

# **Newborn Genome Sequencing**

## Why in news?

Newborn genome sequencing unlocks the blueprint of health.

#### What is genome sequencing?

- 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 DNA that make up the entire set of chromosomes of the humans.
- Genome Sequencing is the state-of-art, robust and high throughput technique to <u>sequence the entire genome of an organism</u>.

To know about the Complete Human Genome, click <u>here</u>



## What methods are deployed for Genome sequencing?

- **Clone-by-Clone method-** It is a *traditional method* that requires high density genome map and works well for larger genomes like eukaryotic genomes.
- Whole genome shotgun sequencing- It is the *improved version of Clone-by-Clone* <u>method</u> that doesn't require a genome map,

- It is the faster method of sequencing but not suitable for larger genomes as they have a number of repetitive DNA sequences.
- Next-Generation Sequencing- It is the *most advanced, robust*, accurate, faster, cheaper and high throughput genome sequencing technique.
- It relies on the chemistry of bridge amplification and can sequence more than 5 separate human genomes simultaneously.
- It is the most trusted Genome Sequencing method so far.

#### Why is it important to sequence new born genome?

- New born genome sequencing helps in *early diagnosis of the disease* that would lead to effective treatment and *prevent from death/disability* of child.
- The rapid diagnosis would consequently translate into a significant  $\underline{reduction \ in \ the}$  $\underline{cost}$  of treatments.
- Population-scale genome-sequencing efforts have provided insights into the *prevalence* <u>of many of the diseases</u> in an unbiased manner.
- The U.K. National Health Services has recently launched a nationwide programme to sequence 100,000 sick newborns.
- It helps in *predictive diagnosis and precision medicine*.
- It identifies *potential causative variants* for further follow-up studies for gene expression and regulating mechanism.

There are 6000 genetic disorder diseases of which 3500 are documented and only few of them have been properly mapped.

## What are the ethical challenges?

- **Privacy issues** The issue of disclosing and managing incidental and secondary findings.
- **Psychological impact on families** Stereotyping and stigmatization of the participants inducted in the research.
- **Issues of justice** Equitable distribution of benefits and burdens associated with accessing and utilizing this technology.

## What lies ahead?

- Since, it is the solution to the rare genetic disorder, it provides for better healthier future.
- Strict adherence to the ethical measures would help to maintain data security and privacy.

## **Quick facts**

- Human Genome project- It is an international research collaboration between 1990 and 2003 with the primary goal to
  - $\circ\,$  Decipher the entire human genetic material.
  - $\circ\,$  Sequence the whole genome at 3 billion base pair per second.

- Improve the tool for data analysis and create a physical map of human genome.
- **Genome India project-** It is a whole genome sequencing of 10,000 individuals representing country's diverse populations.
- The vision is "Cataloguing the genetic variations in Indians" for 3 years (2020-2023).
- IndiGen project- The Council of Scientific & Industrial Research (CSIR) has conducted Whole Genome Sequencing of 1,008 Indians from different populations across the country.

#### References

- 1. The Hindu| New born Genome Sequencing
- 2. Genetic Education | What is Genome Sequencing
- 3. PIB IndiGen Project
- 4. PIB Genome India Project

