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EDITORIAL

Warm greetings to all our readers!

It is the time for monsoons and after the trauma of last years floods, we hope that this season, the elements will be merciful to God's own Country!

Here is a Little Insight into Whole Human Genome Sequencing

The human genome is a "biological blueprint" that reflects our genetic makeup and comprises of more than 3 billion base pairs sequentially structured into double helical DNA strands within every cell of our body. This genetic coding carries information crucial for our existence, and variations mark the "uniqueness" in every individual and among racial populations. A clear knowledge about human genome and its variations is essential to study disease processes that can contribute to improved treatment and preventive measures. Hence, sequencing the whole genome is important.

Whole genome sequencing (WGS) today has opened up wide possibilities in the field of medicine and research.

- Millions are suffering from rare undiagnosed diseases either due to complex genetic heterogeneity and/or unusual clinical presentations. In such cases, WGS is a promising and affordable test which can circumvent multiple specialist visits, laboratory and imaging studies and surgical procedures.
- Genome sequencing could become routine in newborn screening programs which can hasten early diagnosis and allow timely interventions to improve the quality of children's lives. In addition, prenatal and first degree relative screenings can be performed to estimate risk of inheriting diseases to offspring, carrier status, and for family planning.
- As a part of personalized medicine, WGS can help analyze patient's genetic variations to guide selection of safer drugs and treatment protocols that reduce side effects and ensure better outcomes.
- WGS is mostly used for cancer research and application of genomics for treatment is usually limited to targeted sequencing approaches. Nowadays, it is helpful in characterization and monitoring tumors and aids in designing specific treatment.
- In forensics, newer genome sequencing approaches are highly beneficial for DNA typing in mass disasters and crime scenes where DNA samples can be of low quantity, degraded or complex mixtures. WGS has potential application in DNA database construction, ancestry and phenotypic inference and distinguishing monozygotic twins.

However, some drawbacks of WGS are hidden costs of interpretation of data, issues with sequencing and analyzing, quality assurance, standardization protocols, ethical dilemmas and difficulties with interpretation of results.

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