# Necessity of a Genome Sequencing Laboratory in a Nuclear Medicine Facility in Bangladesh for Research, Diagnostic and Therapeutic Services

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## ABSTRACT

The importance of genome sequencing technology in healthcare research and the development of precision diagnostics and therapeutics cannot be overstated. The knowledge of genomic research is pivotal for elucidating how genes contribute to various biological functions, including development, immunity, and disease susceptibility. In Bangladesh, a country with a growing burden of infectious, non-communicable diseases, genetic diseases, and other complex health conditions, establishing a genome-sequencing laboratory within a Nuclear Medicine and Allied Sciences Facility is paramount. This report represents the scientific and medical justifications for establishing a state-of-the-art genome sequencing facility in Bangladesh. It further underscores the necessity of establishing such a laboratory, emphasizing its potential in disease-related research, diagnostic accuracy, and personalized therapy to improve the overall healthcare outcomes in this country.

***Keywords:*** Bangladesh, precision diagnostics, genome sequencing technology, personalized therapy, Nuclear Medicine

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## INTRODUCTION

On April 14, 2003, the world learned that the Human Genome Project had been completed (1). It was one of the greatest scientific accomplishments in history and the first completed human genome sequence. A genome refers to the complete set of genetic material in an organism, encompassing all of its genes and non-coding sequences of DNA. It is the genetic blueprint that represents an organism's structure, function, and development (2). Genomic research has expanded our understanding of biological processes at a molecular level. A primary focus of genomic research is the identification of genetic variants that are associated with complex diseases. Therefore, genome-sequencing technology in healthcare can be helpful in disease detection, treatment, prevention, the identification of genetic predispositions, and the development of personalized therapeutic strategies.

Bangladesh, with a population of over 160 million people, encounters considerable healthcare challenges, including high rates of infectious diseases, such as tuberculosis, malaria, and dengue, as well as an increasing prevalence of non-communicable diseases (NCDs) like cancer, diabetes, and cardiovascular disorders (3). Moreover, genetic disorders like thalassemia and sickle cell anemia are also prevalent. Despite advancements in the healthcare sector, the capacity for comprehensive diagnostics and personalized medicine remains limited.

Nuclear medicine facility in Bangladesh might plan to establish a genome-sequencing laboratory to improve disease diagnosis, treatment, and prevention, enabling researchers to understand disease genetic variations and develop new treatments.

### *The Role of genome sequencing in disease-related research*

Bangladesh faces infectious diseases like tuberculosis, malaria, and dengue fever. Genomic sequencing helps identify genetic variations, predict outbreaks, and develop vaccines and treatments, particularly in identifying multidrug-resistant strains.

Understanding the genetic variability and mutation patterns of Dengue virus (DENV-2, DENV-3) was found to be critical for developing effective control strategies, as these serotypes are more prevalent in Bangladesh (4).

Similarly, during outbreaks like the COVID-19 pandemic, viral genomes were used to identify new variants, offering

insights into transmissibility, vaccine effectiveness, and necessary public health interventions (5).

Apart from infectious diseases, genetic disorders such as thalassemia, sickle cell anemia, and congenital heart defects are prevalent in Bangladesh. Genome sequencing can pinpoint the exact genetic mutations responsible for these conditions, leading to early diagnosis, carrier screening, and genetic counseling.

In oncology, genomic techniques are being used to detect genetic mutations associated with cancers. Cancer is an intricate condition marked by genetic and epigenetic changes (6). Similar to other countries, Bangladesh is experiencing an increased burden of cancer. Gene-expression-profile-based cancer biomarkers involve examining the gene expression patterns in cancer cells to gain a deeper understanding of tumor characteristics, prognosis, and therapeutic response (7). Such as: Oncotype DX is a genomic test that assesses the expression of a panel of about 16 genes involved in breast cancer. MammaPrint is a gene-expression-based assay used to analyze the activity of a set of genes (~18 genes) in breast cancer. It provides a genomic risk score (RS) that helps determine the risk of distant metastasis and assists in treatment decision-making, particularly in early-stage breast cancer. Similarly, VeriStrat is a blood-based protein signature test that measures the expression levels of specific proteins in the serum of lung cancer patients. It categorizes patients as either "VeriStrat Good" or "VeriStrat Poor," indicating the likelihood of response to certain therapies, including EGFR tyrosine kinase inhibitors (TKIs) (6).

***Diagnostic and Therapeutic Applications of Genome Sequencing***

In Bangladesh, where cancer is on the rise, incorporating genome sequencing into diagnostic practices could facilitate personalized treatment regimens that are both more effective and less harmful than conventional methods. Advances in technology, along with a deeper understanding of genetics and disease mechanisms, have been crucial in the development of precision and personalized medicine. Precision medicine involves utilizing genetic and molecular data to identify specific disease subtypes and create targeted treatments. It involves analyzing a patient's DNA, RNA, and protein expression to understand the underlying mechanisms of disease better and customize therapies for the individual (8).

A major advantage of genome sequencing is its ability to detect genetic predispositions to various diseases before the onset of symptoms. Genomic screening programs can uncover individuals who were previously unaware of their increased risk for cancer and heart disease, facilitating risk management and early cancer detection (9).

Additionally, molecular biology techniques, such as polymerase chain reaction (PCR) and differential gene expression, have added important findings to the study of disease pathogenesis. These techniques have been increasingly applied for revealing the different profiles of normal and affected cells or tissues and also for the following-up treatment of certain diseases like minimal residual disease (MRD). The detection of changes in the level of transcription of certain genes using the approach has been a useful tool for the early detection of disease, improving patient survival (10). During the COVID-19 pandemic, the rapid and early detection of severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infections was the key to controlling the disease. Then in Bangladesh, all the laboratories providing emergency testing support for COVID-19 relied on real-time RT-PCR (11).

There are collections of molecular techniques used to analyze biological markers in the genome. Several molecular applications have been incorporated in routine diagnostic laboratories that are more user-friendly, cost-effective, and accurate. Commonly used molecular techniques are: (a) gel electrophoresis (agarose gel electrophoresis, polyacrylamide gel electrophoresis), (b) molecular hybridization (Southern blot, Northern blot, Western blot, dot blot assay, fluorescence in situ hybridization), (c) mutation detection (RFLP, RAPD), (d) DNA sequencing (automated, pyrosequencing, next-generation sequencing), (e) nucleic acid amplification-based techniques (polymerase chain reaction, real-time PCR).

Using these techniques, the presence of a mutation in DNA can easily be identified. DNA analysis can specifically diagnose the inherited disease at the genetic level. DNA-based tests are very useful in discovering the genetic disease of an individual or his offspring in

advance. These are also important for prenatal diagnosis of the hereditary disorders (6).

***Synergy with Nuclear Medicine***

Nuclear medicine techniques, such as positron emission tomography (PET) and single-photon emission computed tomography (SPECT), are frequently used to diagnose and monitor a variety of diseases, particularly cancers, renal diseases, and cardiovascular conditions. Integrating genomic data with nuclear medicine imaging could enhance the precision of diagnosis and enable the development of more targeted therapies. Genetic profiling of tumors could guide the choice of the most appropriate radiotracers, either established or new, enhancing imaging accuracy in cancer diagnosis and treatment monitoring. By combining genome sequencing with nuclear medicine, more precise, timely, and effective medical interventions could be achieved (12).

***Applications of genome sequencing laboratory in nuclear medicine facility***

- Genetic screening of cancer-associated mutations leading to early detection strategies and preventive treatments
- Detecting genomic alterations that contribute to cancer growth, metastases and recurrence
- Development of precision medicine and targeted therapy
- Prenatal diagnosis and testing of diseases
- Forensic science
- Gene therapy

## CONCLUSION

The establishment of a genome sequencing laboratory in a nuclear medicine and allied sciences facility in Bangladesh is essential for advancing disease-related research, improving diagnostic accuracy, and enabling personalized therapeutic approaches. The development of a modern infrastructure and a skilled workforce would be mandatory for such a laboratory, which would mark a significant step towards transforming the country's healthcare system. Securing government investment, along with collaborative partnerships with technologically advanced institutions, would be instrumental in creating a successful infrastructure for both research and public healthcare.

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