GUJARAT'S GENOME SEQUENCING PROJECT: SCIENCE & TECHNOLOGY

NEWS: Gujarat launches India's first tribal genome project to tackle inherited diseases

WHAT'S IN THE NEWS?

Gujarat has launched a pioneering genome sequencing project targeting its tribal population to identify hereditary disease burdens like sickle cell anemia and thalassemia, aiming to create a reference genome database for improved health outcomes. This initiative mirrors broader global efforts like the Human Genome Project and Genome India Project, emphasizing the importance of understanding genetic diversity for personalized healthcare.

Gujarat's Genome Sequencing Project: A Focus on Tribal Health

Gujarat has launched a groundbreaking initiative titled "Creation of Reference Genome Database for Tribal Population in Gujarat," as announced in its 2025–26 Budget. This project is the first of its kind, specifically targeting the state's tribal communities.

Key Aspects of the Project:

- Aim: To sequence the genomes of 2,000 individuals from various tribal communities across 17 districts in Gujarat.
- Focal Point: Identifying the burden of hereditary diseases, particularly sickle cell anemia and thalassemia.
- Implementing Agency: Gujarat Biotechnology Research Center (GBRC).
- Support: The project is supported by the **Department of Science and Technology** and the **Tribal Development Department** of Gujarat.
- **Infrastructure:** It includes state-of-the-art facilities for:
 - Sample collection
 - Genome sequencing
 - Interpretation of genetic data

Understanding Key Diseases:

- **Sickle cell disease:** An inherited blood disorder caused by a genetic mutation affecting hemoglobin, the oxygen-carrying protein in red blood cells.
- **Thalassemia:** A genetic blood disorder that impairs the body's ability to produce hemoglobin. It's inherited when both parents pass on the faulty gene.

Significance of the Project:

• **Bridging Science and Tradition:** It will serve as a link between scientific advancements and traditional knowledge, aiming for a **healthy and prosperous future** for tribal communities.

- **Identifying Genetic Markers:** The project will help identify genetic markers for:
 - Innate immunity
 - Cancer
 - Hereditary diseases
- Targeted Healthcare: Tribal health profiles will be linked to basic health services for targeted medical support.
- Long-Term Health Improvement: Designed not just as a research project, but as a campaign for long-term health improvement.

What is Genome Sequencing?

Genome sequencing is a process that determines the complete DNA sequence of an organism, including both genes and non-coding regions. It identifies the **precise order** of the nucleotide bases (adenine, cytosine, guanine, thymine) in DNA. Determining this order of bases is what we call **sequencing**.

Global Genome Initiatives: An Extra Edge

These projects represent significant milestones in understanding human genetics and have laid the groundwork for precision medicine and advanced genetic research.

Human Genome Project (HGP)

- Goal: A monumental international scientific research project (1990-2003) aimed at determining the complete sequence of human DNA's base pairs and identifying, mapping, and sequencing all human genes from both a physical and functional standpoint.
- **Scope:** Involved researchers from numerous countries, including the United States, United Kingdom, France, Germany, Japan, and China.

• Impact:

- Provided the **first complete "reference sequence" of the human genome**, a foundational resource for global biomedical research.
- Accelerated advancements in **personalized medicine**, where treatments can be tailored to an individual's genetic makeup.
- Improved diagnostic tools and therapeutic strategies for various diseases (e.g., cancer, cystic fibrosis).
- Spurred significant innovations in biotechnology, including gene-editing technologies like CRISPR-Cas9.
- Set a precedent for international scientific collaboration and established principles of **open access to scientific data**.
- Dramatically reduced the cost of genome sequencing over time.

International HapMap Project

- Goal: Launched in 2002, this project aimed to develop a haplotype map (HapMap) of the human genome. A haplotype is a set of DNA variations (polymorphisms) that tend to be inherited together. The project sought to describe common patterns of human genetic variation across different populations.
- **Scope:** An international collaboration involving researchers from Canada, China, Japan, Nigeria, the United Kingdom, and the United States.

• Impact:

- Created a public, genome-wide database of common human sequence variations.
- Provided a catalog of how genetic variants are distributed among diverse populations, crucial for understanding population-specific disease susceptibilities and drug responses.
- Enabled researchers to more efficiently identify genetic variants associated with common diseases (e.g., diabetes, cancer, heart disease) and individual responses to medications and environmental factors.
- Facilitated **genome-wide association studies (GWAS)** by providing a shortcut to analyze genetic variation without sequencing every single base pair.

100,000 Genomes Project (UK)

- Goal: A UK government project, managed by Genomics England, launched in 2012. Its aim was to sequence 100,000 whole genomes from National Health Service (NHS) patients, primarily focusing on rare diseases and common types of cancer, as well as infectious diseases.
- **Scope:** Involved NHS patients and their relatives, with recruitment and sample collection coordinated through 13 NHS Genomic Medicine Centres across England.

• Impact:

- Integrated genomic medicine services into the NHS, making genomic testing more accessible for patients.
- Provided **new diagnoses for many patients with rare diseases** (around one in four participants with a rare disease received a diagnosis for the first time).
- Generated clinically actionable results for a significant portion of cancer patients, leading to opportunities for targeted therapies or clinical trial entry.
- Created a rich dataset that continues to be used by researchers globally to improve understanding of disease causes and develop new treatments, diagnostics, and medicines.
- Served as a model for large-scale genomic initiatives and highlighted the importance of linking genomic data with medical records for comprehensive analysis.

 $Source: \underline{https://www.thehindu.com/sci-tech/science/gujarat-launches-indias-first-tribal-genome-project-to-tackle-inherited-diseases/article69819673.ece$