# Genome study: 180 million genetic variants found in 9,772 individuals

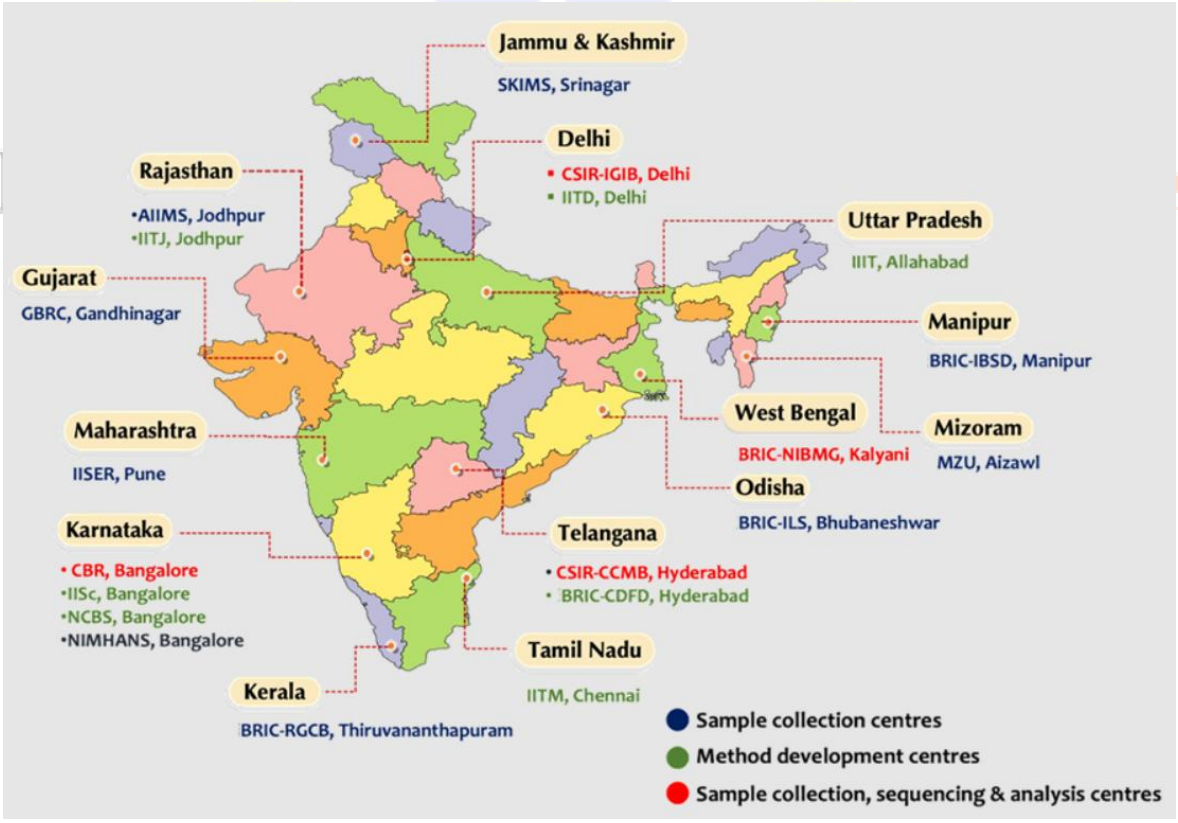
**Context: GenomeIndia Project**

**GenomeIndia** is a pioneering scientific project funded by the **Department of Biotechnology**, Ministry of Science and Technology, Government of India.

- **GenomeIndia** is a government-funded pan-India initiative to catalogue the **genetic diversity of the Indian population**.
- It aims to build a **comprehensive database** of Indian genomes to enhance understanding of:
  - Genetic diseases
  - Drug response variation
  - Population structure
  - Precision medicine potential

**Who is involved?**

The working consortium is represented by 20 national institutes across the country.



## Key Findings of the Preliminary Study

- **Sample Size:**
  - **20,000** individuals enrolled.
  - **10,074** DNA samples sequenced (from 85 diverse populations: 32 tribal + 53 non-tribal).
  - Final analysis on **9,772 individuals (4,696 male + 5,076 female).**
- **Genome Diversity:**
  - **180 million genetic variants identified.**
  - Variants include:
    - Rare variants
    - Community-specific variants
    - India-unique variants
    - Disease-associated variants
    - Variants influencing therapeutic response or resistance.
- **Populations covered:**
  - **Tribal groups**: Tibeto-Burman, Indo-European, Dravidian, Austro-Asiatic.
  - **Non-tribal groups**: Same linguistic categories.
  - **Admixed outgroup** included for comparative insights.
- **Data Storage:**
  - Genome sequence data housed at **Indian Biological Data Centre**, Faridabad, Haryana.

## Scientific & Medical Significance

- **Precision Medicine:**
  - Enables **personalised treatment** based on genetic makeup.
  - Identifies people at risk for specific diseases → **early interventions**.
- **Disease Detection:**
  - Facilitates development of **low-cost diagnostics**.
  - Improves detection of **hereditary conditions** (e.g., thalassemia, cancer predisposition).
- **Drug Response:**
  - Predicts how different groups respond or react adversely to medications.

  - Enhances **pharmacogenomics** (customized drug treatment plans).
- **Public Health:**
  - Informs targeted vaccination and therapy strategies in epidemiology.
  - Can reduce **trial-and-error** treatment, especially in chronic and rare diseases.

**Scientific Methodology & Rigour**

- **Whole Genome Sequencing (WGS)** used — gives high-resolution genetic insights.
- Median sample:
  - 159 per non-tribal group, 75 per tribal group.
  - Sufficient for identifying rare, population-specific variants.
- Integrates genomic data with **biochemistry & anthropometric parameters** — unique approach in India.

**Way Forward**

- Upcoming **detailed paper** to give deeper insight into disease linkages.
- Development of **India-specific genomic risk scores.**
- **Public-private collaborations** for translating genetic insights into affordable diagnostics.
- Integration into **Ayushman Bharat Digital Mission or National Health Stack.**