Genetic Diversity in the Indian Population

For Prelims: Genetic Diversity in the Indian Population, DNA (Deoxyribonucleic Acid)**, Endogamous practices,** Whole-Genome Sequencing.

For Mains: Genetic Diversity in the Indian Population.

Source: TH

Why in News?

Recently, a study by Institute for Human Genetics, University of California, has found stark genetic differences between **people from different regions of the Indian subcontinent.**

What is the Methodology of the Study?

- The researchers collected **DNA (Deoxyribonucleic Acid)** from around 5,000 individuals, mainly people from across India, Pakistan, and Bangladesh. This cohort also contained DNA from some Malay, Tibetan, and other South-Asian communities.
- They performed **Whole-Genome Sequencing** **to identify all the instances** where the DNA either showed a change, was missing, or had additional base-pairs, or 'letters'.

What are the Key Findings of the Study?

- **Endogamous Practices:**
  - There is **little mixing between individuals from different communities in the Indian subcontinent.**
  - Endogamous practices, such as caste-based, region-based, and **consanguineous (Closed Relatives) marriages**, contributed to conserved **genetic patterns** at the community level.
    - In an ideal scenario, there would have been random mating in a population, leading to greater genetic diversity and lower frequency of variants, which are linked to disorders.
- **Regional Trend:**
  - Compared to a relatively outbred population, like that of **Taiwan, the South Asian cohort** – and within it, the South-Indian and Pakistani subgroups – showed a **higher frequency of homozygous genotypes, possibly due to cultural factors.**
    - Humans typically have **two copies of each gene.** When an individual has two copies of the same variant, it is called a Homozygous Genotype.
    - Most genetic variants linked **to major disorders are recessive in nature** and exert their effect only when present in two copies. (Having different variants – i.e. being heterozygous – is usually protective.)
  - The South-Indian and **Pakistani subgroups were estimated to have a high degree of inbreeding** while the Bengali subgroup showed **significantly lower inbreeding.**
    - Not only did the South Asian cohort have a higher number of variants that could

disrupt the functioning of genes, there were also **unique variants that were not found in European individuals.**

- **Risk of Higher Frequency of Homozygous Variants :**
    - The presence of rare homozygous variants increased the risk of disorders like **cardiovascular diseases, diabetes**, **cancers**, and mental disorders.

What are the Other Studies on Genetic DIversity?

- In 2009, a study in Nature Genetics by the group of Kumarasamy Thangaraj, at the Centre for Cellular and Molecular Biology, Hyderabad, revealed that a small group of **Indians are prone to cardiac failure** at relatively young ages.
- The DNA of such individuals **lacked 25 base-pairs in a gene crucial for the rhythmic beating** of the heart (scientists call it a 25-base-pair deletion).
- This deletion was **unique to the Indian population** and, barring a few groups in Southeast Asia, was not found elsewhere.
- This **deletion arose around 30,000 years ago**, shortly after people began settling in the subcontinent, and **affects roughly 4% of the Indian population** today.
    - Identifying such genetic novelties **helps understand population-specific health risks** and vulnerabilities.

What is the Significance of Such Studies on Genetic Diversity?

- Studies have shown that specific genetic novelties are **linked to the health of India's populace**. Understanding these genetic variations **can lead to better interventions for major health concerns.**
- Conducting genetic studies within the country can protect **vulnerable communities from potential exploitation** by multinational companies and foreign research organizations.

What is the Importance of a Detailed Map of the Indian Genome?

- India's incredible diversity necessitates **a detailed** Map of the Indian Genome for various reasons, including economic, matrimonial, and geographical factors.
- Such a map can aid in **understanding the genetic basis of health disparities** and guide population health interventions.

UPSC Civil Services Examination, Previous Year Questions (PYQs)

Q1. With reference to agriculture in India, how can the technique of 'genome sequencing', often seen in the news, be used in the immediate future? (2017)

1. Genome sequencing can be used to identify genetic markers for disease resistance and drought tolerance in various crop plants.
2. This technique helps in reducing the time required to develop new varieties of crop plants.
3. It can be used to decipher the host-pathogen relationships in crops.

Select the correct answer using the code given below:

(a) 1 only
(b) 2 and 3 only
(c) 1 and 3 only
(d) 1, 2 and 3

Ans: (d)