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.

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.
LAMB3 Mutation	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 1st 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

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