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Genetic testing applications in non-communicable diseases (NCDs): reproductive health, rare diseases & oncology

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EDITORIAL

On May 10, 2025, a magnificent assembly of specialists, scientists, and medical professionals gathered for a seminar hosted by **InBOL Healthcare** in partnership with **Thermo Fisher Scientific** and **BioApps** to discuss the genetics of **non-communicable illnesses**. Professionals from a wide range of fields came together at the event to discuss recent developments in genomic research and how they may help us understand non-communicable diseases. Renowned visitors were invited to the conference and offered valuable insights during the talks. **Dr Susmita Banerjee**, a well-known **paediatric nephrologist** from Kolkata; **Dr Ananda Chanda**, a research scientist from **ICMR-NIRBI**; **Dr Debprasad Chattopadhyay**, founder, director, and scientist G at ICMR-National Institute of Traditional Medicine; **Prof. Pranab Roy,** senior consultant at the **Institute of Child Health**, Kolkata; **Prof. Satinath Mukhopadhyay**, InBOL senior professor (former professor and head of IPGMER); Prof. Biswajit Bera, InBOL professor (former director of research, Tea Board of India); and **Prof. Sankar Kumar Ghosh**, professor in the Department of Biotechnology at Assam University and founder of InBOL, were among the invited speakers. Bioapps and Thermo Fisher Scientific both provided representatives to the sessions, who enriched the discussion with their insights and knowledge. **Mrs Anamita Ghosh, Director, InBOL, invited all the participants and experts on NCDs.**

Welcoming the Guests of Honor

A total of **43 participants** attended the seminar, representing a diverse range of academic and professional backgrounds. The audience included clinicians, scientists, researchers, and faculty members, fostering rich and multidisciplinary discussions. Notable medical professionals were present from leading institutions such as **IPGMER, NRS Medical College**, Netaji Subhas Chandra Bose Cancer Hospital, and several **dental and medical colleges in Kolkata** and Jamshedpur. The event also saw participation from faculty, scientists, and research scholars from esteemed institutes, universities, and centres, including **IGE Kolkata, IISER Kolkata**, CSIR-IICB, Assam University, and others across West Bengal and neighbouring regions. This broad representation contributed to an engaging and insightful seminar.

Founder of InBOL Prof SK Ghosh Addressing the guests and participants

The seminar commenced with an inauguration ceremony at 10:30 AM, led by esteemed guests. The inaugural session and facilitation were hosted by **Prof. S. K. Ghosh,** setting a professional and engaging tone for the discussions that followed. The seminar provided a platform for intellectual exchange, **addressing key concerns in the genomics of non-communicable diseases and fostering collaboration among**

Appraisal and Encouragement to InBOL's Effort

Dr Susmita Banerjee, an expert in **paediatric nephrology**, delivered a compelling talk emphasizing **the importance of genetic testing in diagnostics**. She acknowledged InBOL for organizing the seminar, recognizing its significance in bridging the gap between conventional biochemical diagnostic methods and the advanced insights offered by genomic analysis. **She pointed out that while chemical and biochemical parameters are commonly used in disease diagnosis, they fail to provide a comprehensive picture of underlying genetic factors.** Dr. Banerjee also highlighted a crucial challenge—the high costs associated with genetic sample processing—which remains a barrier to widespread adoption of **genomic testing in clinical practice**.

Dr Susmita Banerjee

Dr Ananda Chanda

Dr Ananda Chanda offered a heartfelt reflection on his childhood experiences, weaving them into the broader theme of holistic health. Drawing inspiration from the teachings of Swami Vivekananda, he emphasized that health is not merely the absence of disease but an intricate balance encompassing spiritual and mental well-being. He underscored the importance of **preventive healthcare, aligning with contemporary approaches that aim to mitigate disease risks** before they manifest. In this context, he **commended InBOL for its forward-thinking vision, particularly in West Bengal and across India**, highlighting its contributions to advancing preventive healthcare strategies through genomics.

Dr Debprasad Chattopadhyay shared insights from his extensive research experiences, reflecting on the progress made in clinical investigations. He lauded the founder of InBOL for successfully conceptualizing an institution that brings together numerous intellectuals and thought leaders in the realm of clinical research. He acknowledged that this initiative has fostered collaboration among experts, paving the way for groundbreaking studies and discussions on disease genomics.

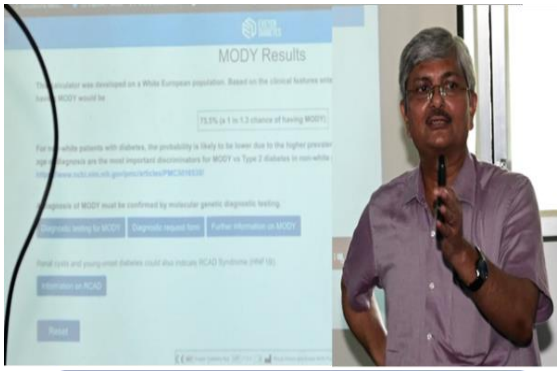
Dr Debprasad Chattopadhyay

Prof Pranab Roy

Prof Pranab Roy from the Institute of Child Health extended his appreciation to InBOL for its innovative approach to medical research. He reminisced about the strong academic bonds among InBOL's founders, mentors, and faculty members, many of whom are alumni of the prestigious Bose Institute. He noted that InBOL has provided them with a rare opportunity to work together once again, reinforcing the spirit of academic camaraderie. Prof. Roy highlighted molecular diagnostics as a crucial frontier in combating rare and inherited diseases and commended InBOL for spearheading initiatives in this transformative field.

Case Studies and Molecular Diagnostics: Need of the Hour

The seminar commenced with an insightful inaugural session led by **Prof. Satinath Mukhopadhyay**, who delivered a lecture on *Monogenic Diabetes*. He provided a detailed overview of various cases of diabetes, highlighting the genes responsible for different pathophysiological manifestations. His presentation emphasized the importance of understanding the genetic basis of diabetes to enable precise diagnosis and targeted therapeutic interventions. By discussing real-world case studies, he illustrated how genetic profiling can aid in distinguishing between different subtypes of diabetes, thereby informing personalized treatment strategies.



Prof Satinath Mukhopadhyay

Dr Dipanjana Datta

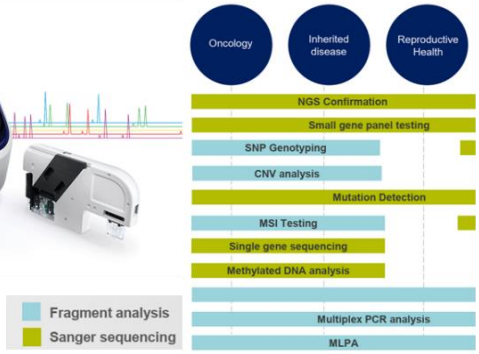
Following the inaugural session and a short tea break, **Dr. Dipanjana Datta**, a distinguished consultant medical geneticist, delivered a compelling talk titled *Management of Genetic Disorders and Importance of Personalized Medicine*. She elaborated on various hereditary diseases, including *Thalassemia*, *Spinal Muscular Atrophy (SMA)*, and *Duchenne Muscular Dystrophy (DMD)*. Her presentation underscored the significance of personalized medicine in managing these disorders, illustrating how genomic advancements have led to improved diagnostic accuracy and tailored treatment approaches. By emphasizing early screening and molecular interventions, she showcased how genetic testing plays a pivotal role in mitigating the impact of inherited disorders.

The next session was conducted by **Prof. Sankar Kumar Ghosh**, who discussed *Non-Invasive Prenatal Testing and Orphan Drugs*. His presentation focused on the application of these methodologies in **InBOL's research on hemoglobinopathies.** He elaborated on how non-invasive techniques can improve prenatal diagnostics by reducing procedural risks while ensuring accuracy in detecting genetic abnormalities. Additionally, he discussed orphan drugs—pharmaceuticals developed specifically for rare conditions—and shared perspectives on their application beyond hemoglobinopathies, advocating for broader implementation in the treatment of other genetic diseases.

Prof Sankar Kumar Ghosh

SeqStudio Sanger Sequencer: Stream Lining Research-Diagnostics at InBOL

The set of case studies and advancement of molecular diagnostics highlighted the future of disease diagnostics and personalized treatments. The presentations set the stage for the Field application specialists from Thermo-Fisher scientific, who introduced multiple diagnostic assays possible through SeqStudio Sanger Sequencer present at InBOL itself.



Dr Sanjib Dey

Before the lunch break, two technical sessions were presented by industry experts. **Dr. Sanjib Dey** from *Thermo Fisher Scientific* introduced the application of *Sanger Sequencing* and *Fragment Analysis* in cancer genomics. He demonstrated how these techniques are utilized for genetic profiling in oncology, facilitating early detection and precise treatment planning. His session illustrated the real-world applications of sequencing methodologies in personalized cancer therapy. Subsequently, **Dr. H Madhuri Doss**, *Senior Application Specialist at BI Biotech India Pvt. Ltd.*, delivered an online talk on *MLPA (Multiplex Ligation-dependent Probe Amplification) Technology* and its utility in cancer genetics. She explained how MLPA enables rapid identification of genetic mutations linked to cancer predisposition, streamlining diagnosis and enhancing targeted treatment approaches.

Post Lunch Session

The post-lunch session was led by **Prof. Sankar Kumar Ghosh,** who gave an in-depth introduction to the different gene panels available for conditions such as breast cancer, neurological disorders, cardiovascular diseases, and hemoglobinopathies. He also shared an innovative concept—**the DNA QR Code for individual human identification,** which could revolutionize personalized healthcare. **Dr Sanjib Dey** delivered another insightful session, discussing the molecular diagnosis of rare and inherited diseases.

Post-lunch, an online session was conducted by **Dr. Anil Kumar**, Technical Application Specialist II at Thermo Fisher Scientific. He provided an in-depth demonstration of various software tools used for analyzing raw genomic data, specifically in diagnosing inherited disorders, rare diseases, and cancer genomics. His session emphasized the importance of computational genomics in interpreting complex datasets to facilitate precision medicine.

Prof Biswajit Bera and Prof Sankar K Ghosh along with InBOL team during query session and vote of thanks

Dignitaries and participants enriching the event with their queries and feedback

Overall, the event was a resounding success, marked by enthusiastic participation from experts, researchers, and clinicians across diverse disciplines. The seminar fostered meaningful discussions on genomics in non-communicable diseases, showcased cutting-edge diagnostic technologies, and highlighted the value of interdisciplinary collaboration. Invited talks, case studies, and industry demonstrations offered participants insightful perspectives and practical knowledge, which were widely appreciated. The positive feedback from speakers and attendees alike reaffirmed the seminar's significance, positioning InBOL Healthcare as a prominent figure in advancing molecular diagnostics and personalized medicine research in the region.

The seminar concluded with an engaging discussion, where experts addressed participant queries and provided valuable feedback. **Prof. Biswajit Bera** and **Prof. S. K. Ghosh** delivered the vote of thanks on behalf of InBOL Healthcare, expressing gratitude to all speakers, researchers, and attendees for their contributions.

InBOL at a Glance: Commitment for Research -Diagnostic services

Services to Project Students/Researchers and Clinicians for the Genome Technology and allied project works. PhD/MD/Master's project/Post MSc/PhD can get services or do hands-on for respective projects. **Genetic and Genome Service@InBOL: please contact admin@inbol.org**

Healthcare and Clinical Services: Diagnostic Testing: Confirms or disproves genetic conditions that may be present in symptomatic individuals (e.g., uncommon disorders or sickle cell disease). o Single-Gene Testing: Detecting alterations in a known gene. Whole Exome Sequencing (WES): Examines all protein-coding genes. WGS: Analysing a person's entire DNA. **Predictive/Pre-symptomatic Testing**: Assessing risk of developing conditions (e.g., hereditary cancer risk like BRCA1/2 mutations) before symptoms arise. **Gene panels:** Obesity, Diabetes, Breast Cancer, PCOS, Autism, Inherited genes, etc. The process of profiling a malignant tumour involves sequencing its DNA to find alterations that can guide targeted therapies (Oncology).

Reproductive Health: Assesses foetal risk and informs family planning through PGD, carrier screening, and **NIPT/NIPS**. **Genetic Counselling**: Expert counsel and support for patients and families on genetic test results, inheritance patterns, and medical care.

Service Research and Development: **Thesis based Genomics services: Sequencing, Analysis, Paper writing and Thesis writing.** Bioinformatics and Data Analysis: Assemble, annotate, and interpret large **Next-Generation Sequencing (NGS)** datasets. Pathogen Genomics: Sequencing infectious pathogen genomes for outbreak surveillance, transmission tracking. Population Genomics: Analysing genetic diversity, illness risk, and gene-environment interactions in large cohorts. **Encourage to do PhD**.

InBOL provides high-throughput research diagnosis on genetic signatures (mutations), NGS-gene panel, miRNA profiles and Gut microbiome profiles to understand or **recommend Precision Medicine**