Endogamy may be the cause of the Persistence of harmful genetic variants in India

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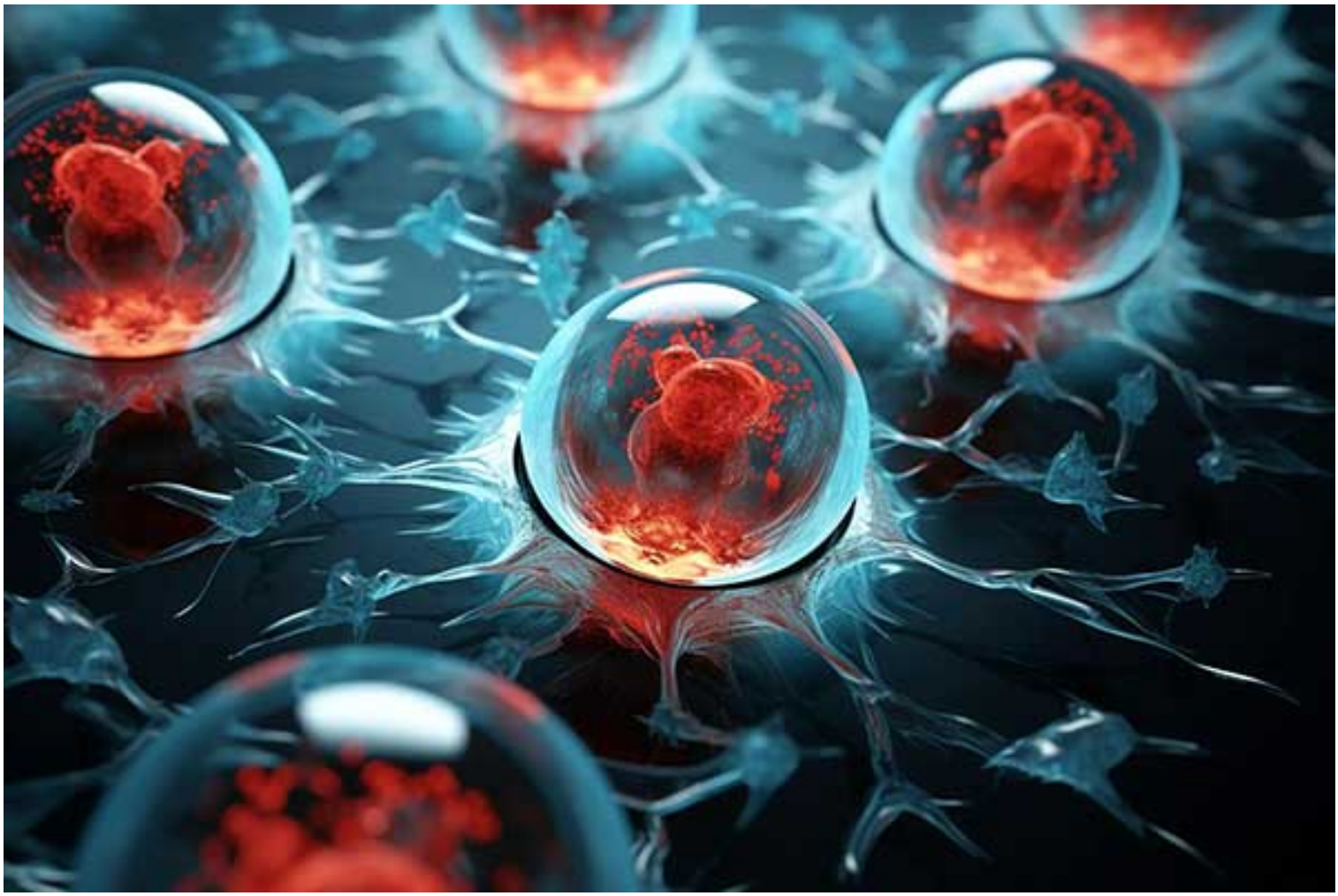

Endogamy may be the cause of the Persistence of harmful genetic variants in India

A study by **Centre for Cellular and Molecular Biology**, Hyderabad has recently found causes of **cardiac failure** at **younger ages** in the **Indian** population.

- **Deoxyribonucleic acid** (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**).

[Ref – Pharmaceutical Technology]

About 25 base pair deletions:

- 25 base pair deletion is a risk **allele** for late-onset **left ventricular (LV) dysfunction**, hypertrophy, and **heart failure.**
- A 25-base pair (25bp) deletion in the **MYBPC3 gene** is enriched in **South Asians,** being **unique** to the **Indian** and **Southeast Asian** population and **not** found elsewhere.
- This affects about **4%** of the **Indian population**.

Key findings of the study:

Genetic differences among populations:

- Whole-genome sequencing of individuals from India, Pakistan, and Bangladesh found **genetic differences** between people from different regions of the **subcontinent**.
- These genetic differences were even found at the level of **smaller geographies** within **India**.
- There was **little mixing** between individuals from **different communities**.
- **Endogamous practices** (including caste-based, region-based, and consanguineous marriages) in the subcontinent are responsible for such **conserved genetic patterns** at the community level.
- In ideal conditions, there would have been **random mating** in a **population**, leading to **greater genetic diversity** and **lower** frequency of **variants** linked to **disorders**.
- The **cultural aspects** of Indians might need mending to improve the population's health.

Homozygous genotypes:

- The South Indian and Pakistani sub-group showed a **higher frequency** of **homozygous genotypes.**
- 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 show their effect

only when present in **two copies**.
- **Heterozygous individuals** are at **lower** risk of getting affected by **genetic** disorders.
- The main cause of **homozygous** genetic composition is **inbreeding** or **consanguineous marriages** which is prevalent in South India and Pakistan.
- The South Asian cohort has a **higher number** of **variants** that could **disrupt** the **functioning of genes**, but there were also **unique variants** that were **not** found in **European** individuals.
    - These variants affect many **physiological parameters**, leading to a higher risk of cardiovascular disorders, diabetes, cancers, and mental disorders.

Indian genome mapping:

- **Human Genome Sequencing** was completed in **2003**.
- **African** and **Chinese** population gene sequencing has been done.
- As **India** has a **diverse population**, there is a **need for genome sequencing** of the Indian population for economic, matrimonial, and geographical reasons.
- The Genome India project has been launched in **2020** to sequence **10,000** Indian human genomes.
- The idea of **genetic puritanism** must be taken away to prevent major **hereditary disorders**.

About Human Genome Sequencing:

- The **Human Genome Project (HGP)** was an international scientific research project for determining the **base pairs** that make up **human DNA**, and identifying, mapping, and sequencing all of the genes of the human genome.
- It was **started in 1990** and was **completed** in **2003**.
- The human genome has approximately **3.1 billion base pairs**.
- There are approximately **22,300 protein-coding genes** in human beings.

Significance of the study:

- The study of sequenced human genes was helpful to **identify** the **genetic variants** that **increase** the **risk** for common diseases like cancer and diabetes.
- The study has shown that **identifying unique genetic variants** can help develop **interventions** for major health concerns.

