Next Generation Sequencing (NGS) Everywhere: "Seq it out" the New Paradigm for Labs

The diagnostic industry in recent times had demonstrated an exponential growth, aiming for more innovative and pioneering efforts to broaden the horizon of precision diagnostics. Witnessing a biotechnological cloudburst boom allowing us to leapfrog from prosaic diagnostics to unfolding the molecular mysteries in genome. The nascency of novel analytical methodologies have empowered to learn the human genetic code sequence. This amplification of DNA served as an "ice breaker" moment for the diagnostic industry to push further the scientific momentum to allow technology to translate preliminary "Sanger sequencing" to accomplish the "Human Genome Project" (HGP).[1] The generational growth enhanced sequencing yield by next generational sequencing (NGS), allowing higher coverage, read length and depth, potentiated further by precise & accurate bioinformatics allowing manageable translation to clinics at desirable scale. Wholesome management systems like Maser (Management and Analysis System for Enormous Reads) has allowed to dig deep to terabytes with more clinical flexibility.[2]

Sequencing techniques whether be the pioneer Sanger technology or the emerging trend towards telomerase to telomerase (T-2T) are surfacing as one the most empowering and seminal game changing biotechnology with potential to supersede most conventional lab methodologies.[3] The conventional histopathology to other pathology science of microbes and other conventions in lab setting are going to be redefined by incorporating multiple types of sequencing techniques for precise and accurate diagnostics. The "good" it provides can be upgraded to next-generational dimensions for pathological sciences and currently happening so in most evolving laboratory setups. Clinical scenario has truly transformed healthcare sciences appear to be targeting almost everywhere and soon the word NGS can be ubiquitous to most clinical laboratories. The conventional newborn screening program shall be replaced by targeted NGS screening thus transforming diagnostic yield and therapeutics.[4] Similarly, the common diseases in society including thalassemia and other genetic diseases can be targeted using advanced sequencing methods from pre-marriage stage, during pregnancy and early as newborn to help prevent and cure with preventive therapies.[5] Why Sequencing formats like NGS and 3rd generational methods adds value to laboratory analytics thus superseding the conventional laboratory? Sequencing methods allow the deeper alterations with DNA code variations which allow can allow physicians to decipher the deep-down pathology whether be the case from oncology, infection and newborn screening or the question related to some forensic requirements. Question marks are raised with regards to inter and intra platform, reproducibility of results and the methodological difficulties but with every passing day the technique is evolving with newer NGS and 3rd generational sequencing methods are emerging with much higher coverage and depth inching further by every day towards refinements with technical robustness in real-time. For example, a case from oncology can be tested to learn resistant genetic mutations and to find gene targets precision and personalized solutions superseding conventional immunohistochemistry biomarkers.[6] Alongside sequencing technologies have also revolutionized the future health prediction and has also been more frequently incorporated in forensic and ancestry linkage analysis. In addition to conventional pathology specimen the genomic markets are witnessing a whole new lot of direct to consumer (DTC) as point of care testing, where the liquid biopsy may emerge as a tool for diagnostics, measure of tumor metastasis, assessment of in utero genetic disorders and infections which may change the way we used to diagnose pathology.[7] Furthermore, the sequencing market is is depicting a paradigm shift with unprecedented demand not just as targeted diagnostics, but also as transcriptomic, next-general sequencing and epigenomic sequenced markers. Provided the phenomenal growth experienced by genomic analytics with explosive rise in sequencing needs displacing traditional diagnostics, growing world economies like Pakistan seems to be facing a retarded growth in this industry as well. Not only Pakistan needs a big thrust and catchment diagnostic market to overcome where it should not only grow self-suffice in its clinical needs but also can emerge stalwart to provide such services to neighbors. Following the simplistic clinical molecular diagnostic needs starting from sequence-based gold-standard diagnostics, it can be anticipated that innovative scientific concepts like genome editing, epigenetics and pharmacogenomics are going to define the future of precision medicine. Foremost remains to appreciate

the application of newer molecular diagnostics at scale with country's need and then to plan small business enterprises with ongoing research and development to simply follow what the models in the west. Examples in this regard remain available.[8, 9]

A strong recommendation for both public and private sector be issued by medical authorities be issued to not only address the biotechnological markets to pace up sequencing methodologies and onward genomic science/molecular laboratories to help the growing medical needs and also to attract healthcare tourism. Disallowing or following a less vigorous approach will neither be beneficial for the sick but also remain an additional burden on the economy. The author humbly believe that sequencing will pioneer the next-generational medicine for precision, ultimate cost, taxonomy of medical literature and the way we believe in science.

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