Human Pangenome Map

For Prelims: Human Pangenome Map, Genome**,** DNA**, Genes, Reference Genome,** Genome India Project**.**

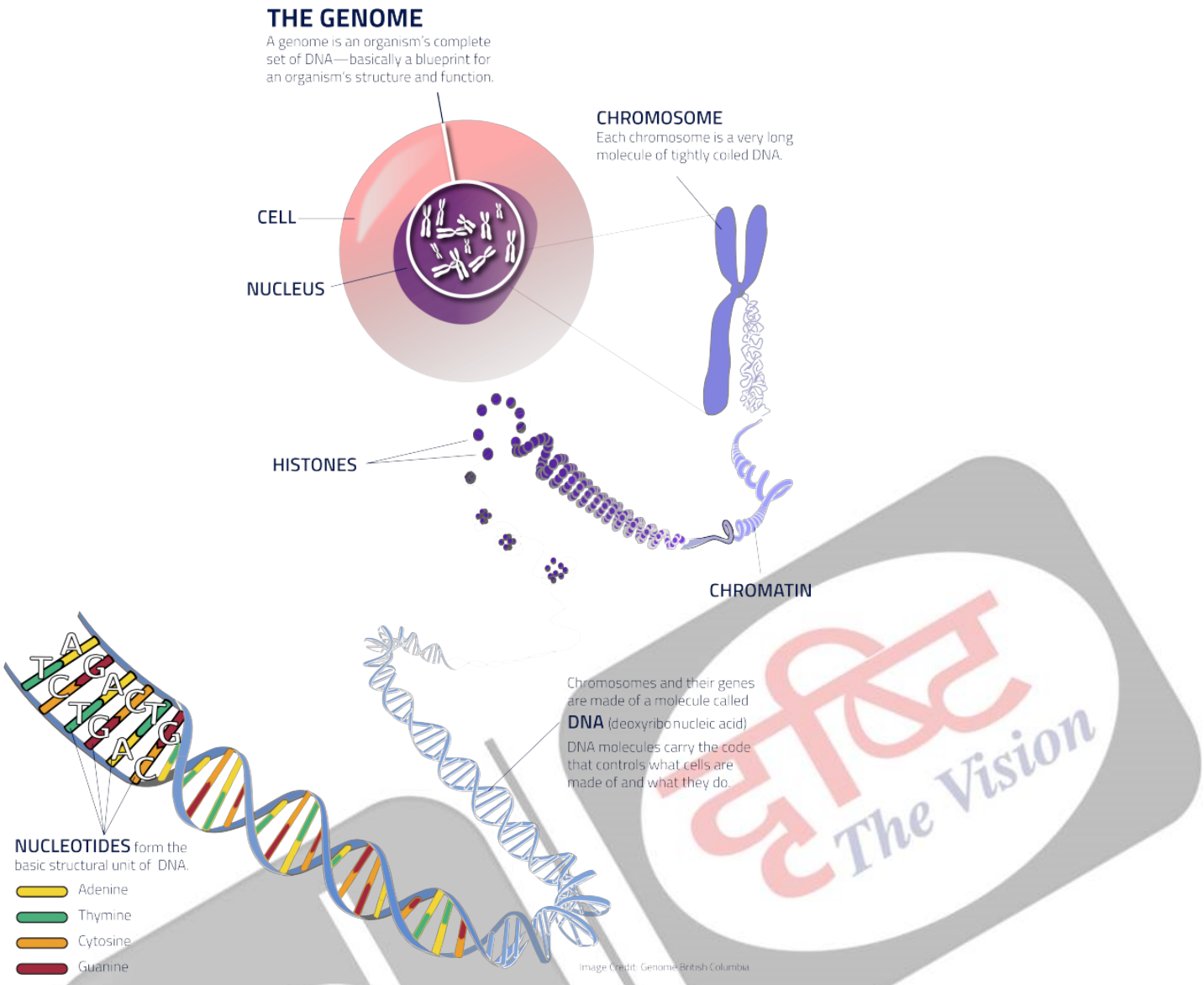
For Mains: Human Pangenome Map and its Significance.

Why in News?

Recently, a new study has been published in the Nature journal describing a **Pangenome Reference Map,** built using genomes from 47 anonymous individuals (19 men and 28 women), mainly from Africa but also from the Caribbean, Americas, East Asia, and Europe.

What is Genome?

- **Genome:**
  - The **Genome** is like a blueprint or instruction manual for life. It contains all the **genes and the spaces between the genes that make up our chromosomes.**
  - Our chromosomes are made up of **DNA (Deoxyribonucleic Acid)**, which is a long string composed of four **building blocks called nucleotides or bases (A, T, G, and C).** These building blocks are arranged in different combinations and repeated millions of times to create **23 pairs of chromosomes.**
  - The genome provides **valuable information about our genetic makeup** and helps researchers explore various aspects of human biology and health.
- **Genome sequencing:**
  - Genome sequencing is the method used to **determine the precise order of the four bases (A, T, G and C)** and how they are arranged in chromosomes.
  - By sequencing individual genomes, scientists can **learn about human genetic diversity and understand how certain diseases** may affect us.



What is a Reference Genome?

- **About:**
  - A reference genome or reference map is like a standard map that scientists use when they sequence and study new genomes. It serves as a guide to **compare and understand the differences between the newly sequenced** genome and the reference genome.
- **Significance:**
  - The first reference genome created in **2001 was a significant scientific achievement.** It helped scientists discover disease-related genes, understand genetic aspects of diseases like cancer, and develop new diagnostic tests. However, it had limitations and wasn't perfect.
- **Drawback:**
  - It was mostly based on the genome of one individual with **mixed African and European ancestry**, and it had some gaps and errors.
  - While the new **reference genome or Pangenome** is comprehensive and error-free, it still doesn't represent the full diversity of human genetics.

What is a Pangenome Map?

- **Pangenome Map:**

    - The pangenome, unlike the previous linear reference genome, is represented as **a graph.** Each chromosome in the pangenome **can be imagined as a bamboo stem with nodes.**
    - These nodes represent stretches of sequences that are **similar among all 47 individuals.** The internodes between the nodes vary in length and represent genetic variations among individuals from different ancestries.
    - To create complete and continuous maps of the chromosomes in the pangenome project, researchers used a technology called **long-read DNA sequencing, creating complete and continuous chromosome maps by producing accurate, long DNA strands.**
- **Significance of Pangenome Map:**
    - Although any two humans share more than 99% of their DNA, there is **still about a 0.4% difference between any two individuals.** This may seem small but considering the vast size of the human genome (3.2 billion nucleotides), the difference **amounts to around 12.8 million nucleotides.**
    - A complete and accurate pangenome map of the human genome can help better understand these **differences and explain the diversity among individuals.**
    - It will also assist in studying genetic **variations that contribute to underlying health conditions.**
    - Although the current map doesn't include genomes from Indians, it will still be beneficial in **comparing and mapping Indian genomes against the existing accurate reference genomes.**
    - Future pangenome maps that include high-quality Indian genomes, including those from diverse and isolated populations within the country, **will provide valuable insights into disease prevalence,** the discovery of new genes related to rare diseases, improved diagnostic methods, and the development of novel drugs for these diseases.
- **Drawbacks:**
    - However, the current pangenome map lacks representation from diverse populations like **Africa, the Indian subcontinent, indigenous groups in Asia** and Oceania, and West Asian regions.

Is there any Genome Mapping Initiative in India?

- In April 2023, the government announced that it aims to sequence 10,000 genomes by the end of the year 2023 under the **Genome India Project (GIP).**
- The GIP aims to create a database of Indian genomes, researchers can learn about these unique **genetic variants and use the information to create personalized drugs** and therapies.
    - The United Kingdom, China, and the United States are among the countries that have programmes to sequence at least 1,00,000 of their genomes.

UPSC Civil Services Examination, Previous Year Question (PYQ)

Q. 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)

- Chinese scientists decoded rice genome in 2002. The Indian Agricultural Research Institute (IARI) scientists used the genome sequencing to develop better varieties of rice such as Pusa Basmati-1

and Pusa Basmati-1121, which currently makes up substantially in India's rice export. Several transgenic varieties have also been developed, including insect resistant cotton, herbicide tolerant soybean, and virus resistant papaya. **Hence, 1 is correct.**

- In conventional breeding, plant breeders scrutinize their fields and search for individual plants that exhibit desirable traits. These traits arise spontaneously through a process called mutation, but the natural rate of mutation is very slow and unreliable to produce all the plant traits that breeders would like to see. However, in genome sequencing it takes less time, thus it is more preferable. **Hence, 2 is correct.**
- The host-pathogen interaction is defined as how microbes or viruses sustain themselves within host organisms on a molecular, cellular, organism or population level. The genome sequencing enables the study of the entire DNA sequence of a crop, thus it aids in understanding of pathogens' survival or breeding zone. **Hence, 3 is correct.**
- **Therefore, option (d) is the correct answer.**

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