## Whole Human genome mapped

April 4, 2022

<u>In news-</u> Recently, the scientists have published the first complete human genome, filling in gaps remaining after previous efforts.

Key updates-

- The current mapping offers new promise in the search for clues regarding disease-causing mutations and genetic variation among the world's 7.9 billion people.
- Researchers in 2003 unveiled what was then billed as the complete sequence of the human genome. But about 8% of it had not been fully deciphered, mainly because it consisted of highly repetitive chunks of DNA that were difficult to mesh with the rest.
- Addressing the remaining 8 per cent of the genome, the Telomere-to-Telomere (T2T) Consortium has now presented a complete 3.055 billion base pair sequence.
- The full sequencing builds on the work of the Human Genome Project, which mapped about 92% of the genome.
- Researchers said that the last eight per cent includes numerous genes and repetitive DNA and is comparable in size to an entire chromosome.
- They generated the complete genome sequence using a human cell line with only one copy of each chromosome, unlike most human cells, which carry two copies of each chromosome.
- Each strand of the human DNA is made up of a pair of chromosomes, drawing one each from the parents.
- The Telomere to Telomere (T2T) consortium's full version is composed of 3.055 billion base pairs, the units from which chromosomes and our genes are built and 19,969 genes that encode proteins.
- The finding provided new insights into the workings of the DNA, how it influences the risks of disease and how

cells keep it neatly organized.

- Decoding the human genome opens up new avenues of research to enhance human health and find new ways to treat diseases and identify genetic diversities behind them.
- With the full genome sequence now available, researchers can create a more accurate map of chromosomes, and thus, of the DNA.
- They can **find answers to basic biological questions** about how chromosomes properly segregate and divide.
- Such insights are vital for understanding the genetic contributions to certain diseases, and the DNA's response during fighting a disease, including a new one.

What is a genome?

- A genome is a chemical compound that represents an organism's complete set of DNA.
- It contains all the information of the past evolution and codes for instructions needed to develop and direct activities to run the life of the organism.
- A set of DNA is made of two twisting paired strands, often referred to as a double helix.
- A unit of DNA containing a particular set of attributes and information is called gene, the word more commonly used by people to refer to hereditary matters.
- Whole genome sequencing (WGS), also known as full genome sequencing, complete genome sequencing, is the process of determining the entirety, or nearly the entirety of the DNA sequence of an organism's genome at a single time.
- Each DNA strand is made of four chemical substances, called nucleotide bases that carry genetic information.
- Each base is represented by one of the four letters A,
   T, C and G and they are arranged in pairs.
- Virtually every single cell in the body contains a

complete copy of the approximately 3 billion DNA base pairs, or letters, that make up the human genome.

•With its four-letter language, DNA contains the information needed to build the entire human body.

## Further

## reading:

https://journalsofindia.com/end-to-end-genome-sequencing-telom
ere-to-telomere-consortium/