Curriculum

DrNB Super Specialty

Medical Genetics

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I. 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 thal 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.

II. PROGRAMME GOAL & OBJECTIVES

1. 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 n 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.

2. 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 instill 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:

- The Role of Genetics in Medicine
- Biology of the cell
- Structure, Function and Behaviour of Genes, and Chromosomes
- Anatomy of the human genome – Gene mapping, linkage and positional Cloning
- Genetic variation - its origin and detection
- Family history and type of inheritance: Mendelian (autosomal dominant, autosomal recessive, x-linked)
- Non-Traditional inheritance – mitochondrial, uniparental disomy, genomic imprinting
- Multifactorial Inheritance and common diseases
- Population genetics
- Statistics as applied to Genetic epidemiology and Research
- Consanguinity and its Consequences
- Chromosomes and their disorders
- Biochemical genetics and inborn errors of metabolism

- Molecular genetics of human diseases - Hemoglobinopathies, Duchenne Muscular Dystrophy (DMD), Spinal Muscular Atrophy, Triplet repeat expansion etc.
- Immunogenetics and HLA
- Developmental genetics - Malformations and Teratology
- Cancer Genetics
- 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.
- Genetic testing and Genetic screening
- Genetic counselling & Prenatal diagnosis
- Reproductive genetics, Pre-pregnancy and pregnancy counselling,
- screening in 1s and 2nd trimester, obtaining and analysing fetal tissues for genetic disease, counselling
- Therapy of genetic disorders
- Human genome project and after, Genome wide association studies
- Sequencing, Deep sequencing, Exome sequencing
- Pharmacogenetics&Personalised medicine
- 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 non-isotopic 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, productsiof conception
- Fluorescent in situ hybridization
- Cytogenetic studies using microarrays or bacs-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

4. 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

III. 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. Thecontributions 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.

IV. 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, Mini satellites, 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
   - XerodermaPigmentosa
   - 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 imperfects 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

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 may – 31st October 2015	Clinical Genetics – 24 weeks	End Assessment by MCQs
	20the July – 1st Aug 2015 National Gourse – 1, Clinical Genetics at SGPGI, Lucknow – 2 weeks	
Semester 2 1st November 2015 – 30th April 2016	Metabolic genetics, includes lysosomal disorders &newborn screening – 12 wk	End assessment by MCQs
	Prenatal genetics – 4 weeks	
	Cancer genetics – 4 weeks	
	√ Clinical Cytogenetics – 4 weeks	
	National Course – 2, Cytogenetics at SGRH, Delhi – 2 weeks	

Semesters 1 & 2	Once every two months inter-institutional educational programm, 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	
	Introduction to Laboratory Medicine in Genetics and it Clinical Application	
Semester 3 1st May 2016-31st October 2016 Laboratory Genetics	Molecular genetics – 6 weeks	Practical Assessment
	Cytogenetics – 6 weeks	
	Biochemical genetics – 6 weeks	
	Immuno-hematology + HLA – 4 weeks	
	Prenatal procedures – 2 weeks	
	National course 3 Molecular Laboratory Genetics at CDFD, Hyderabad – 2 weeks	
Semester 4 1st Nov. – 30th 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 Eopidemiology as applied to Genetics at NIBMG, Kolkata – 2 weeks	End assessment by MCQs
Semesters 3 and 4	Once every two months inter-institutional educational programme, Guest lectures From 3 – 5 PM, Second Tuesday Monthly half day Educational Course organised by SGPGI, Lucknow, Second Wednesday of each month	End assessment by MCQs

	from 3.30 – 5 PM	
Semester 5 1st May 2017 – 31st Oct. 2017	Research – 14 weeks	Assessment of work done
	Clinical Genetics – 10 weeks	Log Books
Semester 6 1st Nov. 2017 – 31st April 18	National Course 5, Clinical And Laboratory genetics – at CDFD, Hyderabad – 2 weeks	End Assessment by MCQs
	Research – 14 weeks, Thesis Writing – 6 weeks	
	National Course 6, Public Health Genetics & Leadership training- PHFI, Delhi – 2 week	
	Monthly Half day inter-institutional educational programm, Guest lectures Monthly half day Educational Course organised by SGPGI, Lucknow	
	National Research Retreat – Trainees present tgheir research	Final written, clinical & Oral Examination

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:

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

- Conduct a general and systemic physical examination for proper evaluation of patients with genetic disorders. Ex: Hearing evaluation
- Interview pregnant women to identify those at risk for abnormalities in the fetus and learn their management
- Conduct genetic counseling sessions
- Carry out: Sweat chloride test, Muscle biopsy, Nerve biopsy, and Skin biopsy.
- Carry out: Fetal autopsies.
- Infer from Dysmorphology databases
- Attend postings for 1-2 weeks in the Neurology department, Development clinic.

Details of lectures by faculty and seminars by students List of Faculty Lectures

- The role of Genetics in Medicine
- Approach to history taking and examination in a patient with a genetic disorder
- Interpreting Family history and Mendelian Inheritance patterns
- Non- Mendelian patterns of inheritance
- Multifactorial inheritance and common diseases
- Genetic Counselling
- Chromosomes- Structure, Classification and Identification
- Structure of Genes, Types of mutation with examples
- Approach to a child with dysmorphism
- Common Microdeletion syndromes
- Prenatal screening for genetic disorders
- Microarray analysis- Basics and indications in clinical practice
- Approach to diagnosis of IEM's
- Genetic causes of short stature
- Recurrent pregnancy loss
- Genetic causes of Autism
- Opthalmological clues to genetic disorders
- Approach to a child with macrosomia
- Intoduction to Hemoglobinopathy

- Approach to diagnosis of Skeletal dysplasia
- Principles of USG and evaluation of fetal growth
- Duchene muscular dystrophy

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

V. 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.

VI. 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.

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- Strachan T and Read AP: Human Molecular Genetics (4th edition,2010)
- Garland Science/ Taylor & Francis Group. Tobias ES
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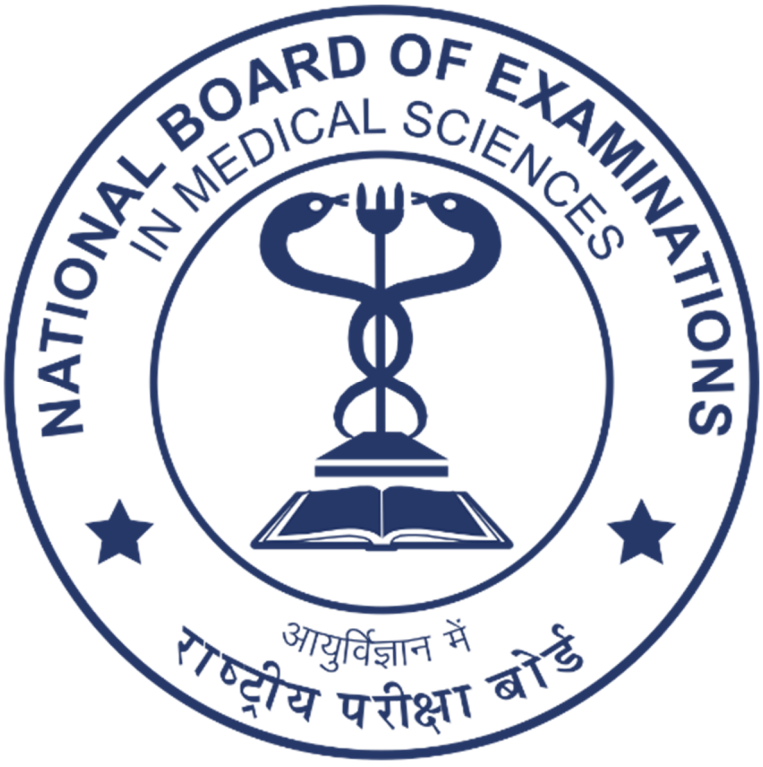
- Turnpenny PD and Ellard S. editors, Emery's Element of Medical Genetics, (14 ed. 2011)
- Vogel and Motulsky's Human Genetics (4th Edition, 2010) Springer.
- Young I: Introduction to Risk Calculation in Genetic Counseling (3rd Edition,2006) Oxford University Press.



आयुर्विज्ञान में राष्ट्रीय परीक्षा बोर्ड

स्वास्थ्य एवं परिवार कल्याण मंत्रालय, भारत सरकार

मेडिकल एन्क्लेव, अंसारी नगर, नई दिल्ली – 110029

NATIONAL BOARD OF EXAMINATIONS IN MEDICAL SCIENCES

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