# The Complete Human Genome

## Why in news?

Scientists published the first complete human genome, offering new promise in the search for clues regarding disease-causing mutations and genetic variation.

## What is human genome?

- A genome is defined as an organism's complete set of Deoxyribose Nucleic Acid (DNA), including all of its genes.
- It consists of all of the approximately three billion base pairs of deoxyribonucleic acid (DNA) that make up the entire set of chromosomes of the humans.
- **Non-uniformity**- Human genome is mostly the same in all people, but a very small part of the DNA does vary between one individual and another.
- Excepting identical (monozygous) twins, no two humans on Earth share exactly the same genomic sequence.
- **Non-static**- It is susceptible to certain changes.

## How did the mapping of human genome evolve?

- **Human Genome Project-** The HGP was the international, collaborative research program for complete mapping and understanding of all the genes of human beings.
- It began in 1990 and was completed in 2003.
- It contained information from a region of the human genome known as the euchromatin where the chromosome is rich in genes and the DNA encodes for protein.
- The 8% was left out and was called heterochromatin and it does not produce protein.
- Heterochromatin was given lower priority because it was thought to be "junk DNA" while euchromatin contained more genes that were simpler to sequence with the tools available at the time.
- Now, a large team has accounted for that 8%, completing the picture of the human genome for the first time.
- The fully sequenced genome is the result of the efforts of a global collaboration called the Telomere-2-Telomere (T2T) project.

## What is the significance of this mapping?

- Knowledge of the human genome provides an understanding of
    - the origin of the human species
    - the relationships between subpopulations of humans
    - the health tendencies or disease risks of individual humans
- The new reference genome, called T2T-CHM13, includes highly repetitive DNA sequences found in and around the telomeres (structures at the ends of chromosomes) and the centromeres (at the middle section of each chromosome).
- The new sequence is known to play important roles in evolution and disease.

- A complete human genome makes it easier to study genetic variation between individuals or between populations.
- Constructing a complete human genome can serve as a reference to understand which variations might be responsible for disease.
- The new T2T reference genome will complement the standard human reference genome, known as Genome Reference Consortium build 38 (GRCh38), which originated from the Human Genome Project.

## What were the earlier projects regarding genome mapping?

- **Genome India Project**- It was spearheaded by Centre for Brain Research.
- It aims to build a grid of Indian reference genome to understand the type and nature of diseases and traits that comprise the diverse Indian population.
- **International HapMap Project**- It is a partnership of scientists and funding agencies from Canada, China, Japan, Nigeria, the United Kingdom and the United States.
- The goal is to determine the common patterns of DNA sequence variation in the human genome and to make this information freely available in the public domain.
- **1000 Genomes Project**- It began in 2008 and created a catalogue of common human genetic variation, using openly consented samples from people who declared themselves to be healthy.
- The International Genome Sample Resource (IGSR) maintains and shares the human genetic variation resources built by the 1000 Genomes Project.

**References**


1. https://indianexpress.com/article/explained/complete-human-genome-explained-7848706/
2. https://www.britannica.com/science/human-genome/Social-impacts-of-human-genome-research
3. https://www.internationalgenome.org/
4. https://www.nature.com/articles/nature02168
5. https://www.genome.gov/human-genome-project/What