GENOME INDIA PROJECT – SCIENCE & TECHNOLOGY

NEWS: The **Department of Biotechnology (DBT)**, a government body responsible for advancing biotechnology research in India, has **made the genome data of 10,000 individuals publicly accessible** as part of the **Genome India Project (GIP)**.

WHAT'S IN THE NEWS?

- The dataset contains **genetic sequences from 99 ethnic populations** across India, making it the first such effort to comprehensively map the **genetic diversity of the Indian population**.
- This genetic database serves as a **"digital public good"**, meaning that it is available to researchers worldwide for **scientific and medical advancements**.
- The information can be used to **develop new diagnostic tools**, create **targeted therapies for diseases**, and **identify previously unknown rare diseases**.
- A major motivation behind this initiative is the **underrepresentation of Indian genomes** in global genetic databases. By making Indian genetic data publicly available, this project ensures that **Indian genetic diversity is adequately represented in global research**, leading to more effective and personalized healthcare solutions.

Key Takeaways of the Genome India Project (GIP)

Project Approval and Objectives

- The **Genome India Project (GIP) was approved by the Government of India in 2020** as a national initiative to create a **comprehensive catalogue of genetic variations** found in the Indian population.
- A **genetic diversity map** is essential for **understanding human evolution, disease susceptibility, and genetic disorders** unique to the Indian population.
- Existing **international genomic databases**, primarily based on **European and North American populations**, do not accurately reflect the **genetic makeup of Indians**.
- Indian genetic diversity is **likely different** from other global populations due to **unique evolutionary and environmental factors**, making it necessary to **develop a separate Indian genomic dataset**.

Collaboration and Expansion Goals

- The first phase of the project involved collaboration among **20 scientific institutions across India**, which worked together to **sequence the genomes of 10,000 individuals**.
- The project now has a **data storage facility**, a **data-sharing platform**, and an **operational framework** to ensure **efficient management and accessibility of genomic data**.

- With these foundational elements in place, the **Department of Biotechnology (DBT) aims to expand the initiative**, with the goal of sequencing **up to 1 million genomes in the future**.

Second Phase – Disease-Specific Sequencing

- The second phase of the project will focus on sequencing **the genomes of individuals with specific diseases**.
- This approach will allow scientists to **compare the genetic makeup of healthy individuals with those suffering from diseases**, leading to the **identification of genetic risk factors**.
- By pinpointing **specific genes that contribute to diseases**, researchers can develop **targeted therapies and preventive strategies**.

Significance of Creating a Genetic Database

Identifying Genetic Risk Factors for Diseases

- The genetic map developed through this project helps in identifying **genetic mutations or risk factors** responsible for various diseases.
- These insights can be used to create **precision medicine**, where treatments are tailored based on an individual's genetic profile.
- With this knowledge, researchers can **modify, delete, or add specific genes** to treat diseases that have a **genetic basis**.

Discovering Unique Indian Genetic Variants

- Researchers working on the **Genome India Project** have identified **135 million genetic variations** in the 10,000 genomes sequenced so far.
- Out of these, **7 million variations are unique to the Indian population** and are **not found in any global genetic databases**.
- This discovery reinforces the need for an **Indian genome database**, as many disease-related genetic markers **may not be present in global datasets**.

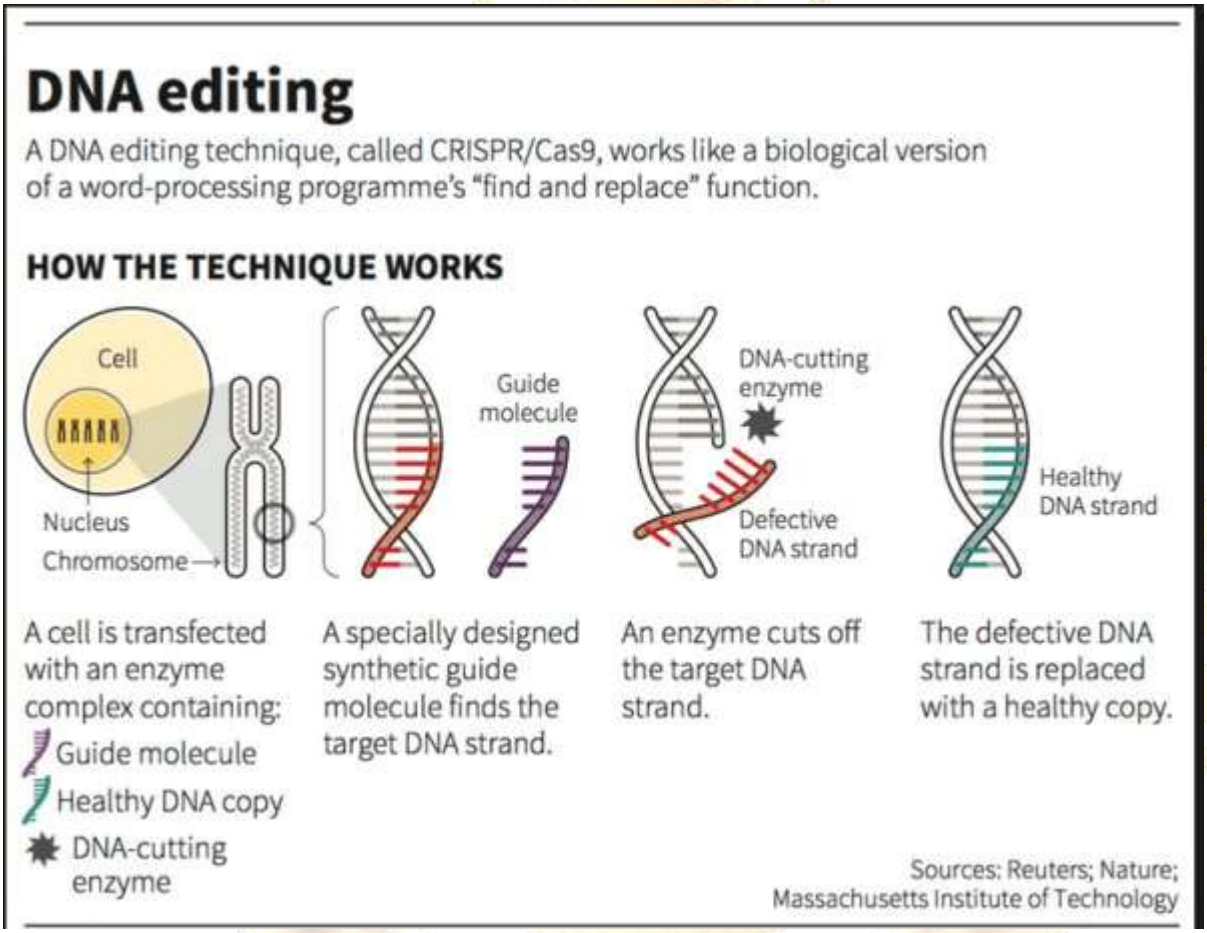
Understanding Disease Prevalence in India

- Population-wide genetic sequencing can help determine **how frequently certain genetic variations occur**, which in turn helps estimate **how common a particular genetic disease is**.
- **Example 1:** A mutation in the **MYBPC3 gene**, which increases the risk of **cardiac arrest at a young age**, is found in **4.5% of the Indian population** but is rare globally.
- **Example 2:** A mutation in the **LAMB3 gene**, which causes a fatal skin disorder, is present in **4% of the population near Madurai**, but is absent from global databases.

- These examples highlight **why India needs its own genome dataset**, as **genetic risks in the Indian population differ significantly from other populations**.

Identifying and Treating Rare Genetic Diseases

- Many rare genetic diseases remain **undiagnosed and untreated** due to the **lack of genetic data**.
- By studying genetic variations, scientists can identify **previously unknown rare diseases** and develop **gene therapies** to treat them effectively.



Identifying Drug Resistance Variants

- Certain **genetic mutations** can make **specific drugs ineffective or even harmful** for certain populations.
- **Example:** A sect of the **Vaishya community in South India** lacks a gene needed to **properly process common anesthetics**. If these anesthetics are administered, it can lead to **prolonged unconsciousness or even death**.
- By understanding such genetic risks, doctors can **personalize treatments and avoid prescribing ineffective or dangerous drugs**.

Understanding Human Genome and Genome Sequencing

The Human Genome – A Biological Blueprint

- The human genome is a **biological instruction manual**, composed of **3 billion base pairs**, which determine **all physical and biological characteristics**.
- The genome is written using **four chemical bases: A (adenine), C (cytosine), G (guanine), and T (thymine)**.
- These bases **combine in unique sequences**, defining an individual's **physical traits, disease risk, and biological functions**.

How Genome Sequencing Works

- Scientists extract **DNA from blood samples** and **break it into smaller pieces** for easier sequencing.
- The **A, C, G, and T sequences** of these fragments are recorded using **DNA sequencing machines**.
- Advanced computational techniques are then used to **reconstruct the complete genome** from these smaller fragments.

Global Genome Sequencing Efforts

- **2003:** The **Human Genome Project** completed and published the **first-ever human genome sequence**.
- **2012:** The **1,000 Genome Project** sequenced **1,092 genomes** through international collaboration.
- **2018:** The **UK Government sequenced 100,000 genomes** for medical research.
- **Ongoing:** A **European initiative** aims to sequence **1+ million genomes across 24 countries**.

Genome Editing – Correcting Genetic Disorders

What is Genome Editing?

- Genome editing is a technique that allows scientists to **cut and modify DNA** to **correct genetic defects, prevent inherited disorders, or enhance physical traits**.

CRISPR Technology – A Revolutionary Tool

- **CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats)** is a groundbreaking technology that enables **precise editing of the genetic code**.
- It works similarly to a **"cut-copy-paste"** function in a computer, where faulty DNA sequences can be removed and replaced with correct ones.

Potential Applications of Genome Editing

- Genome editing has the potential to **cure a vast number of genetic disorders**, including:
    - **Blood disorders** like **sickle cell anemia**.
    - **Eye diseases** such as **color blindness**.
    - **Cancers, diabetes, HIV, and heart diseases**.
- By combining **genome sequencing and gene editing**, scientists may soon be able to **find permanent cures** for many genetic diseases.

Conclusion

- The **Genome India Project** marks a **significant step forward in genetic research** and **personalized medicine in India**.
- It will lead to **better diagnostics, more effective treatments, and targeted therapies for various diseases**.
- Advancements in **genome sequencing and gene editing** hold immense potential for **transforming global healthcare**.

Source: https://indianexpress.com/article/upsc-current-affairs/upsc-essentials/genome-india-project-gene-editing-important-upsc-knowledge-nugget-9804177/