

## Genome India project

### Why in news?

The Department of Biotechnology announced the completion of the Genome India Project the largest initiative to conduct sequencing of 10,000 genomes of the Indian population.

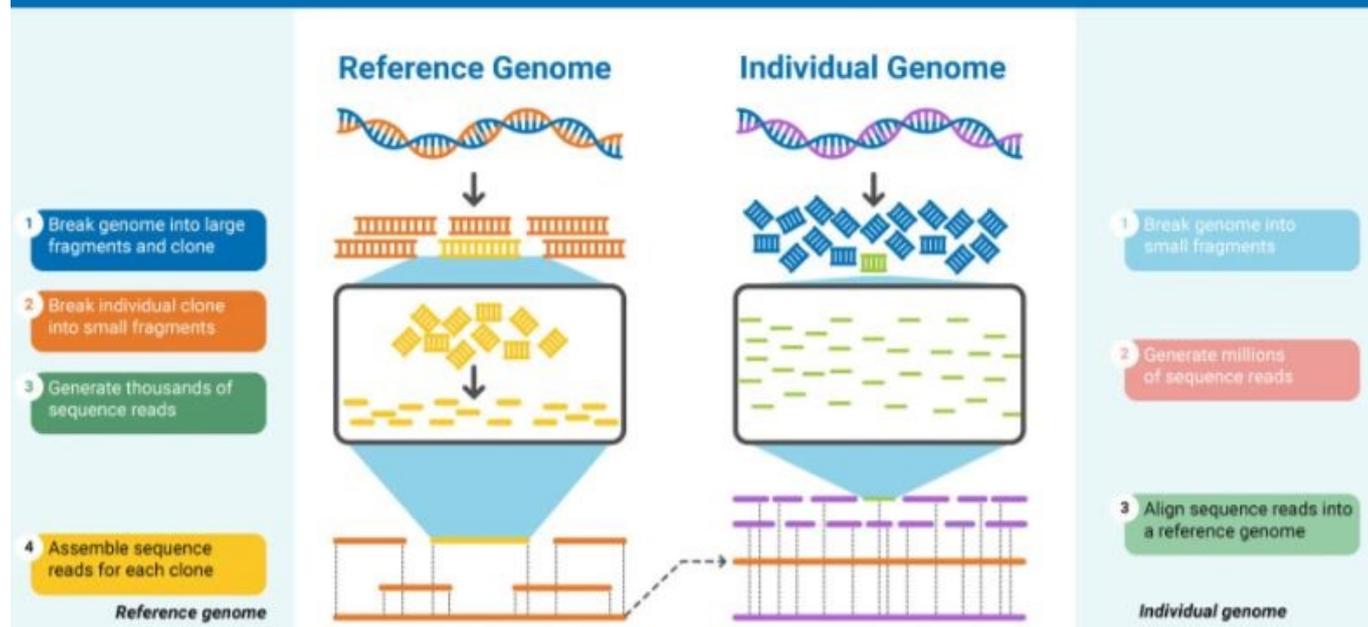
### Genome India Project

- **Launch year** - 2020.
- **Aim**- To create a comprehensive catalogue of 10,000 genetic samples from citizens across India, to build a reference genome.
- **Vision**- “Cataloguing the genetic variations in Indians” for 3 years (2020-2023).
- **Institutional support** - Indian Institute of Science’s (IISc) Centre for Brain Research.
- **Partner organisations** - 20
- **Fund** - By the Department of Biotechnology.
- **Genome sequencing method**- The project is based on the [Next-Generation Sequencing \(NGS\)](#) platform.
- **Next-Generation Sequence**- It involves fragmenting DNA/RNA into multiple pieces, adding adapters, sequencing the libraries, and reassembling them to form a genomic sequence.

### What is genome sequencing?

- **Genome**- It is defined as an organism’s complete set of *Deoxyribose Nucleic Acid (DNA)*, including all of its genes.
- Each genome contains all of the information needed to build and maintain that organism.
- In humans, a copy of the entire genome — *more than 3 billion DNA base pairs* — is contained in all cells that have a nucleus.
- The discovery that DNA is structured as a “*double helix*” was the spark in the long, continuing quest for understanding how genes dictate life.
- **Genome sequencing**- It is the state-of-art, robust and high throughput technique to sequence the entire genome of an organism.

# WHOLE GENOME SEQUENCING



## What are the advantages of the project?

- **Study genetic diversity-** The diverse genetic makeup of India, with over 4,635 anthropologically defined population groups, adds complexity and richness to the dataset.
- **Understand disease-** The data will help to compare and contrast the impact of genetic variations on physical health as India is the “largest genetic lab in the world.”
- **Rich dataset-** The project has successfully created a reference genetic database and a biobank containing blood samples from across the country.
- **Reference genetic database-** The completion of sequencing 10,000 genomes culminates in the establishment of a 'reference' Indian human genome which serves as a foundational template offering insights into the genetic makeup of the population
- **Database storage-** The project generated an extensive dataset of 8 petabytes, requiring 80 GB storage for each sequence making the dataset accessible to researchers.
- **Digital public good-** The data will be stored at the Indian Biological Data Centre in Faridabad, serving as a "digital public good.", it aims to foster advancements in diagnostics, therapies, and disease understanding.
- **Personalised health care-** A specific genetic mutations have been identified within the Indian population.

Genetic mutation	Effect	Prevalence
MYBPC3 Mutation	It is associated with cardiac arrest occurring at a young age	India-It is found in approximately 4.5% of the Indian population.
		Global- This mutation is considered rare.

<b>LAMB3 Mutation</b>	It leads to lethal condition	India- It is present in nearly 4% of the population near Madurai, Tamil Nadu.
		Global- This mutation is not observed in global databases

- **Precision medicine-** An Indian genome database will aid in understanding the genetic makeup to develop targeted treatments, especially for rare diseases resulting from genetic anomalies.
- **Drug discovery-** The database can contribute to the development of new diagnostics in potentially identify resistance-indicating variants.
  - **mRNA vaccines-** It is based on genetic mutations and identifying populations with specific resistance or susceptibility to certain medicines.
- **Gene therapy advancements-** The database holds immense potential for understanding genetic predispositions to diseases like cancer and lung diseases, it is also considered vital in comprehending infectious diseases like COVID-19.
- **Indigenization-** The project address the unique genetic challenges by deploying *Indian solutions using Indian data for Indian problems*, showcasing the country's commitment to utilizing its diversity for scientific advancements.
- **Technological advancement-** The Genome India Project highlights the remarkable progress in genomic sequencing technology because the 1<sup>st</sup> whole genome project required 13 years and 3 billion dollars to complete the project whereas this project is completed in 3-4 months.

### What are the challenges?

- **Expensive medicines-** The genome sequencing has opened up new complexities, despite advancements the accessibility and affordability of medicines for rare genetic conditions remain a challenge.
- **Monogenic disease-**The [Human Genome Project](#), completed in 2003, promised to decode the secrets of the genome and pave the way for personalized medicine but subsequent decades have revealed that only a small fraction of diseases are monogenic (caused by a single gene).
- **Privacy issues-** The issue of disclosing and managing incidental and secondary findings.
- **Huge dataset management-** The massive dataset generated (8 petabyte), poses significant challenges in terms of storage, management, and data security.
- **Genetic discrimination-** Some regions or ethnic groups may be underrepresented, affecting the project's dataset.
- **Limited representation-** The 10,000 genomes is a significant achievement but it may not fully capture the immense genetic diversity present in India's population of 1.4 billion people.

*There are over 4,600 distinct groups, and achieving comprehensive representation is challenging.*

## What lies ahead?

- As Genome India Project moves forward, its impact on healthcare, genetics, and scientific research is poised to be transformative, ushering in a new era of understanding and utilizing the genetic diversity within the country.
- The findings of the project should not remain confined to academia but involve collaborations with scientists, students, technology companies, ethicists, and social scientists.

## References

1. [The Hindu- Decoding Genome India Project](#)
2. [Indian Express- Explained Genome India Project](#)
3. [The Print- India largest genetic lab in the world](#)

