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 *sequence the entire genome of an organism*.

To know about the Complete Human Genome, click here



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 method* 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 *reduction in the cost* of treatments.
- Population-scale genome-sequencing efforts have provided insights into the *prevalence of many of the diseases* 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
  - Decipher the entire human genetic material.
  - 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

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