ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 11 | November - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

TRAINING OF IN-SERVICE CLINICIANS' FROM GOVERNMENT HOSPITALS AND OUTREACH PROGRAMME 'UMMID'

Medical Education

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ABSTRACT

Rural and tribal areas in a country like India have a high prevalence of inherited disorders due to various factors including inadequate medical training for clinicians in basic clinical and diagnostic genetics. UMMID (Unique Methods of Management and treatment of Inherited Disorders) and NIDAN (National Inherited Diseases Administration), supported by the Department of Biotechnology (DBT) , aimed to provide six-month clinical and diagnostic genetics training to in-service clinicians to identify genetic disorders in vulnerable populations of aspirational districts. This initiative highlights the pressing need for additional genetics training programs for clinicians to effectively manage and prevent these disorders at the community level.

KEYWORDS

Training Clinician 'UMMID'

BACKGROUND AND AIM

Genetic disorders are a major cause of illness and death worldwide, particularly in developing countries [1,2] .While there is currently a focus on training medical professionals and providing care at the college level, there's an urgent need to consider community-level programs for managing and preventing genetic disorders, especially in rural and tribal areas of countries like India. These areas may have a higher prevalence of inherited disorders due to consanguinity [3], along with other challenges like low awareness, low education levels, poor infrastructure and medical training. [4] This makes all the achievements in public health service sector in these states under National Rural Health Mission (NRHM) insignificant which can lead to missing of high risk mothers and children with genetic disorders resulting in high maternal and infant mortality rates. [5] Initiatives like UMMID and NIDAN supported by the DBT aimed to assess needs, provide training, and evaluate basic screening for genetic and endocrine disorders in vulnerable populations in aspirational districts. This initiative highlights the genetics training program conducted for clinicians to effectively manage and prevent these disorders at the community level.

MATERIAL AND METHODS

Training occurred between May 6, 2019, and Oct 31, 2022.

Trainees were divided into four batches for hands-on training:

1. First Batch: September 2019 - March 2020
2. Second Batch: March 2021 - August 2021
3. Third Batch: September 2021 - March 2022
4. Fourth batch: May 2022 – Oct 2022

They received training in various techniques, including newborn screening (NBS) using dried blood spots (DBS) for conditions like hemoglobinopathies, thalassemia (Hb HPLC, Capillary electrophoresis), congenital hypothyroidism, congenital adrenal hyperplasia, biotinidase deficiency, galactosemia, and G-6PD deficiency by fluorimetric assays. diagnostic techniques like GCMS, TMS (LC-MS/MS) for inborn errors of metabolism (IEM) , karyotyping, chromosomal analysis and FISH (Fluorescence-in-situ hybridization) were covered. Bio dosimetry training for radiotherapy cases, PCR (Polymerase chain reaction), RNA/DNA extraction, cDNA synthesis, western blot assay, cell culture techniques, primer designing, and the use of tools like FASTA/BLAST were also part of the training. This extensive training took place in the genetic laboratory at Maulana Azad Medical College (MAMC) Delhi with each batch, lasting six months. Additionally, trainees gained experience in collecting DBS samples on filter paper from postnatal wards at MAMC. The training also encompassed principles, software usage for data interpretation, bioinformatics, test report interpretation, and prenatal risk estimation using biochemical test analytes, including next generation sequencing (NGS) /exome reports.

Clinical training involved evaluating patients with genetic disorders in morning clinics and a dedicated genetic clinic, lasting 4-6 hours each. Prenatal clinical training included observing invasive procedures like amniocentesis, Chorionic Villus sampling (CVS) , and cordocentesis at the department of Obstetrics and Gynecology, conducted once a week. The training also covered fetal autopsy, prenatal serum sampling for aneuploidies, and genetic counseling. Trainees maintained a log book to record their experiences.

RESULTS

A total of 33 in-service clinicians from the government and 2 from the private sector applied for training. Seven clinicians received training as scheduled, with some delays for the second batch due to the COVID-19 pandemic. Summary results have been shown in Table1 and Table 2.

Table 1: Diagnostic And Clinical Genetics Training

Diagnostic Genetics Training		
All four batches received diagnostic genetics training in the following aspects		
	All the four batches received training	Second and third batches received additional training

Molecular Genetics	❖ Extensive details of diagnostic techniques mentioned in material and methodology. ❖ Direct and indirect mutation screening and detection techniques ❖ Quality control and assurance measures ❖ CVS DNA extraction with tools to exclude maternal cell contamination	❖ Oxford Nanopore technology for COVID-19 genome sequencing ❖ Designing adapters, library preparation, and loading on the flow cell ❖ Working with the commander software
Cytogenetics	❖ Cytogenetic diagnostic techniques mentioned in material and methodology.	❖ Flow cytometry, comet assay, and single and double-stranded DNA break assays.
Biochemical Genetics	❖ Enzyme estimation using leucocytes and dried blood spots ❖ Biomarkers analysis using Enzyme Linked Immunosorbent Assay (ELISA) and untargeted metabolomics. ❖ Neonatal and prenatal screening using Victor 2D platform. ❖ Neonatal screening using Genomic Screening Processor-with trouble shooting. ❖ Qualifying biochemical phenotypes and literature review. ❖ Quality control and assurance measures, utilization of LJ curves, and QC sample preparation.	
Clinical Cytogenetics	❖ Interpretation of Microarray	
NBS techniques for hemoglobinopathies	❖ Isoelectric focusing and capillary electrophoresis ❖ Identification of differential elution in beta thalassemia diagnosis	ARMS PCR for common thalassemia mutations, and the third batch conducted Sanger sequencing for the HBB gene.
Clinical training		
- Trainees personally cared for over 250 patients with various genetic disorders in outpatient and inpatient settings during the first phase. - Due to the second wave of COVID-19, the cumulative clinical exposure in the second phase was limited.		
Prenatal Clinical Training included observation of	❖ Ultrasonography ❖ Amniocentesis (22 cases), Chorionic Villus Sampling (12 cases), and Cordocentesis (3 cases).	
Prenatal Serum Screening covered	❖ Aneuploidies using time-resolved fluoroimmunoassay. ❖ Pretest and post-test counseling. ❖ DBS for first-trimester screening.	
Fetal Autopsy	❖ The second batch also received exposure to perinatal pathology, including mitochondriopathy with electron microscopy .	

Table 2: Workshops, Additional Training And Academic Activities

For first batch	
Workshops, seminars, and journal clubs	❖ All topics mentioned in the methodology. ❖ Biostatistics, approach to IEMs, and Real-time PCR. ❖ Online seminars on basic genetic principles, genetic tests, genomic variation, IEMs, and Bioinformatics. (Also attended by second batch)
Additional trainings	❖ Dysmorphology and various Human Phenotype Ontology terms through Face 2 gene. ❖ Molecular docking experiments. ❖ Creation of electronic health records, including pedigree drawing. ❖ App development for a live dashboard for newborn screening. ❖ Development of an artificial tool for monitoring Retinopathy of Prematurity in the periphery.
Training in new technologies	❖ Field testing for hearing using oto-acoustic emission (OAE) and automated brain stem response. ❖ Newborn screening for hemoglobinopathies. ❖ Pulse oximetry for peripheral use.
For the second batch	
Training included	❖ Inherited renal pathologies, including tissue culture and initiating a cell line. ❖ Electron microscopy. ❖ Flow cytometry.
Academic activities comprised seminars on	❖ Adrenoleukodystrophy. ❖ GC-MS. ❖ Celiac disease. ❖ Prenatal testing. ❖ Various permutation combinations.
Additional training involved	❖ Estimation of free fatty acids. ❖ Estimation of omega 3 and 6 fatty acids on DBS.
For the third and fourth batch	
Cytogenetics and Molecular Genetics	❖ Topics mentioned in methodology covered.
Biochemical Genetics	❖ Demonstrations and hands-on experience covering all diagnostic techniques and clinical training as outlined in the methodology.
Additional training	❖ Estimating prenatal risk using biochemical test analytes, supported by software. ❖ HLA-Typing (PCR-based), TTg, citrulline, and zonulin levels estimation for diagnosing celiac disease. ❖ Estimating urinary gycosaminoglycans for diagnosing mucopolysaccharidoses and other enzymatic estimation assays for lysosomal disorders, biotinidase, and aryl sulfatase. ❖ Genetics Clinic training, including dysmorphology assessment, pedigree analysis, interpreting genetic testing reports, genetic counseling and syndromic diagnosis using dysmorphology databases. ❖ Assessing and evaluating suspected mitochondrial disorders, peroxisomal disorders and disorders presenting with developmental delay and neuroregression.

	❖ Gained experience in immunohistochemistry. ❖ Interpretation of x-rays for diagnosing skeletal dysplasias and interpreting CT and MRI reports for diagnosing neurodegenerative and other neurological disorders.
Seminars and academic topics	❖ NBS. ❖ Prenatal diagnosis. ❖ Commander software in Oxford Nanopore microscopy and FISH. ❖ OAE. ❖ Thalassemia and Hemoglobinopathies in NBS. ❖ Participation in the mission NEEV (Neonatal early evaluation vision) sensitization program and metabolic-related activities.

DISCUSSION

For right diagnosis and appropriate genetic testing, it is important to understand a patient's needs. Unfortunately, many medical students, doctors and public lack basic genetics and comprehensive genetic testing awareness. [6,7] Aggarwal and Phadke's 2015 report highlighted the scarcity of qualified Clinical Geneticists in India, with only 40 trained professionals available[8]. Poor genetic knowledge of physicians results in delayed diagnosis and treatment which increases the overall burden of disease, raising challenges for both patients and doctors. Hence, it is important to integrate clinical and diagnostic genetics training programs for clinicians and preventive measures into India's public health system. Such integration can identify high-risk individuals and help them understand the implications of genetic testing, facilitating informed decisions.

Although integration of genome testing into public health settings is a complex and expensive process but it can be made more affordable and accessible by joint efforts like government funding, insurance companies, and public-private partnerships. Physicians can benefit from various awareness programmes such as personal interactions, awareness campaigns, seminars, workshops and mass media, which will provide accurate information and improve patient care.[6,9]

The involvement of the Medical Council of India and the Board of Education is crucial in training clinicians to incorporate genomics into their practice. NITI Aayog has recommended measures to harness AI-based genomics algorithms in India.

The six-month genetic test training program for clinicians under the UMMID initiative at MAMC underscores the need for more similar training programs and certificate courses for clinicians which will enhance the accessibility and affordability of healthcare for all in India.[10]

Acknowledgments

- We thank the Dean of Maulana Azad Medical College and the Medical Superintendent of Lok Nayak Hospital.
- Gratitude to the Department of Biotechnology, Division of Genetics MAMC , Department of Obstetrics and Gyanae, MAMC , GB Pant Hospital, Department of Pathology MAMC, Faculty members, Patients and Trainees.

Source of Financial Support: Department of Biotechnology. No. BT/Med/NIDAN-TRG/04/2018, 28.03.2019

Conflict of Interest: None

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