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

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