GENOME SEQUENCING - SCI & TECH GS III MAINS

Q. Genome sequencing has a scope to revolutionize every sector starting from pharmaceuticals to agriculture. Discuss (10 marks, 150 words)

News: *The transformative benefits of population-level genome sequencing | Explained*

What's in the news?

- In the last decade, genomics has undergone a revolutionary shift with the advent of technologies that have significantly improved through and reduced the cost of whole-genome sequencing, giving rise to population-scale genome-sequencing programmes – where scientists decipher the complete genetic makeup of large populations, offering unprecedented insights into the intricate view and tapestry of human diversity.

Genome Sequencing:

- Genome sequencing is a process of determining the complete DNA sequence of an organism's genome.
- Breakthrough in human genome sequencing has potential to benefit mankind especially in healthcare, disease prevention etc.

Applications of Genome Sequencing:

1. Biological research:

- The ability to read genetic sequences is extremely useful in biological research because the base sequence contains information for making proteins as well as regulating gene functions.

2. Forensics:

- Sequencing has proven to be a powerful tool in forensics. Because differences in DNA and RNA sequences can differentiate organisms down to species and individual levels, it can help to classify diseases, identify therapeutic targets and customize treatments.

3. Diagnostics:

- **Pre-natal screening:** It has also been used in prenatal screening to determine whether the foetus has any genetic disorders or anomalies.
- **Evaluate disorders:** Genome sequencing has been used to assess rare disorders, preconditions for disorders and even cancer from a genetic viewpoint, rather than as diseases of specific organs.
- **Drug Efficacy:** Genome sequencing can provide information about drug efficacy or adverse drug effects.

The relationship between drugs and the genome is called pharmacogenomics.

4. Vaccine development:

- Sequencing of viruses (e.g. ebola, coronavirus) and bacteria has led to the development of vaccines against them, once knowing their variants or strains.
- Genomic data of pathogens could reveal hidden pathways of transmission.

5. Population Studies:

- Advanced analytics and AI could be applied to critical datasets generated by collecting genomic profiles across the population, allowing for a better understanding of disease causation and potential treatments.
- This is especially relevant for rare genetic diseases, where large datasets are required to find statistically significant correlations.

6. Agriculture and food security:

- Genome sequencing has the power to revolutionize food security and sustainable agriculture by reducing the risks from disease outbreaks and improving agriculture through effective plant and animal breeding, detecting multiple pathogens, etc.

Limitations of Genome Sequencing:

1. Data Analysis:

- A vast amount of data is generated, which requires extensive analysis and interpretation.

2. Structural variants:

- While technologies used to sequence DNA are highly accurate at deciphering the sequence, the majority of available technologies have limited scope in being able to determine so called structural variants.
- These are alterations that affect large segments of DNA at a time, such as duplications, deletions, and inversions.

3. Incomplete research and irrelevant data:

- Despite growing knowledge in genomics, many genes still have unknown roles and a large number of genomic variants have not been identified as benign or pathogenic.

4. Ethical concerns:

- Storing the large amount of data generated by WGS poses challenges related to capacity, cost, and privacy concerns, including potential ethical dilemmas with insurance companies and family members.

5. Not suitable for larger genomes:

- Despite being a faster method, whole genome sequencing is not suitable for larger genomes because they have a number of repetitive DNA sequences for which assembling processes is sometimes challenging.

WAY FORWARD:

1. Training:

- It is important to train more clinicians for gene data interpretation and rope in more labs for sequencing. Training more physicians to study medical genetics for speedier analysis is needed.

2. Data security:

- Given the sensitivity of genomic data, every effort must be made to minimise the likelihood of data breaches and to maintain public trust in institutions that gather, store and use such data.
- A practical and feasible solution to build such a reliable and safe database is the application of blockchain technology to secure genomic data.

3. Effective policy:

- There is a need for a comprehensive and effective policy to guide the use of genomic information, with significant emphasis on protecting the privacy of research subjects.
- One way to make this possible is through 'dynamic consent' by which people who wish to participate in a research project can register themselves and provide consent on an ongoing basis.

4. Collaborative effort:

- A collaborative and harmonised effort is needed to balance sharing of genomic data with an individuals' privacy.
- A framework must be designed transparent enough to specify the purpose of the collected genomic data and the duration for which it will be stored in the database.