

## Leveraging NGS-Tech in Healthcare Applications

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**Amidst the slow but steady growth, India has a long road ahead to become a master in the NGS market**



India is making huge strides in the next-generation sequencing (NGS) market. It has to be remembered that the sequencing of the SARS-CoV-2 has helped to develop vaccines in record time, track the emergence of viral variants and invest in timely strategic plans to control the spread. Having a genome database in place for the Indian population will ease the process of developing companion diagnostics and personalised medicine. Realising the potential of genome sequencing, governments, startups and healthcare companies are foraying into the market.

Human genome sequencing has the potential to determine the role of individual genes and their ability to cause diseases. Data generated through these sequencing technologies offer opportunities to assess genetic predisposition to a disease, diagnose heritable cancers and develop tests to prevent adverse drug reactions. The availability of the genome database for the Indian population is likely to ease the process of developing companion diagnostics and personalised medicine and offers access to stratified samples for the research.

### The India market

Way back in 2009, India announced the sequencing of the first human genome. The IndiGen programme, funded by the Council for Scientific and Industrial Research (CSIR), aimed to undertake whole genome sequencing of a thousand Indian individuals representing diverse ethnic groups from India. The programme aims to create a pilot dataset to enable genetic epidemiology of carrier genetic diseases towards enabling affordable carrier screening approaches in India. The human genome datasets would also be utilised for prioritising Pharmacogenomics variants specific to the Indian population for optimising therapy and minimising adverse events.

In the public sector, NGS is being used mostly for R&D activities and genome sequencing for building databases and for public health surveillance. In the private sector mostly working towards delivery. India is slowly adopting NGS as it has become an enabling technology for any life science domain. From basic research applications and is moving well towards discovery, development, quality assurance and quality control (QA & QC) and diagnostic applications.

According to the Indian Council of Medical Research (ICMR), as on March 23, 2022 India has NGS facilities available in 84 institutes spread across 24 States and 3 UTs, with 63 operational sites. These NGS platforms are mainly from Illumina, Ion Torrent, and Nanopore. However, a lot of NGS facilities in academic and government labs and private companies are already being phased out. With advances in technologies and applications, these facilities are unable to match world standards.

Back in 2015, there were majorly 2 to 3 players, while in 2021 there are about 13 to 14 primary players in the Indian market and about 3 to 4 companies have an optimal level of capacity and infrastructure to perform NGS workflows. The key advantage is the diversity in the populations, which assists in collecting a large pool of samples.

As **Dr Deepak Modi, Scientist, ICMR-National Institute for Research in Reproductive and Child Health** points out, "India has a bright future and will be the largest consumer market for NGS. With the 10000 Indian genomes sequenced and the availability of a reference database, we will be in a position to rapidly deploy in the diagnostic and personalised medicine segment in India."

### The growth story

Bengaluru-based Bionivid Technology has collaborated with numerous research institutions and universities in India and abroad, that offer contract research services in bioinformatics, genomics and NGS data analytics. The company plans to expand in two strategically important cities in the next six months.

Says **Roli Budhwar, Chief Operating Officer, Bionivid Technology**, "India holds a huge potential to be one of the biggest genomics markets based on NGS technology due to its rich genetic diversity. Healthcare diagnostics is currently the focus of various industry players. Other fields are still under-explored and carry huge potential. Promoting public-private partnerships, regulated by policies given the human resources with the right skill set and training, can push India to the front on the global stage. Overall the genomics industry in India is expected to grow in the coming years, driven by advancements in tech, increase in demand for precision medicine, expanding applications in agriculture and funding support."

Bengaluru-based Eurofins Genomics India delivers projects using advanced Illumina NextSeq500 platform, HiSeq2500, MiSeq, PacBio sequel with high-performance computational infrastructure. The company provides high-quality DNA/RNA oligonucleotides, NGS, Gene synthesis and sequencing services.

According to **Dr Vedam Ramprasad, Chief Executive Officer, MedGenome Labs**, NGS will remain one of the key technologies for the genomics industry across applications and will find additional use cases in combination with technologies for other omics. The increasing popularity and adoption of genetic testing in India will likely result in terms like NGS becoming common parlance (similar to RT-PCR in this post-COVID world). Primary growth drivers are improved awareness among clinicians and consumers, more accurate and precise diagnostics, new targeted therapeutics and a projected increase in disposable income of the Indian middle-class population.

MedGenome Labs has its primary lab in Bengaluru and according to the company, it is the South Asia's largest CAP-accredited NGS lab and houses the latest NGS equipment (at par with global industry leaders). The company additionally has five other labs across the country focusing on various molecular and genetic diagnostic technologies. These facilities apart from processing thousands of samples for genomic testing every month also support patients with genetic counselling and walk-in sample collections.

Mumbai-based Godrej Memorial Hospital recently introduced comprehensive genome testing that identifies an individual's predisposition to over 150 conditions and genetic traits. The hospital has partnered with HaystackAnalytics, based out of IIT Bombay, to launch the Health Genometer Smart Plan.

**Dr Anirvan Chatterjee, Chief Executive Officer, HaystackAnalytics** says that the human genome is a blueprint for life and DNA sequencing is the key to unearthing these insights. Genomics is changing the face of healthcare in therapy areas such as oncology, tuberculosis and infectious diseases as the technology of NGS has given breakthrough solutions in identifying the right pathogens and helping in the timely diagnosis of the disease.

Reliance Industries has announced its foray into genome testing through the acquisition of Bengaluru-based Strand Life Sciences in 2021 for a sum of Rs 393 crore for 80 per cent stake. The test kits developed by Strand Life Sciences will be rolled out at Rs 12000 i.e. 86 per cent cheaper than other available options in the market.

In October 2022 Illumina partnered with GenoScreen, an innovative genomics company, to launch a package combining Illumina products and the GenoScreen Deeplex Myc-TB assay. The targeted NGS based test for the rapid and extensive detection of anti-TB drug resistance.

Agilent, having its Diagnostics and Genomics (DGG) Center of Excellence (CoE) based at Mansear, Haryana, offers an extensive portfolio of NGS research products for quality control of nucleic acids, for library preparation and automation. It also offers tools for NGS data analysis and reporting. Agilent SureSelect library preparation and target enrichment reagents include custom and catalog panels, such as the SureSelect Human All Exon v8 panel and the SureSelect Cancer Comprehensive Genomic Profiling assay.

## Challenges

Amidst the slow but steady growth, India has a long road ahead to become a master in the NGS market. A host of challenges are currently being faced, the major ones being awareness, affordability and education and skilled resources. Very few healthcare workers are aware of the power of NGS technology. Another challenge is the required skill for NGS data processing and analysis.

Says **Dr Raja Mugasimangalam, Founder and Chief Executive Officer, Genotypic Technology**, "Rapid changes in technology, particularly the need to buy new equipment every three years is a big challenge. Pricing of the reagents, particularly with no local supplier and everything imported is a big problem. Lower-cost services from Korea and China are a big challenge for Indian service providers. Diagnostic companies that have received funds for molecular diagnostics based on NGS are cutting costs just to show some topline and entering the genomics services market."

Even one requires good hands-on wet lab protocol-based molecular biology techniques specifically for NGS data generation. Another important thing missing is the skill needed for analysing the big data generated through the NGS platform. Integration of computational technologies like Bioinformatics, Artificial Intelligence (AI), Machine Learning (ML) and biological sciences is the need of the hour.

Very few academic institutions have courses on NGS data analytics or bioinformatics in general. There is a lot of data being generated but the workforce with the right skill set to analyse and interpret this data is missing.

As experts point out, there are needs for some computational skills such as using Linux interface, R or python and also some coding abilities. The younger generation of biology graduates should acquire skills to make themselves market ready.

According to **Arpita Ghosh, Assistant Director Bioinformatics, Eurofins Genomics India**, there are too many small and new players after COVID-19 who have received some surplus funds. A huge price war is killing the market for service providers. It may be noted that government research institutes are not going for quality and heading for the lowest quote which is pulling the research level down.

Dr Modi goes on to add that quality control is another challenging area for the NGS market in India. There are a few India-specific guidelines for minimal essential standards for NGS data. As a consequence, there is often poor-quality data floating around and even used for clinical decision making which is quite dangerous. Also there is no Indian equivalence of the American College of Medical Genetics and Genomics (ACMG) guidelines to interpret and use the NGS data for clinical decision making. This makes it difficult for the counsellors and clinicians for effective decision making.

Talking about the challenges, **Samir Vyas, Country General Manager, India, Agilent** says, "The biggest challenge for the Indian NGS market is the lack of genotype-phenotype correlations for Indians at a population-wide and an individual-level, and most of the available databases are based on Caucasian populations. Large-scale genetic studies are required to identify actionable mutations and to create a database specific to the Indian population. A few initiatives have been taken in this direction. An example includes the Department of Biotechnology Genome India (a pan-India project which aims to catalogue

genetic variation in India) and is focussed on Whole Genome Sequencing of representative populations across the country."

Relatively high cost of sequencers and consumables, unavailability of local suppliers and vendors, low awareness and affordability for clinical applications and lower spending on genomics research in India, when compared to developed countries such as the US, are a few of the notable challenges in this sector. Apart from this, there are no national consensus guidelines on the use of clinical genomics to improve patient care.

### **Future of NGS in India**

NGS has been critical in decreasing the cost of sequencing, from \$1 million per whole genome in 2007 to \$200 in 2023, resulting in the relatively wide adoption of genomics (NGS based) across a variety of applications. Though India is lagging in NGS in the global arena, there is a huge scope for the country to excel in this sector.

Government should promote NGS and need to go hand in hand with the private players and ensure the right skill with the requisite funding.

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