

Sequencing the Y Chromosome

Why in news?

Scientists have fully sequenced the Y chromosome for the first time, uncovering information that could have implications for the study of male infertility and other health problems.

What is Y chromosome?

DNA is a molecule that carries genetic information for the development and functioning of an organism.

- In the nucleus of a human cell, each DNA molecule is packaged into a long thread like structure called chromosome.
- Most human cells contains 23 pairs of chromosomes. One half of each pair of chromosomes from one parent, while other half comes from other parent.
- The 23rd pair are X and Y chromosomes, often called as **sex chromosomes**. The other 22 pairs called as **autosomes**.
- Females have a pair of X chromosomes, whereas males have X and Y chromosome.
- The Y chromosome is male-determining because it bears a gene called **SRY**, which directs the development of a ridge of cells into a testis in the embryo.
- The embryonic testes make male hormones, and these hormones direct the development of male features in a baby boy.



What is the difficulty in sequencing Y chromosome?

- **Repetition** - The Y chromosome was a particularly hard nut to crack because it is unusually repetitive.
- While all human chromosomes contain repeats, more than 30 million letters of the Y chromosome — out of 62.5 million — are repetitive sequences, sometimes called **satellite DNA or junk DNA**.
- Repetitive DNA complicates the assembling of data from genetic sequencing.
- **Palindromes** - The Y chromosome also contains palindromes — sequences of letters that are the same backward and forward, like radar.
- **Degeneration of Proto- Y** - The proto-Y is degenerating at a faster pace, losing about 10 active genes per million years, reducing the number from its original 1,000 to just 27.
- There has been great debate about whether this degradation continues, because at this rate the whole human Y would disappear in a few million years

The Y is the last human chromosome to have been sequenced end-to-end, or telomere to telomere (T2T)

How the scientists unravelled the complex Y chromosome?

- **Sequencing** - Advanced "*long-read*" *sequencing technology* and computational methods enabled researchers to achieve a complete reading of the Y chromosome.
- This accomplishment added over 30 million repetitive base pairs to the human reference genome.
- The new technology has allowed sequencing of bases along individual long DNA molecules, producing long-reads of thousands of bases.
- It effectively dealt with repetitive sequences and transformed raw sequencing data into a usable resource.
- These longer reads are easier to distinguish and can therefore be assembled more easily.
- **Findings**- Overall, the combined research determined that the Y chromosome has 106 protein-coding genes.
- 42 were found that were new, but many still appear to be repeats.

What is the importance of the study?

- **Advanced diagnostics**- The study empowers future sequencing endeavours to explore into health and disease aspects through comprehensive Y chromosome inclusion.
- To study whether loss of the Y chromosome is a biomarker of biological aging or has a direct effect on the health of men.
- **Infertility**- It will help to study conditions and disorders linked to the chromosome, such as lack of sperm production that leads to infertility.
- **Health**- Genes have been identified on the Y chromosomes that have been shown to be required for the prevention of cancer and cardiovascular disease.
- **Dark matter**- It represents the 'dark matter' of the genome. This analysis will allow us to better understand the regions of the Y chromosome that have regulatory functions and may encode mRNA and proteins.
- **Human evolution**- Assembling complete sequences of Y chromosomes across space and time not only helps to investigate sex chromosome evolution but also human evolution.
- **Gene therapy**- It will open up avenues to treat diseases that may linked to Y chromosomes.
- **Future studies**- The findings provide a solid base to explore how genes for sex and sperm work, how the Y chromosome evolved, and whether as predicted will disappear in a few million years.

References

1. [The Hindu- Gene Sequencing of Male Y chromosome](#)

2. [Live Science- Full sequencing of Male Y chromosome](#)

