrays or beads-on-beads

3. Biochemical Genetics & HLA

- ✓ Enzyme analysis for storage disorders and neurodegenerative disorders
- ✓ Assays for various metabolites in blood and urine: e.g. succinylacetone, alpha 1 antitrypsin, ceruloplasmin

- ✓ Metabolic tests in urine, thin layer chromatography and High performance Liquid Chromatography for analysis of amino acids
- ✓ HLA typing by serology
- ✓ HLA typing by DNA PCR SSP
- ✓ Cross-matching for transplantation
- ✓ PRA antibodies, Donor specific antibodies

III. ATTITUDES

At the end of the course, the students should learn to:

- *Respect* patients religious moral and ethical beliefs and biases, even if they differ from the students own beliefs;
- *Present* all available options fairly, accurately and non-directively.
- *Appreciate* 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.
- *Apply* appropriate techniques for conveying difficult genetic information.
- *Recognize* the importance of imparting information to patients who are anxious or unfamiliar with the concepts being presented
- *Make* appropriate referral to genetics support groups community groups or other resources that can benefit the patients and their family.
- *Respect* the autonomy of all patients but also provide guidance with decision making when requested.
- *Cope* emotionally with patient responses.
- *Recognize* the limitations of their own skills and seek consultation when necessary

ELIGIBILITY CRITERIA FOR ADMISSIONS TO THE PROGRAMME

(A) DNB Medical Genetics Course:

1. Any medical graduate with DNB/MD/MS in Pediatrics, General Medicine or Obstetrics and Gynecology qualification, who has qualified the **Entrance Examination** conducted by NBE and fulfill the eligibility criteria for admission to DNB **Super Specialty** courses at various NBE accredited Medical Colleges/ institutions/Hospitals in India is eligible to participate in the Centralized counseling for allocation of DNB **Medical Genetics** seats purely on merit cum choice basis.
2. Admission to 3 years post MBBS DNB **Medical Genetics** course is only through **Entrance Examination** conducted by NBE and Centralized Merit Based Counseling conducted by National Board of Examination as per prescribed guidelines.

Duration of Course: 3 Years

Every candidate admitted to the training programme shall pursue a regular course of study (on whole time basis) in the concerned recognized institution under the guidance of recognized post graduate teacher for assigned period of the course.

TEACHING AND TRAINING ACTIVITIES

The fundamental components of the teaching programme should include:

1. Case presentations & discussion- once a week
2. Seminar – Once a week
3. Journal club- Once a week
4. Grand round presentation (by rotation departments and subspecialties)- once a week
5. Faculty lecture teaching- once a month
6. Clinical Audit-Once a Month

7. A poster and have one oral presentation at least once during their training period in a recognized conference.

The rounds should include bedside sessions, file rounds & documentation of case history and examination, progress notes, round discussions, investigations and management plan) interesting and difficult case unit discussions.

The training program would focus on knowledge, skills and attitudes (behavior), all essential components of education. It is being divided into theoretical, clinical and practical in all aspects of the delivery of the rehabilitative care, including methodology of research and teaching.

Theoretical: The theoretical knowledge would be imparted to the candidates through discussions, journal clubs, symposia and seminars. The students are exposed to recent advances through discussions in journal clubs. These are considered necessary in view of an inadequate exposure to the subject in the undergraduate curriculum.

Symposia: Trainees would be required to present a minimum of 20 topics based on the curriculum in a period of three years to the combined class of teachers and students. A free discussion would be encouraged in these symposia. The topics of the symposia would be given to the trainees with the dates for presentation.

Clinical: The trainee would be attached to a faculty member to be able to pick up methods of history taking, examination, prescription writing and management in rehabilitation practice.

Bedside: The trainee would work up cases, learn management of cases by discussion with faculty of the department.

Journal Clubs: This would be a weekly academic exercise. A list of suggested Journals is given towards the end of this document. The candidate would summarize and discuss the scientific article critically. A faculty member will suggest the article and moderate the discussion, with participation by other faculty members and resident doctors. The

contributions made by the article in furtherance of the scientific knowledge and limitations, if any, will be highlighted.

Research: The student would carry out the research project and write a thesis/ dissertation in accordance with NBE guidelines. He/ she would also be given exposure to partake in the research projects going on in the departments to learn their planning, methodology and execution so as to learn various aspects of research.

SYLLABUS

A. PRINCIPLES OF HUMAN GENETICS

1 The History and Impact of Genetics in Medicine

- Gregor Mendel and the Laws of Inheritance
- DNA as the Basis of Inheritance
- The Origins of Medical Genetics
- Types of Genetic Disorders (single gene disorders, Chromosomal disorders, Polygenic disorders, Somatic cell genetics, mitochondrial disorders)
- Major New Developments- The Human Genome Project,
- Internet resources for medical genetics

2 Basic Cell Biology: Structure and Function of genes

- DNA, RNA and Protein:
- DNA replication, Transcription, Translation,
- Regulation of gene expression
- Mutations and Mutagenesis- Types of mutations, Structural and Functional effects of mutation on the protein, Different mutagens

3 Chromosomes and Cell Division

- Human Chromosomes
- Cell Division- Mitosis, Meiosis, Gametogenesis
- Chromosome Abnormalities- Numerical, Structural and Mosaicism

4 Patterns of Inheritance

- Family Studies
- Mendelian Inheritance- Autosomal dominant, Autosomal recessive, Sex-linked recessive and dominant inheritance
- Multiple Alleles and Complex Traits
- Genetic heterogeneity
- Variable expressivity

- Genetic instability and Anticipation
- Mosaicism
- Uniparental Disomy
- Genomic Imprinting
- Mitochondrial Inheritance

5 Polygenic and Multifactorial Inheritance

- Polygenic Inheritance and the Normal Distribution
- Multifactorial Inheritance-The Liability/Threshold Model, Continuous and discontinuous traits
- Rules of polygenic inheritance
- Heritability
- Identifying Genes that Cause Multifactorial Disorders- Linkage analysis, Association studies, GWAS studies

B. MOLECULAR GENETICS

1 DNA Technology and Applications

- Structure of a gene
- DNA Cloning and The PCR
- Techniques of DNA Analysis- Nucleic acid probes, Nucleic acid hybridization assays
- DNA Sequencing – Sanger, and massively parallel
- Application of DNA sequence polymorphisms- SNPs, VNTRs, Minisatellites, Microsatellites

2 Mapping and Identifying Genes for Monogenic Disorders

- Position-Independent Identification of Human Disease Genes
- Positional Cloning
- The Human Genome Project and its Applications
- Epigenetics
- Microarray in research and clinical practice

C. CYTOGENETICS

- Methods of chromosome analysis- Karyotyping and chromosomal banding
- Fluorescent In-Situ Hybridization
- Comparative Genomic Hybridization
- Chromosome Nomenclature
- Prenatal cytogenetics
- Cancer cytogenetics

D. IMMUNOGENETICS

- Immunity
- Innate Immunity
- Specific Acquired Immunity
- Inherited Immunodeficiency Disorders
- Blood Groups
- HLA and Immunology of transplantation

E. POPULATION AND MATHEMATICAL GENETICS

- Hardy-Weinberg Principle and its Applications
- Factors that alter gene frequency- non-random mating, small populations, selection,
Mutations,, Migration and gene flow.
- Consanguinity and its consequences
- Genetic Polymorphism
- Segregation Analysis
- Genetic Linkage
- Risk Calculation
- Probability Theory
- The Use of Linked Markers
- Bayesâ Theorem and Prenatal Screening
- Empiric Risks

F. GENETICS IN MEDICINE

1 Hemoglobin and the Hemoglobinopathies

- Structure and Developmental Expression of Hemoglobin
- Synthesis and Control of Hemoglobin Expression
- Disorders of Hemoglobin – alpha , beta and gamma
- Clinical Variation of the Hemoglobinopathies
- Antenatal and Newborn Hemoglobinopathy Screening

2 Disorders of coagulation and bleeding

- Factor VIII
- Factor IX
- Afibrinogenemia
- Other disorders of coagulation
- Wiskott Aldrich syndrome

3 Biochemical Genetics

- Introduction of Inborn Errors of Metabolism
- Disorders of Amino Acid and Branched-Chain Amino Acid Metabolism
- Urea Cycle Disorders
- Disorders of Carbohydrate Metabolism
- Disorders of Steroid Metabolism
- Disorders of Lipid Metabolism,
- LDL receptor defects
- Lysosomal Storage Disorders
- Disorders of Purine/Pyrimidine Metabolism
- Disorders of Porphyrin Metabolism
- Organic-Acid Disorders
- Disorders of Copper Metabolism
- Peroxisomal Disorders
- Disorders Affecting Mitochondrial Function
- Prenatal Diagnosis of Inborn Errors of Metabolism

4 Pharmacogenetics

- Definition
- Drug Metabolism
- Genetic Variations Revealed by the Effects of Drugs
- Pharmacogenetics

5 Cancer Genetics

- Differentiation between Genetic and Environmental
- Factors in Cancer
- Oncogenes
- Tumor Suppressor Genes
- Epigenetics and Cancer
- Genetics of Common Cancers
- Genetic Counseling in Familial Cancer
- Tumor profiling in cancer and identifying targets for drug therapy

6 Genetic Factors in Common Diseases

- Genetic Susceptibility to Common Disease
- Approaches to Demonstrating Genetic Susceptibility to Common Diseases
- Disease Models for Multifactorial Inheritance
- Diabetes
- Crohn Disease
- Hypertension
- Coronary Artery Disease
- Schizophrenia
- Alzheimer Disease
- Hemochromatosis
- Venous Thrombosis
- Age-Related Macular Degeneration

G. CLINICAL GENETICS

1 Congenital Abnormalities and Dysmorphic Syndromes

- Incidence
- Definition and Classification of Birth Defects
- Genetic Causes of Malformations
- Environmental Agents (Teratogens)
- Malformations of Unknown Cause

2 Chromosome Disorders

- Incidence of Chromosome Abnormalities
- Disorders of the Sex Chromosomes
- Chromosome Deletion and Microdeletion Syndromes
- Disorders of Sexual Differentiation
- Chromosomal Breakage Syndromes
- Recurrent miscarriage
- Xeroderma Pigmentosa
- Chromosomal, FISH studies, Microarray-CGH Analysis
- X chromosome inactivation
- X-linked mental retardation and Fragile X

3 Single-Gene Disorders

- Huntington Disease
- Myotonic Dystrophy
- Hereditary Motor and Sensory Neuropathy
- Neurofibromatosis
- Marfan Syndrome
- Cystic Fibrosis
- Inherited Cardiac Arrhythmias and Cardiomyopathies
- Spinal Muscular Atrophy
- Duchenne Muscular Dystrophy
- Hemophilia
- Collagen disorders – Osteogenesis imperfecta and Ehlers Danlos syndrome

4 Screening for Genetic Disease

- Screening Those at High Risk
- Carrier Testing for Autosomal Recessive and X-Linked Disorders
- Presymptomatic Diagnosis of Autosomal Dominant Disorders
- Ethical Considerations in Carrier Detection and Predictive Testing
- Population Screening
- Neonatal Screening
- Population Carrier Screening
- Genetic Registers

5 Genetic Counseling

- Establishing the Diagnosis
- Calculating and Presenting the Risk
- Discussing the Options
- Communication and Support
- Outcomes in Genetic Counseling
- Ethical issues

6 Prenatal Testing and Reproductive Genetics

- Techniques Used in Prenatal Diagnosis
- Prenatal Screening
- Indications for Prenatal Diagnosis
- Special Problems in Prenatal Diagnosis
- Termination of Pregnancy
- Preimplantation Genetic Diagnosis
- Assisted Conception and Implications for Genetic Disease
- Non-Invasive Prenatal Diagnosis
- Prenatal Treatment

7 Treatment of Genetic Disease

- Conventional Approaches to Treatment of Genetic Disease
- Therapeutic Applications of Recombinant DNA Technology

- Gene transfer and Therapy
- RNA Modification
- Targeted Gene Correction
- Stem Cell Therapy

8 Ethical and Legal Issues in Medical Genetics

- General Principles
- Ethical Dilemmas in the Genetic Clinic
- Ethical Dilemmas and the Public Interest

9 Developmental Genetics

- Fertilization and Gastrulation
- Developmental Gene Families
- Role of Cilia in Developmental Abnormalities
- The Limb as a Developmental Model
- Developmental Genes and Cancer
- Positional Effects and Developmental Genes
- Hydatidiform Moles
- Sexual Differentiation and Determination
- Epigenetics and Development
- Twinning

8a. Semester Based Teaching

The 3 year course will be divided into six semesters.

Each semester will consist of clinical teaching by practice in a continuous way.

The postings for the various topics, including clinical and laboratory, are shown in the program.

There will be a joint session once a month, through teleconferencing, where student and faculty from all the three centers will make presentations, One hour will be allotted to each centre.

Once a month all students will participate in the tele conference on medical genetics run from the Sanjay Gandhi Institute of Postgraduate Medical Education and Research.

In each semester there will be a one week course on different advanced topics in genetics, organized in the three centres or other genetic centres in the country, for the candidates.

After each module of posting, the candidates will have to assessment as outlined below.

There will be a 2 weeks holiday in each semester.

Details of Semester Teaching:

Three years curriculum

Semester	Activity	Assessment
Semester 1 1st th May – 31 th October 2015	Clinical Genetics - 24weeks 20 th July – 1 st Aug 2015 National Course – 1, Clinical Genetics at SGPGI, Lucknow – 2 weeks	End Assessment by MCQs
Semester 2 1 st November 2015 – 30 th April 2016	Metabolic genetics, includes lysosomal disorders & newborn screening – 12 wk	

	Prenatal genetics – 4 weeks	
	Cancer genetics – 4 weeks	
	✓ Clinical Cytogenetics – 4 weeks	
	National Course – 2, Cytogenetics at SGRH, Delhi – 2 weeks	
Semesters 1 & 2	<p>Once every two months inter-institutional educational program , Guest lectures</p> <p>From 3- 5PM, Second Tuesday</p> <p>Monthly half day Educational Course organised by SGPGI, Lucknow, Second Wednesday of each month from 3.30-5 PM</p>	End assessment by MCQs
Semester 3 1 st May 2016 – 31 st October 2016 Laboratory Genetics	Introduction to Laboratory Medicine in Genetics and its Clinical Application	
	Molecular genetics – 6 weeks	
	Cytogenetics – 6 weeks	
	Biochemical genetics – 6 weeks	
	Immuno-hematology + HLA – 4 wks,	
	Prenatal procedures - 2 weeks	Practical Assessment
	National Course 3 Molecular Laboratory Genetics at CDFD, Hyderabad – 2 weeks	
Semester 4 1 st Nov – 30 th April 2017	Clinical Genetics – 12 weeks	Log book
	Research Elective – 12 weeks Development clinic, pediatric neurology	Assessment of work done
	National Course 4, Population Genetics and Epidemiology as applied to Genetics at NIBMG,	End Assessment by MCQs

	Kolkata– 2 wk	
Semesters 3 and 4	Once every two months inter-institutional educational program , Guest lectures From 3- 5PM, Second Tuesday Monthly half day Educational Course organised by SGPGI, Lucknow, Second Wednesday of each month from 3.30-5 PM	End assessment by MCQs
Semester 5 1 st May 2017 – 31 st Oct 2017	Research – 14 weeks	Assessment of work done
	Clinical Genetics – 10 weeks	Log book
	National Course 5, Clinical And Laboratory genetics - at CDFD, Hyderabad – 2 weeks	End Assessment by MCQs

<p>Semester 6 1st Nov 2017 – 31st April 18</p>	<p>Research – 14 wk, Thesis writing – 6 wk</p>	
	<p>National Course 6, Public health genetics & Leadership training – PHFI, Delhi – 2 wk</p>	

	<p align="center">Monthly Half day inter-institutional educational program , Guest lectures Monthly half day Educational Course organised by SGPGI, Lucknow</p>	
	<p align="center">National Research Retreat – Trainees present their research</p>	<p align="center">Final Written, Clinical & Oral examination</p>

General outline for the 1st Semester

General outline that will be followed by the DNB Medical Genetics students during the first semester- **Clinical Genetics (26 weeks)** will be as follows:

- Attend Genetic clinic daily
- Two formal case presentations every week

- Faculty lecture once every week
- Student seminar once every week
- Journal club will be presented by both students and faculty once every week

In addition to the above, emphasis will be made on development of specific clinical skills:

- 9) *Elicit* a comprehensive family medical history, construct an appropriate pedigree and recognize patterns of inheritance
- 10) *Evaluate* cases of mental retardation, dysmorphology, disorders of sexual development, short stature and other common genetic problems
- 11) *Learn* to record anthropometric measurements used in clinical genetics
- 12) *Conduct* a general and systemic physical examination for proper evaluation of patients with genetic disorders. Ex: Hearing evaluation
- 13) *Interview* pregnant women to identify those at risk for abnormalities in the fetus and learn their management
- 14) *Conduct* genetic counseling sessions
- 15) *Carry out*: Sweat chloride test, Muscle biopsy, Nerve biopsy, and Skin biopsy.
- 16) *Carry out*: Fetal autopsies.
- 17) *Infer from* Dysmorphology databases
- 18) *Attend postings* for 1-2 weeks in the Neurology department, Development clinic.

2. Details of lectures by faculty and seminars by students

List of Faculty Lectures

1. The role of Genetics in Medicine
2. Approach to history taking and examination in a patient with a genetic disorder
3. Interpreting Family history and Mendelian Inheritance patterns
4. Non- Mendelian patterns of inheritance
5. Multifactorial inheritance and common diseases

6. Genetic Counselling
7. Chromosomes- Structure, Classification and Identification
8. Structure of Genes, Types of mutation with examples
9. Approach to a child with dysmorphism
10. Common Microdeletion syndromes
11. Prenatal screening for genetic disorders
12. Microarray analysis- Basics and indications in clinical practice
13. Approach to diagnosis of IEM's
14. Genetic causes of short stature
15. Recurrent pregnancy loss
16. Genetic causes of Autism
17. Ophthalmological clues to genetic disorders
18. Approach to a child with macrosomia
19. Introduction to Hemoglobinopathy
20. Approach to diagnosis of Skeletal dysplasia
21. Principles of USG and evaluation of fetal growth
22. Duchene muscular dystrophy

3.List of Student Seminars

1. Common Chromosomal disorders
2. Basic DNA techniques
3. Approach to hepatosplenomegaly
4. Approach to patients with intellectual disability
5. Neuromuscular disorders
6. Approach to a child with Neuroregression
7. Acute encephalopathy – genetic causes and approach to diagnosis and management
8. Inherited Cardiomyopathies
9. Disorders with deafness
10. Thalassemia
11. Coagulation disorders

12. Mitochondrial disorders
13. Intrauterine infections
14. Fragile X- syndrome
15. Child with hypotonia

Biostatistics, Research Methodology and Clinical Epidemiology

Ethics

Medico legal aspects relevant to the discipline

Health Policy issues as may be applicable to the discipline

THESIS PROTOCOL & THESIS

The candidates are required to submit a thesis at the end of three years of training as per the rules and regulations of NBE.

Guidelines for Submission of Thesis Protocol & Thesis by candidates

Research shall form an integral part of the education programme of all candidates registered for DNB degrees of NBE. The Basic aim of requiring the candidates to write a thesis protocol & thesis/dissertation is to familiarize him/her with research methodology. The members of the faculty guiding the thesis/dissertation work for the candidate shall ensure that the subject matter selected for the thesis/dissertation is **feasible, economical** and **original**.

Guidelines for Thesis Protocol

The protocol for a research proposal (including thesis) is a study plan, designed to describe the background, research question, aim and objectives, and detailed methodology of the study. In other words, the protocol is the 'operating manual' to refer to while conducting a particular study.

The candidate should refer to the NBE Guidelines for preparation and submission of Thesis Protocol before the writing phase commences. The

minimum writing requirements are that the language should be clear, concise, precise and consistent without excessive adjectives or adverbs and long sentences. There should not be any redundancy in the presentation.

The development or preparation of the Thesis Protocol by the candidate will help her/him in understanding the ongoing activities in the proposed area of research. Further it helps in creating practical exposure to research and hence it bridges the connectivity between clinical practice and biomedical research. Such research exposure will be helpful in improving problem solving capacity, getting updated with ongoing research and implementing these findings in clinical practice.

Research Ethics: Ethical conduct during the conduct and publication of research is an essential requirement for all candidates and guides, with the primary responsibility of ensuring such conduct being on the thesis guide. Issues like Plagiarism, not maintaining the confidentiality of data, or any other distortion of the research process will be viewed seriously. The readers may refer to standard documents for the purpose.

The NBE reserves the right to check the submitted protocol for plagiarism, and will reject those having substantial duplication with published literature.

PROTOCOL REQUIREMENTS

1. All of the following will have to be entered in the online template. The thesis protocol should be restricted to the following word limits.
 - Title : 120 characters (with spacing) page
 - Synopsis [structured] : 250-300
 - Introduction : 300-500
 - Review of literature : 800-1000
 - Aim and Objectives : Up to 200
 - Material and Methods : 1200-1600
 - 10-25 References [ICMJE style]
2. It is mandatory to have ethics committee approval before initiation of the research work. The researcher should submit an appropriate application to the ethics committee in the prescribed format of the ethics committee concerned.

Guidelines for Thesis

1. The proposed study must be approved by the institutional ethics committee and the protocol of thesis should have been approved by NBE.
2. The thesis should be restricted to the size of 80 pages (maximum). This includes the text, figures, references, annexures, and certificates etc. It should be printed on both sides of the paper; and every page has to be numbered. Do not leave any page blank. To achieve this, following points may be kept in view:
 - a. The thesis should be typed in 1.5 space using Times New Roman/Arial/ Garamond size 12 font, 1" margins should be left on all four sides. Major sections viz., Introduction, Review of Literature, Aim & Objectives, Material and Methods, Results, Discussion, References, and Appendices should start from a new page. Study proforma (Case record form), informed consent form, and patient information sheet may be printed in single space.
 - b. Only contemporary and relevant literature may be reviewed. Restrict the introduction to 2 pages, Review of literature to 10-12 pages, and Discussion to 8-10 pages.
 - c. The techniques may not be described in detail unless any modification/innovations of the standard techniques are used and reference(s) may be given.
 - d. Illustrative material may be restricted. It should be printed on paper only. There is no need to paste photographs separately.
3. Since most of the difficulties faced by the residents relate to the work in clinical subject or clinically-oriented laboratory subjects, the following steps are suggested:
 - a. The number of cases should be such that adequate material, judged from the hospital attendance/records, will be available and the candidate will be able to collect case material within the period of data collection, i.e., around 6-12 months so that he/she is in a position to complete the work within the stipulated time.
 - b. The aim and objectives of the study should be well defined.
 - c. As far as possible, only clinical/laboratory data of investigations of patients or such other material easily accessible in the existing facilities should be used for the study.
 - d. Technical assistance, wherever necessary, may be provided by the department concerned. The resident of one specialty taking up some problem related to some other specialty should have some basic knowledge about the subject and he/she should be able to perform the investigations independently, wherever some specialized laboratory investigations are required a co-guide may

be co-opted from the concerned investigative department, the quantum of laboratory work to be carried out by the candidate should be decided by the guide & co-guide by mutual consultation.

4. The clinical residents are not ordinarily expected to undertake experimental work or clinical work involving new techniques, not hitherto perfected OR the use of chemicals or radioisotopes not readily available. They should; however, be free to enlarge the scope of their studies or undertake experimental work on their own initiative but all such studies should be feasible within the existing facilities.
5. The DNB residents should be able to freely use the surgical pathology/autopsy data if it is restricted to diagnosis only, if however, detailed historic data are required the resident will have to study the cases himself with the help of the guide/co-guide. The same will apply in case of clinical data.
6. Statistical methods used for analysis should be described specifically for each objective, and name of the statistical program used mentioned.

General Layout of a DNB Thesis:

- **Title-** A good title should be brief, clear, and focus on the central theme of the topic; it should avoid abbreviations. The Title should effectively summarize the proposed research and should contain the PICO elements.
- **Introduction-** It should be focused on the research question and should be directly relevant to the objectives of your study.
- **Review of Literature** - The Review should include a description of the most relevant and recent studies published on the subject.
- **Aim and Objectives** - The 'Aim' refers to what would be broadly achieved by this study or how this study would address a bigger question / issue. The 'Objectives' of the research stem from the research question formulated and should at least include participants, intervention, evaluation, design.
- **Material and Methods-** This section should include the following 10 elements: Study setting (area), Study duration; Study design (descriptive, case-control, cohort, diagnostic accuracy, experimental (randomized/non-randomized)); Study sample (inclusion/exclusion criteria, method of

selection), Intervention, if any, Data collection, Outcome measures (primary and secondary), Sample size, Data management and Statistical analysis, and Ethical issues (Ethical clearance, Informed consent, trial registration).

- **Results-** Results should be organized in readily identifiable sections having correct analysis of data and presented in appropriate charts, tables, graphs and diagram etc.
- **Discussion**—It should start by summarizing the results for primary and secondary objectives in text form (without giving data). This should be followed by a comparison of your results on the outcome variables (both primary and secondary) with those of earlier research studies.
- **Summary and Conclusion-** This should be a précis of the findings of the thesis, arranged in four paragraphs: (a) background and objectives; (b) methods; (c) results; and (d) conclusions. The conclusions should strictly pertain to the findings of the thesis and not outside its domain.
- **References-** Relevant References should be cited in the text of the protocol (in superscripts).
- **Appendices** -The tools used for data collection such as questionnaire, interview schedules, observation checklists, informed consent form (ICF), and participant information sheet (PIS) should be attached as appendices. Do not attach the master chart.

Thesis Protocol Submission to NBE

1. DNB candidates are required to submit their thesis protocol within 90 days of their joining DNB training.
2. Enclosures to be submitted along with protocol submission form:
 - a) Form for Thesis Protocol Submission properly filled.
 - b) Thesis Protocol duly signed.
 - c) Approval letter of institutional Ethical committee. (*Mandatory, non receivable of any one is liable for rejection*)

Thesis Submission to NBE

1. As per NBE norms, writing a thesis is essential for all DNB candidates towards partial fulfillment of eligibility for award of DNB degree.

2. DNB candidates are required to submit the thesis before the cut-off date which shall be 30th June of the same year for candidates appearing for their scheduled December final theory examination. Similarly, candidates who are appearing in their scheduled June DNB final examination shall be required to submit their thesis by 31st December of preceding year.
3. Candidates who fail to submit their thesis by the prescribed cutoff date shall NOT be allowed to appear in DNB final examination.
4. Fee to be submitted for assessment (In INR): 3500/-
5. Fee can be deposited ONLY through pay-in-slip/challan at any of the Indian bank branch across India. The challan can be downloaded from NBE website www.natboard.edu.in
6. Thesis should be bound and the front cover page should be printed in the standard format. A bound thesis should be accompanied with:
 - a. A Synopsis of thesis.
 - b. Form for submission of thesis, duly completed
 - c. NBE copy of challan (in original) towards payment of fee as may be applicable.
 - d. Soft copy of thesis in a CD duly labeled.
 - e. Copy of letter of registration with NBE.
7. A declaration of thesis work being bonafide in nature and done by the candidate himself/herself at the institute of DNB training need to be submitted bound with thesis. It must be signed by the candidate himself/herself, the thesis guide and head of the institution, failing which thesis shall not be considered.

The detailed guidelines and forms for submission of Thesis

Protocol & Thesis are available at

www.natboard.edu.in.thesis.php

LOG BOOK

A candidate shall maintain a log book of operations (assisted / performed) during the training period, certified by the concerned post graduate teacher / Head of the department / senior consultant.

This log book shall be made available to the board of examiners for their perusal at the time of the final examination.

The log book should show evidence that the before mentioned subjects were covered (with dates and the name of teacher(s)) The candidate will maintain the record of all academic activities undertaken by him/her in log book .

1. Personal profile of the candidate
2. Educational qualification/Professional data
3. Record of case histories
4. Procedures learnt
5. Record of case Demonstration/Presentations
6. Every candidate, at the time of practical examination, will be required to produce performance record (log book) containing details of the work done by him/her during the entire period of training as per requirements of the log book. It should be duly certified by the supervisor as work done by the candidate and countersigned by the administrative Head of the Institution.
7. In the absence of production of log book, the result will not be declared.

Leave Rules

1. DNB Trainees are entitled to leave during the course of DNB training as per the Leave Rules prescribed by NBE.
2. A DNB candidate can avail a maximum of 20 days of leave in a year excluding regular duty off/ Gazetted holidays as per hospital/institute calendar/policy.
3. MATERNITYLEAVE:
 - a. A female candidate is permitted a maternity leave of 90 days once during the entire duration of DNB course.
 - b. The expected date of delivery (EDD) should fall within the duration of maternity leave.
 - c. Extension of maternity leave is permissible only for genuine medical reasons and after prior approval of NBE. The supporting medical documents have to be certified by the Head of the Institute/hospital where the candidate is undergoing DNB training. NBE reserves its rights to take a final decision in such matters.
 - d. The training of the candidate shall be extended accordingly in case of any extension of maternity leave being granted to the candidate.
 - e. Candidate shall be paid stipend during the period of maternity leave. No stipend shall be paid for the period of extension of leave.
4. Male DNB candidates are entitled for paternity leave of maximum of one week during the entire period of DNB training.
5. No kind of study leave is permissible to DNB candidates. However, candidates may be allowed an academic leave as under across the entire duration of training program to attend the conferences/CMEs/Academic programs/Examination purposes.

DNB COURSE	NO. OF ACADEMIC LEAVE
DNB 3 years Course (Broad & Super Specialty)	14 Days
DNB 2 years Course (Post Diploma)	10 Days
DNB Direct 6 years Course	28 days

6. Under normal circumstances leave of one year should not be carried forward to the next year. However, in exceptional cases such as prolonged illness the leave across the DNB training program may be clubbed together with prior approval of NBE.
7. Any other leave which is beyond the above stated leave is not permissible and shall lead to extension/cancellation of DNB course.
8. Any extension of DNB training for more than 2 months beyond the scheduled completion date of training is permissible only under extraordinary circumstances with prior approval of NBE. Such extension is neither automatic nor shall be granted as a matter of routine. NBE shall consider such requests on merit provided the seat is not carried over and compromise with training of existing trainees in the Department.
9. Unauthorized absence from DNB training for more than 7 days may lead to cancellation of registration and discontinuation of the DNB training and rejoining shall not be permitted.

10. Medical Leave

- a. Leave on medical grounds is permissible only for genuine medical reasons and NBE should be informed by the concerned institute/hospital about the same immediately after the candidate proceeds on leave on medical grounds.
- b. The supporting medical documents have to be certified by the Head of the Institute/hospital where the candidate is undergoing DNB training and have to be sent to NBE.
- c. The medical treatment should be taken from the institute/ hospital where the candidate is undergoing DNB training. Any deviation from this shall be supported with valid grounds and documentation.
- d. In case of medical treatment being sought from some other institute/hospital, the medical documents have to be certified by the Head of the institute/hospital where the candidate is undergoing DNB training.

- e. NBE reserves its rights to verify the authenticity of the documents furnished by the candidate and the institute/hospital regarding Medical illness of the candidate and to take a final decision in such matters.

11.

- a. Total leave period which can be availed by DNB candidates is $120+28 = 148$ days for 6 years course, $60+14=74$ days for 3 years course and $40+10 = 50$ days for 2 years course. This includes all kinds of eligible leave including academic leave. Maternity / Paternity leave can be availed separately by eligible candidates. Any kind of leave including medical leave exceeding the aforementioned limit shall lead to extension of DNB training. It is clarified that prior approval of NBE is necessary for availing any such leave.
- b. The eligibility for DNB Final Examination shall be determined strictly in accordance with the criteria prescribed in the respective information bulletin.

EXAMINATION

FORMATIVE ASSESSMENT

Formative assessment includes various formal and informal assessment procedures by which evaluation of student's learning, comprehension, and academic progress is done by the teachers/ faculty to improve student attainment. Formative assessment test (FAT) is called as "Formative" as it informs the in process teaching and learning modifications. FAT is an integral part of the effective teaching. The goal of the FAT is to collect information which can be used to improve the student learning process.

Formative assessment is essentially positive in intent, directed towards promoting learning; it is therefore part of teaching. Validity and usefulness are paramount in formative assessment and should take precedence over concerns for reliability. The assessment scheme consists of Three Parts which has to be essentially completed by the candidates.

The scheme includes:-

Part I:- Conduction of theory examination

Part-II :- Feedback session on the theory performance

Part-III :- Work place based clinical assessment

Scheme of Formative assessment

PART – I	CONDUCT OF THEORY EXAMINATION	Candidate has to appear for Theory Exam and it will be held for One day.
PART – II	FEEDBACK SESSION ON THE THEORY PERFORMANCE	Candidate has to appear for his/her Theory Exam Assessment Workshop.
PART – III	WORK PLACE BASED CLINICAL ASSESSMENT	After Theory Examination, Candidate has to appear for Clinical Assessment.

The performance of the resident during the training period should be monitored throughout the course and duly recorded in the log books as evidence of the ability and daily work of the student

1. Personal attributes:

- **Behavior and Emotional Stability:** Dependable, disciplined, dedicated, stable in emergency situations, shows positive approach.
- **Motivation and Initiative:** Takes on responsibility, innovative, enterprising, does not shirk duties or leave any work pending.

- **Honesty and Integrity:** Truthful, admits mistakes, does not cook up information, has ethical conduct, exhibits good moral values, loyal to the institution.
- **Interpersonal Skills and Leadership Quality:** Has compassionate attitude towards patients and attendants, gets on well with colleagues and paramedical staff, is respectful to seniors, has good communication skills.

2. Clinical Work:

- **Availability:** Punctual, available continuously on duty, responds promptly on calls and takes proper permission for leave.
- **Diligence:** Dedicated, hardworking, does not shirk duties, leaves no work pending, does not sit idle, competent in clinical case work up and management.
- **Academic ability:** Intelligent, shows sound knowledge and skills, participates adequately in academic activities, and performs well in oral presentation and departmental tests.
- **Clinical Performance:** Proficient in clinical presentations and case discussion during rounds and OPD work up. Preparing Documents of the case history/examination and progress notes in the file (daily notes, round discussion, investigations and management) Skill of performing bed side procedures and handling emergencies.

3. Academic Activity: Performance during presentation at Journal club/ Seminar/ Case discussion/Stat meeting and other academic sessions. Proficiency in skills as mentioned in job responsibilities.

FINAL EXAMINATION

The summative assessment of competence will be done in the form of DNB Final Examination leading to the award of the degree of Diplomate of National Board in Emergency Medicine. The DNB final is a two-stage examination comprising the theory and practical part. An eligible candidate who has qualified the theory exam is permitted to appear in the practical examination.

Theory Examination

1. The theory examination comprises of **Three / Four** papers, maximum marks 100 each.
2. There are 10 short notes of 10 marks each, in each of the papers. The number of short notes and their respective marks weightage may vary in some subjects/some papers.
3. Maximum time permitted is 3 hours.
4. Candidate must score at least 50% in the aggregate of **Three/ Four** papers to qualify the theory examination.

5. Candidates who have qualified the theory examination are permitted to take up the practical examination.

a) Practical Examination:

1. Maximum Marks: 300.
2. Comprises of Clinical Examination and Viva.
3. Candidate must obtain a minimum of 50% marks in the Clinical Examination (including Viva) to qualify for the Practical Examination.
4. There are a maximum of three attempts that can be availed by a candidate for Practical Examination.
5. First attempt is the practical examination following immediately after the declaration of theory results.
6. Second and Third attempt in practical examination shall be permitted out of the next three sessions of practical examinations placed alongwith the next three successive theory examination sessions; after payment of full examination fees as may be prescribed by NBE.
7. Absentation from Practical Examination is counted as an attempt.
8. Appearance in first practical examination is compulsory;
9. Requests for Change in center of examination are not entertained, as the same is not permissible.
10. Candidates are required not to canvass with NBE for above.

Declaration of DNB Final Results

1. DNB final is a qualifying examination.
2. Results of DNB final examinations (theory & practical) are declared as PASS/FAIL.
3. DNB degree is awarded to a DNB trainee in the convocation of NBE.

RECOMMENDED TEXT BOOKS AND JOURNALS

Essential Journals:

- American Journal of Human Genetics
- American Journal of Medical Genetics
- Clinical Chemistry
- Clinical Genetics
- European Journal of Human Genetics
- European Journal of Medical Genetics
- Genes Chromosomes and Cancer
- Genetic testing and Molecular Biomarkers
- Genetics in Medicine
- Human Genetics
- Human Molecular Genetics
- Human Mutation
- J Molecular Diagnosis
- Journal of American Medical Association
- Journal of Fetal Medicine
- Journal of Inherited Metabolic disease
- Journal of Medical Genetics
- Lancet
- Molecular Genetics and Metabolism
- Nature Genetics
- Nature Reviews Genetics
- New England Journal of Medicine
- Orphanet Journal of Rare Diseases
- Prenatal Diagnosis
- Science & Translational medicine
- Trends in Genetics

Essential Texts:

- Alberts B, Johnson A, Lewis J, Morgan D, Roberts K, Walter P. Molecular Biology of the Cell. 6th ed. 2014. Garland.
- Blau N, Duran M, Gibson KM (Eds). Laboratory Guide to the Methods in Biochemical Genetics. 2008. Springer Online (accessed November 2012). ISBN 9783540766988
- Harper P: Practical Genetic Counselling, (7th edition 2010). Oxford University Press
- Jones KL and Smith DW : Smith's Recognizable patterns of Human malformations (7th Edition, 2013): Elsevier Health Sciences
- Jorde LB, Carey JC and Bamshed MJ: Medical Genetics (4th Edition, 2010): Elsevier Health Sciences.
- Krebs JE, Goldstein ES, Kilpatrick ST. Lewin's Genes 11 ed. Jones and Bartlett. 2012.

- Kumar D, Eng C. Genomic Medicine, Principles and Practice. 2nd ed. Oxford University Press. 201
- Milunsky A and Milunsky JM: Genetic disorders and the fetus. 6th edition. Wiley and Blackwell. 2010.
- Nussbaum RL, McInnes RR, Willard HF: Thompson & Thompson Genetics in Medicine (7th Edition, 2007) Elsevier Health Sciences.
- Rimoin DL, Connor JM, Pyeritz RE, Korf BR : Emery and Rimoin's Principles of Medical Genetics (5th Edition, 2006) Elsevier Health Sciences.
- Saudubray J-M, Berghe G vd, Walter JH (Eds). Inborn Metabolic Diseases. Diagnosis and Treatment. 5th Edition 2012 Springer Online (accessed November 2012). ISBN 9783642157202

- Saudubray, Jean-Marie, Berghe, Georges van den, Walter, John H. (Eds.) Inborn Metabolic Diseases. Diagnosis and Treatment 5th ed. 2012, XXVIII, 660 p.
- Schaefer GB, Thompson Jr JN Medical Genetics – an integrated approach. McGraw Hill. (2014)
- Scriver, C.R., Beaudet, A.L., Valle, D., Sly, W.S., Vogelstein, B., Childs, B., Kinzler, K.W. Scriver's OMMBID The Online Metabolic & Molecular Bases of Inherited Disease
- Strachan T and Read AP: Human Molecular Genetics (4rd edition,2010) Garland Science/Taylor & Francis Group. Tobias ES, Connor M, and FergusonSmith M., Essential Medical Genetics Edition ; 6th ed ; Oxford, Cambridge, Mass. : Blackwell Science, 2011
- Turnpenny PD and Ellard S. editors, Emery's Element of Medical Genetics, (14 ed. 2011)
- Vogel and Motulsky's Human Genetics (4th Edition, 2010) Springer.
- Young I: Introduction to Risk Calculation in Genetic Counseling (3rd Edition,2006) Oxford University Press.