GENOME SEQUENCING IN INDIA

INTRODUCTION

WHAT IS GENOME SEQUENCING & PRECISION MEDICINE

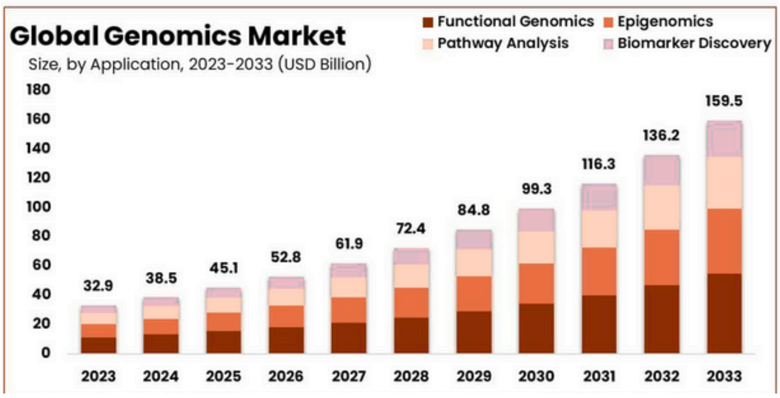
- Whole Genome Sequencing (WGS), also known as full genome sequencing or simply genome sequencing, is the process of determining the complete DNA sequence of an organism's genome in a single analysis. This involves decoding the entire set of chromosomal DNA, along with mitochondrial DNA in humans.
- In the context of healthcare, genome sequencing allows for the comprehensive analysis of an individual's genetic code, approximately 3 billion base pairs in humans. This provides critical insights into genetic variations that may contribute to health conditions, helping identify mutations responsible for inherited or acquired diseases.
- Precision medicine is a personalized approach to healthcare that uses genetics, lifestyle, and environment to guide diagnosis and treatment. It moves beyond the "one-size-fits-all" model to improve effectiveness and reduce side effects.

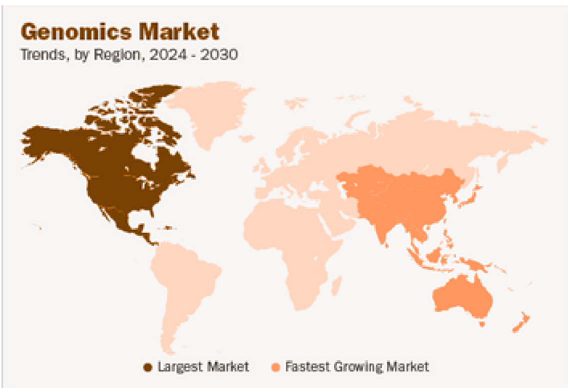
RATIONALE

- India's vast genetic diversity, along with the rising burden of non-communicable and rare diseases like thalassemia and sickle cell anemia, makes genomic sequencing vital for accurate diagnosis and targeted treatment. Yet, despite making up over 17% of the global population, India is underrepresented in genomic studies, with the Genome India Project covering only a small fraction of its 4,600+ populations. Expanding large-scale sequencing is crucial to capture this diversity, advance precision medicine, and bridge the global genomic equity gap.
- Beyond local healthcare, genomic sequencing gives India a strategic edge in global research. Mapping its genetic mosaic can reveal population-specific disease risks, drug responses, and preventive strategies that benefit both India and the world. Broader sequencing efforts will ensure inclusive data, reduce disparities, and position India as a major player in global biomedical innovation.

THE FUTURE OF GENOMICS

The future of healthcare is being transformed by advances in genetics and genomics, making personalized medicine increasingly achievable. Affordable genome sequencing, pharmacogenomics, and ancestry-based insights will enable precise disease prediction, tailored treatments, and preventive strategies, while wearables integrated with electronic health records will allow real-time, individualized care. Emerging tools like telegenetics, AI chatbots, and advanced computational methods will expand access to genetic counseling and improve interpretation of rare variants, while microbiome research may unlock targeted therapies to restore gut health. As healthcare systems adapt and professionals are trained in genomics, North America is expected to lead this growth, followed closely by the Asia-Pacific region.





GLOBAL ADVANCES IN GENOME ECOSYTEM

01 **Next Generation Sequencing:** Next-generation sequencing (NGS) has revolutionized DNA sequencing by enabling the simultaneous analysis of millions of DNA fragments, offering detailed insights into genome structure, genetic variation, gene activity, and behavior. Recent advances have improved speed, accuracy, cost-efficiency, and data analysis, unlocking new potential in genomics and disease understanding.

02 **Third Generation Sequencing:** Third-generation sequencing technologies represent the latest advancements in DNA sequencing, offering new approaches that overcome the limitations of previous generations. These technologies provide long-read sequencing capabilities, enabling the sequencing of much larger DNA fragments compared to earlier methods. Examples include PacBio and Oxford Nanopore

03 **CRISPR-Based Targeted Enrichment:** CRISPR-based enrichment methods use Cas9 to isolate specific genomic regions for sequencing without PCR amplification, greatly improving detection of structural variants, short tandem repeats, fusion genes, and minor alleles in mixed samples

04 **Spatial Transcriptomics:** Spatial transcriptomics maps gene expression across tissues while keeping spatial context intact. Tools like 10× Visium, Slide-seq, and DBiT-seq enable near single-cell resolution, showing where genes are active within tissues. Advanced models like stIHC, SpaRED, and SpaCKLE improve analysis accuracy and reduce missing data, making this method powerful for cancer and brain research.

05 **In Situ and Imaging-based Methods:** Microdissection-based techniques (e.g. Laser Capture Microdissection, Geo-seq), in situ hybridization (e.g. MERFISH, seqFISH), and in situ sequencing (e.g. STARmap, FISSEQ) capture spatial gene expression directly in tissue while preserving architecture

INDIA'S PLACE IN GENOMIC ECOSYSTEM

Cost Efficiency

Compared to markets like the US or Europe, India offers significant price advantages: high-quality genomics research can be performed at a fraction of the cost .

Expanding Genomic Infrastructure

India's genomic data infrastructure is growing rapidly boosting its role in affordable genomic data management among low-cost countries.

Direct Clinical and Commercial Relevance

With its high prevalence of CDs and NCDs, coupled with a large, genetically diverse population, India is well-positioned for medical innovation and is a strong market for such solutions.

Policy Support and Market Growth

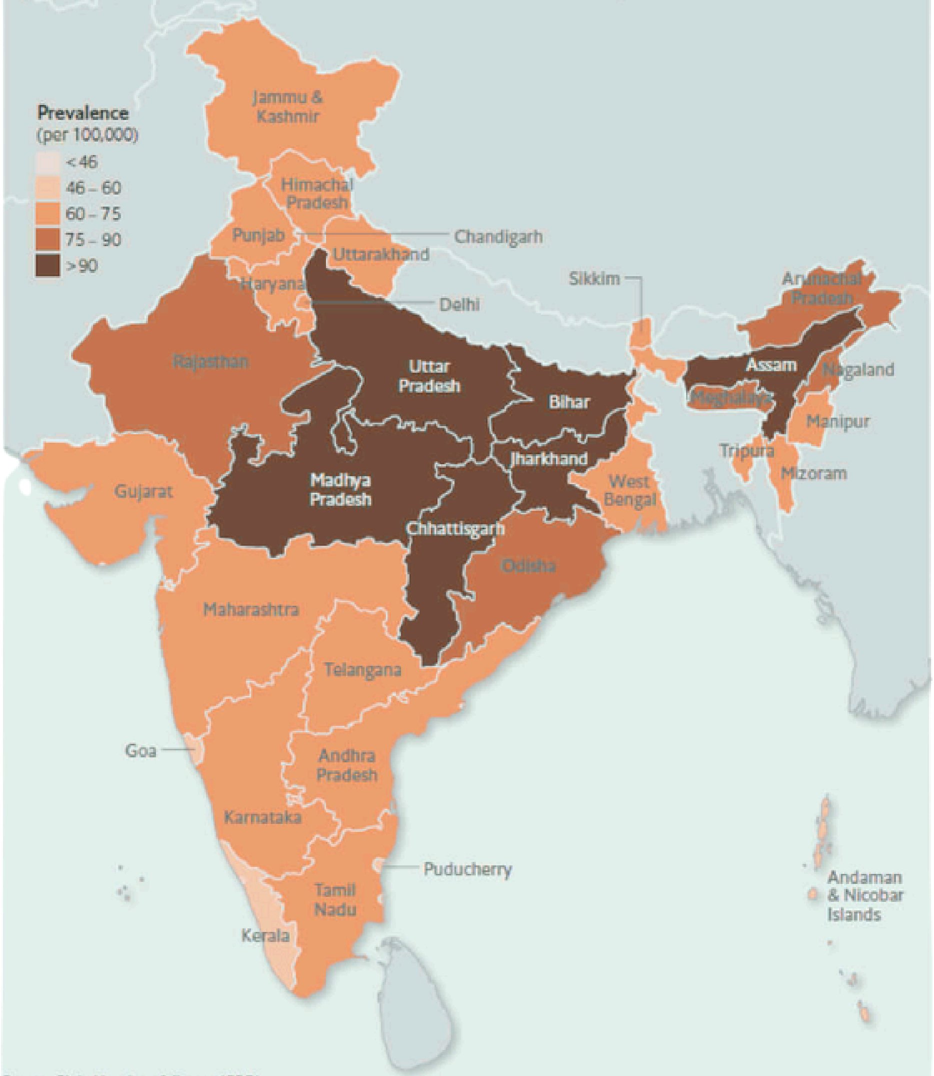
India's strong government push for genomics and a $1B NGS market by 2025 signal major growth and opportunities for innovation and partnerships.

INDIA'S DISEASE LANDSCAPE

India's genomic disease landscape is shaped by deep population structure and long-standing endogamy, producing strong founder effects across hundreds of caste, regional, and tribal groups. Genome-wide studies show founder events dating back ~120–3,500 years across diverse ethno-linguistic groups, implying elevated recessive disease risk in many large communities. This structure manifests in concentrated burdens of haemoglobinopathies and rare disorders—for example, sickle cell disease (SCD) clusters in central, western, and eastern tribal belts, with systematic reviews estimating higher prevalence in states such as Madhya Pradesh, Chhattisgarh, and Maharashtra. β-thalassaemia carrier frequency ranges from ~0.3% to ~17% depending on community, further underscoring how genetic risk is tied to local ancestry rather than uniform national averages.

Addressing these challenges requires systematic genomic integration into public health. Initiatives such as the National Sickle Cell Anaemia Elimination Mission are already scaling up community-level screening, genetic counseling, and early treatment in high-prevalence tribal belts. Broader solutions include expanding premarital and prenatal carrier screening programs, particularly in regions with known high-risk communities; building population-specific genomic reference databases for accurate diagnostics; and strengthening rare disease registries to support early detection and access to therapies for conditions like lysosomal storage disorders. While there have been some efforts by government schemes and a few private initiatives, the scale of India's inherited disease burden leaves considerable room for further innovation. More companies can step in to design low-cost, ancestry-specific testing, extend services to Tier II and Tier III cities, and collaborate with public health systems—creating significant opportunities for advancing preventive genomics and improving equitable healthcare delivery.

Figure 3: Prevalence of sickle cell disease across Indian states, 2017



Source: Global burden of disease (GBD).

India

India has one of the most genetically diverse populations, raising susceptibility to genomic disorders

70 Mn

Population affected by Genetic Diseases in India *~ICMR*

Sickle Cell

is one of most common genetic disease in India

215,380

No. of People with Sickle Cell Disease in India as of 2025 *~NSCP*

THE STRATEGIC NEED FOR GENOME SEQUENCING IN INDIA

OPPORTUNITIES

GENETIC DIVERSITY ACROSS REGIONS AND COMMUNITIES

- Wide Variability – India's vast ethnic, linguistic, and geographic diversity creates a rich genetic mosaic with distinct variations across regions and communities.
- Disease Impact – This genetic diversity influences susceptibility and resistance to various diseases, making it crucial to study diverse populations for accurate diagnosis and treatment.
- Strategic Advantage – Mapping this diversity through genome sequencing supports precision medicine tailored to India's unique population and boosts global biomedical research.

HIGH BURDEN OF NON COMMUNICABLE AND RARE DISEASES

- Non-communicable diseases (NCDs) - Like diabetes and heart disease are rising rapidly in India, posing a major health and economic burden.
- Rare genetic disorders - Often inherited, affect thousands but remain underdiagnosed due to limited genomic data.
- Examples include thalassemia prevalent in eastern India, sickle cell anemia among tribal groups, and cystic fibrosis found in select populations.

LACK OF REPRESENTATIVE GENOMIC DATA FOR INDIAN POPULATION

- India makes up over 17% of the global population but remains underrepresented in major global genomic studies.
- Its diverse genetics and social structures require broader and more inclusive genomic research.
- The Genome India Project's 10,000 genomes cover only a small fraction of India's 4,600+ distinct populations.

BRIDGING THE GLOBAL GENOMIC EQUITY GAP

- In conclusion, expanding India's diverse genomic data is essential to bridge the global genomic equity gap and enable inclusive, precise healthcare for all populations.

16.10%

CAGR FOR INDIA GENOMICS MARKET

INVESTMENT LANDSCAPE

- **CENTRAL GOVERNMENT FUNDING AND POLICY SUPPORT**

The Government of India provides substantial funding for genomic research through agencies like the Department of Biotechnology (DBT), Council of Scientific and Industrial Research (CSIR), and other central bodies. These funds support large-scale genome sequencing projects, development of sequencing infrastructure, bioinformatics platforms, and training of skilled personnel. Dedicated grants are also allocated for disease-specific studies, pathogen surveillance, and precision medicine initiatives, enabling the generation and application of genomic data to improve healthcare outcomes across the country.

- **ROLE OF SCIENTIFIC BODIES (DBT, CSIR, ICMR)**

Department for Biotechnology: A unique digital identity for secure access to health records across providers.

Council of Scientific and Industrial Research: Funds research programs like IndiGen, supporting genome sequencing and population genetics studies.

Indian Council of Medical Research: Allocates funds for disease-specific genomics and translational research linking genomics to public health.

Together, these funding initiatives help strengthen India's genomic research ecosystem by supporting infrastructure, training skilled personnel, and translating genomic data into actionable healthcare and public health solutions.

DBT, CSIR, and ICMR form India's scientific backbone, driving achievements like the indigenous DNA vaccine, large-scale genome projects, and key public health research.

- **GLOBAL COLLABORATIONS AND GRANT-BASED PARTNERSHIPS**

- Ensures protection of personal data, including sensitive health information, building trust in digital health technologies.
- Establishes clear guidelines for data consent, storage, and usage, promoting transparency and accountability.
- Enables healthcare providers and digital health solutions (e.g., telemedicine platforms, health apps) to handle patient data responsibly.
- Enhances data security and privacy, fostering wider adoption of digital health systems.
- Supports the growth of the digital health ecosystem by safeguarding patient information and ensuring compliance with data protection standards.

- **VENTURE CAPITAL AND PRIVATE SECTOR INTEREST**

- Promotes local manufacturing of high-quality medical devices, reducing reliance on imports.
- Supports the digital health ecosystem by ensuring access to affordable devices like wearable health trackers, diagnostic tools, and telemedicine equipment.
- Encourages innovation in medical device development, fostering integration with digital health applications.
- Creates opportunities for advanced solutions such as remote monitoring tools and diagnostic aids.
- Boosts the domestic healthcare industry by strengthening the supply chain and increasing self-reliance in medical technologies.

KEY NATIONAL INITIATIVES AND PROGRESS

India has steadily built a robust genomic research ecosystem through coordinated national policies, infrastructure development, and funding support. Over the past decade, the country has expanded sequencing facilities, established bioinformatics platforms, and promoted collaborative networks across research institutes, universities, and hospitals. These efforts harness genomic data for public health planning, disease surveillance, and precision medicine, with investments in high-throughput technologies, data repositories, and skilled manpower enabling advances in both human and pathogen genomics.

Progress has also been reinforced by regional and state-level initiatives capturing genetic diversity across India's populations. Programs targeting tribal and rural communities, as well as disease-specific cohorts, generate datasets that inform public health strategies and population genetics studies. Integration of genomics with digital health systems, like the National Digital Health Mission, is translating research into actionable healthcare interventions. Collectively, these initiatives highlight India's growing capabilities in genomics and reflect a strategic vision to leverage genomic insights for innovation in medicine, disease prevention, and global scientific leadership.

GENOME INDIA PROJECT

The GenomeIndia Project, led by the Department of Biotechnology (DBT) and coordinated by the Centre for Brain Research at IISc Bengaluru, is mapping India's genetic diversity by sequencing whole genomes from geographically, linguistically, and ethnically representative groups. A total of 20,000 samples from 83 diverse populations have been collected nationwide, forming a robust biobank for future research, with sequencing completed for 10,000 samples. The resulting India-specific genomic reference and allele frequency data will enable genomic medicine, diagnostics, and precision health solutions tailored to the country's unique genetic landscape, supported by publicly accessible databases and portals for researchers and clinicians.

INSACOG INDIAN SARS-COV-2 GENOMICS CONSORTIUM

The Indian SARS-CoV-2 Genomics Consortium (INSACOG), set up in 2020, is a network of 58 labs that sequences virus samples across India to track variants, inform public health measures, and support research, diagnostics, and vaccines, including detecting variants like NB.1.8.1 and LF.7.

INDIGEN PROEJCT BY CSIR

The IndiGen Project, launched by CSIR in 2019, sequences genomes of 1,008 Indian individuals to map genetic variations, understand disease predispositions, and create a database for public health and precision medicine, helping identify carriers of genetic disorders across India.

PRECISION MEDICINE & THE PROMISE OF TARGETED THERAPIES

- **ROLE OF GENOMIC SEQUENCING IN PERSONALISED DRUG DEVELOPMENT**

Genome sequencing involves decoding the order of the four chemical building blocks, or nucleotides, that make up an individual's DNA. This process provides a wealth of information about an individual's genetic makeup, including variations and mutations that may be associated with the disease.

Personalized medicine is a new approach to medical practice that utilizes an individual's **genetic makeup (Genomic sequences)** to make informed decisions about disease prevention, diagnosis, and treatment. Genetic information can assist doctors in selecting the most appropriate medication or therapy, as well as determining the correct dosage or treatment plan.

- **BENEFITS TO PHARMACEUTICAL R&D AND CLINICAL TRIALS**

Precision medicine has transformed pharmaceutical research and development by making drug discovery and clinical trials more targeted and efficient. Through improved drug target identification, researchers can develop therapies tailored to specific genetic or molecular patient profiles, increasing the likelihood of clinical success. This targeted approach contributes to higher success rates in clinical trials, reducing late-stage failures and optimizing resource use. It also enables patient stratification, allowing trials to focus on patients most likely to respond, which decreases trial size, duration, and adverse events, enhancing cost efficiency.

In addition, precision medicine supports the repurposing of existing drugs for new patient subgroups, leveraging known safety profiles and shortening development timelines. The use of companion diagnostics and biomarker-guided therapies provides regulatory advantages, such as accelerated approvals and precise labeling, strengthening both clinical and commercial viability. These combined benefits make precision medicine a key driver of innovation and efficiency in modern pharmaceutical R&D and clinical trials.

EMERGING STARTUPS IN INDIA

Company	City	Year Founded	Key Focus Areas
MEDGENOME	Bengaluru	2013	Genomic Diagnostics,Preventive Wellness Testing, Research & Collaborations,Drug Discovery & Pharma Licensing,AI/ML & Bioinformatics
4baseCare Together, We Beat Cancer	Bengaluru	2018	Precision Oncology Genomics, AI-Driven Clinical Interpretation, Indian-Population Cancer Panels , Solid & Liquid Biopsy (SoLiq)
strand	Bengaluru	2000	Bioinformatics & AI Platforms, NGS-based Clinical Diagnostics (oncology, rare diseases, NIPS), Preventive Genomic Wellness, Pharma & Biomarker Discovery, Global Precision-Medicine Partnerships
VGENOMICS	Delhi	2022	AI-driven Genomic Diagnostics for Rare Diseases, Whole Genome/Exome & RNA Sequencing, Clinical & Preventive Genetic Testing

EXISTING GAPS AND CHALLENGES IN GENOMIC ECOSYSTEM

PAIN POINTS

LACK OF AWARENESS AND SKILLED PROFESSIONALS

- India faces a significant shortage of trained human resources in genomics—especially in clinical genetics, bioinformatics, and genetic counseling. The IndiGen project, for instance, underscored the need to train more clinicians to interpret genomic data effectively.
- Furthermore, reports highlight that many labs lack trained manpower to provide genetic counseling and follow-up services, limiting the meaningful deployment of genetic tests.

LIMITED GENOME LITERACY AMONG PUBLIC AND PRACTITIONERS

- Public understanding of genomic science remains low, posing hurdles for acceptance and informed decision-making. Educating the public is crucial to dispel misconceptions, increase participation in research, and guide responsible genetic testing.
- Similarly, limited genome literacy among practitioners constrains their ability to integrate genomics into routine healthcare.

ETHICAL AND REGULATORY AMBIGUITIES

- India lacks a robust, unified legal framework governing the ethical use, regulation, and privacy of genomic data. While there is reliance on broad health ministry clearances, significant numbers of samples are sequenced or analyzed overseas with minimal oversight.
- Fragmented regulation leads to concerns over quality control, misuse of data, and potential genetic discrimination—for example, affecting insurance access or stigmatizing communities.

INFRASTRUCTURE & DATA STORAGE CONCERNS

- The vast volumes of data generated by genomic sequencing—up to 120 GB per genome—pose considerable storage, processing, and computational challenges. India still lacks a centralized, accessible data infrastructure; proposals to build a national “hub-and-spoke” repository under the Department of Biotechnology remain a priority.

< 100

CLINICAL GENETICISTS AND A HANDFUL OF COUNSELORS SERVE 1.4 BILLION PEOPLE IN INDIA

THE ROAD AHEAD BRIDGING THE GAP

- **STRENGTHENING POLICY,ETHICS AND DATA GOVERNANCE**

India's progress in genomic sequencing for precision medicine requires stronger policy, ethics, and data governance to maximize health benefits and reduce risks. Ethical frameworks must reflect India's diverse population, safeguard privacy, and ensure long-term data relevance through anonymization, informed consent, and the right to withdraw. Participatory governance models like the CARE Principles—Collective Benefit, Authority to Control, Responsibility, and Ethics—can enable community empowerment, equitable benefit-sharing, and protection from misuse or stigma. Building stringent, inclusive regulations and oversight bodies, aligned with global standards, will be key to creating a transparent and sustainable precision medicine ecosystem.

- **SCALING PUBLIC-PRIVATE PARTNERSHIPS**

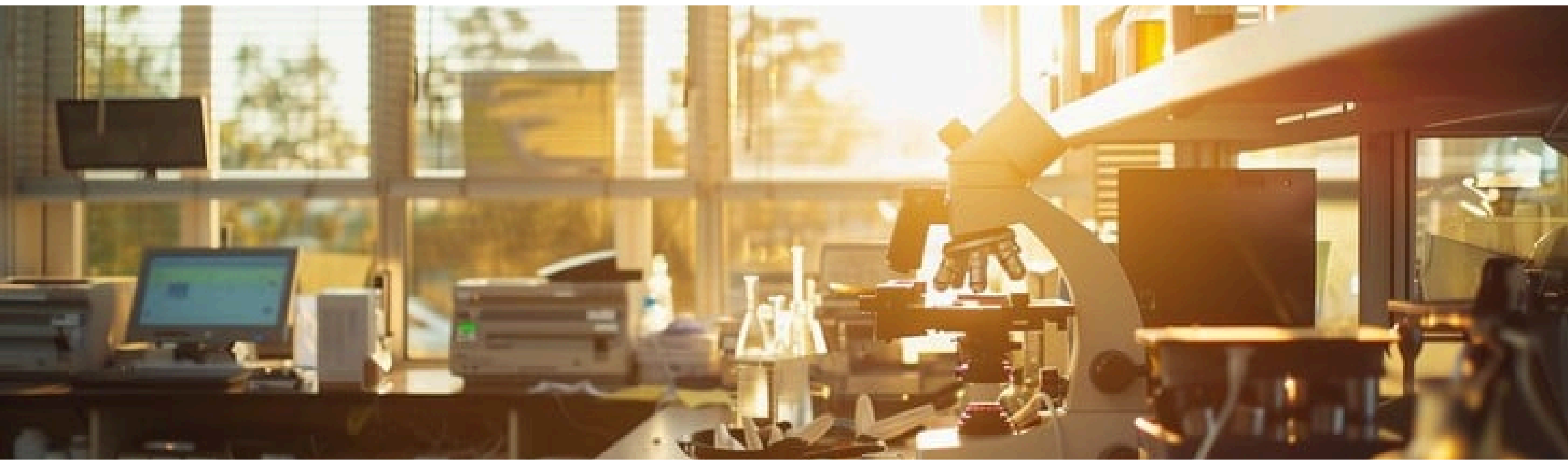
The road ahead for PPPs in India's genomic sequencing lies in establishing a dedicated Genome Research Council to regulate and drive initiatives with accountability. This body would unite public institutions, private industry, and philanthropy, including CSR funds, to pool resources while ensuring compliance and data privacy. Key priorities include co-development platforms, innovation hubs, and blended financing models to accelerate translational research and expand access. Strengthening government capacity in funding integration, data repositories, and oversight will be vital to scaling impactful PPPs that rival global efforts and deliver equitable outcomes for India's diverse population.

- **LOCAL MANUFACTURING AND INDIGENOUS TECHNOLOGY DEVELOPMENT**

The road ahead requires strengthening local manufacturing and indigenous technology to reduce dependence on imports and lower costs. Building robust R&D ecosystems, incentivizing startups, and fostering industry–academia collaboration will be crucial. Public support through policy incentives, infrastructure, and funding can accelerate homegrown innovation, while strategic partnerships and technology transfer can scale production. Together, these steps will ensure affordable, accessible, and sustainable genomic solutions tailored to India's diverse needs.

- **CAPACITY BUILDING IN GENOMICS EDUCATION AND RESEARCH**

The road ahead requires expanding genomics education through specialized university programs and integration into MBBS curricula, alongside continuous training for healthcare professionals. National training hubs, fellowships, and global collaborations can nurture skilled talent, while academia–industry partnerships and translational research centers will drive innovation. Strengthening public genomic literacy and ethical awareness will further build trust, enabling India to advance precision medicine and equitable healthcare.



CONCLUSION

India is at the cusp of a genomic revolution, moving from limited data and infrastructure to building the foundations of a world-class ecosystem. With initiatives like the Genome India Project and the Indian Biological Data Centre, the country has begun addressing critical gaps in data, research, and policy. Strengthening ethical governance, ensuring data privacy, and fostering inclusive participation will be key to creating a sustainable and transparent framework that empowers both researchers and citizens.

At the same time, investing in indigenous research, local manufacturing, and capacity building in genomics education will prepare a skilled workforce and reduce dependence on imports. Public–private partnerships, innovation hubs, and global collaborations can accelerate India's progress while ensuring equity and affordability. With coordinated efforts, genomics can transform Indian healthcare, positioning the country as a global leader in precision medicine and contributing solutions that extend beyond its borders.

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