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EDITORIAL

Genomics in Pakistan

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ABSTRACT

An overview is provided of the globally emerging field of Genomics, highlighting important landmarks and milestones achieved so far. The current status of this promising and futuristic domain of research and clinical application in Pakistan is contrasted against global developments, and recommendations given for purposeful directions so that its immense potential could be exploited for economic and health benefits.

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INTRODUCTION

Genomics is a science that studies the genetic material of all living organisms. It deals with decoding the genome and how it works together within the body of a living organism.[1,2] Genomics also compare nucleotides, identify variants (errors) and classifies genomes.[3] Error(s) in such network of genes can lead to various problems in internal communication and result in genetic diseases.[4] Gene sequencing methods are developed to decode the genes involved in various functions. The detection of errors in genes is helpful in determining various diseases such as cancer and can lead to better and robust treatments.[5] Next Generation Sequencing (NGS) technologies can be effectively deployed to counter genetic diseases.[6] The detection of cancer and other genetic diseases and personalizing its treatment is now possible by using various NGS technologies.[7] Rehman Medical Institute (RMI) located in Peshawar, Khyber Pakhtunkhwa, Pakistan became the first institute in the country to deploy Illumina MiSeq, a cutting-edge machine, to train new scientists, provide diagnostic services to patients, and further explore unknown genetic diseases thereby providing world class treatment.

NGS is a paradigm shift, playing a role in detection, prevention, and provision of personalised treatment. Such technologies are at the forefront of making Personalised Medicine a possibility. NGS provides a clear picture of genes that are involved in a specific disease and the opportunity to repurpose and/or design new drugs on a case-by-case basis for accurate and precise treatment.[7-9] Such technologies also provide quick and accurate detection of all kind of genetic diseases. Currently, such technologies are used to ascertain the landscape of several genetic diseases that includes majority of the cancers.[10] Having acquired such cutting-edge technology, RMI will not only create jobs, train scientists but also become the only facility in leading the way to making Personalised Medicine a reality in Pakistan.

NGS machines status

A number of machines are available in the market to be selected for various applications.[11] Machine selection is a critical decision based on the country and organization needs. Price of the machine, each run cost, service fee, engineer availability, skilled workforce, ease of use, throughput, and data accuracy are some of the key factors in acquiring a sequencer.[12,13]

A whole lineup of machines are available in the market however, currently available sequencers are Illumina, Ion torrent, Pacific Bioscience (PacBio), BGISeq (not in the market yet), and Nanopore. All these machines come with its own individual pros and cons.[14-16] However, it is worth mentioning that usually the machine service fee ranges from 10-20% of the purchase price per year and thus needed to be included in the budget. The higher the throughput, the harder to gather samples for the run but a lower throughput can cost more per base pair sequence.

Currently, there are a number of Illumina MiSeq machines available in Pakistan and one may want to venture into a pacific bioscience (PacBio machine) as it will serve as a complementary to the MiSeq machines available in the country and will help ease the process. Nanopore machine seems like a good option, however it comes with a higher error rate and may not be suitable as a stand-alone machine at the moment especially if used for diagnostic purposes.[17]

Genomics status in Pakistan

The first-ever Genomics laboratory in Pakistan was initiated on August 12, 2015 at RMI by acquiring an Illumina MiSeq NGS machine. The laboratory faced an uphill battle at the start due to lack of understanding of genomics among scientists, oncologists, cardiologists, clinicians, bio-technologists and other relevant scientists. The Center for Genomic Sciences (CGS) at RMI provides a one-stop solution from project initiation to execution, data analysis, and interpretations.

The newly initiated NGS lab was successful in convincing scientists and funding agencies regarding the benefits and uses of NGS.

Eventually, several funding agencies provided funds to various researchers including Center for Genomic Sciences (CGS) at RMI and the NGS machine was utilized up to some extent. Until now, diagnostic level work is minimum and requires further awareness to fully reap the benefits of the technology. A total of five (05) Illumina machines, including the one in RMI Peshawar, are installed in Pakistan. There is also a discussion on acquiring an Ion Torrent in Pakistan which may not be necessary and may end up a benchtop showpiece. Machine acquisition seemed to have become a fashion but overall, awareness, genomic skills, will, patience, and full determination are still lacking in Pakistani society which is greatly hampering the output of NGS.

Future of Genomics in Pakistan

It is known that genomics is the way forward which is making personalized medicine a reality. Genomics is the way to prevention, early detection, targeted therapies, and prognosis.[18] All ethnicities must move ahead with decoding their own population to make sure their own profile for personalized medicine. Besides dealing with human diseases and genetic manipulation, genomics is the key to changing the genetic makeup of other living organisms for the better future and/or expediting genetic improvements using genetic markers based breeding and selection.[7-9, 19-21]

Currently, the government of Pakistan has included biotechnology together with genomics among the six priority areas of the science and technology. During the last few years various funding agencies have invested around 2 billion PKR on research and development (R&D) in the field of biotechnology by establishing and upgrading various laboratories. However, genomics was least understood and was not given due attention. Genomics needs to be given further attention and importance for achieving quick and drastic improvements. It is genomics which will lead to creating jobs, generating revenues, reducing import and increasing export, however, skilled work force need to be produced through merit based induction at the universities level at the minimum. Furthermore, there must be check and balance with fair accountability combined with severe punishments.

RECOMMENDATIONS

Together with genomics, CRISPR based genome editing and marker-assisted breeding programs need to be established without further delay. The following are some priority areas to be launched with in the country:

- Development and initiation of DNA markers assisted based selection systems.
- Improving the quality and gene yield by adopting genomic section based methods in livestock and crop breeding strategies. NGS based rapid diagnostic tools for early and accurate detection of pathogens and pests.
- Development of Insect and pathogen-resistant plants to reduce the chemical impact on the environment and improve yields.
- Incorporation of NGS based tools and strategies to promote drought and salt-tolerant crops.
- Development of NGS based novel breeding technologies such as CRISPR-Cas coupled with speed breeding.
- Incorporation of high throughput NGS based tools and strategies in various fields of science such as medical biotechnology, neurosciences, medicinal and aromatic plants based research, molecular medicine, industry and environment, DNA Forensics, Microbial, Viral, and Fungal Genomics, and Bioarchaeology, Anthropology, Evolution, and Human Migration.
- Identification, characterization, and development of herbal drugs and nutraceuticals through genetic engineering.

In addition to above proposed strategies, there is a substantial need to develop recombinant and edible vaccines particularly for Cholera, Hepatitis and Rabies. These vaccines need to be tested for large scale production. Furthermore, using NGS based biotechnology tools there is a need to produce biofertilizers and biopesticides. Serious steps are need to incorporate bioprospecting and molecular taxonomy, to improve the nutritional quality of crops and livestock. Serious steps are needed to incorporate and implant bioprocessing in agriculture, livestock breeding and marine biotechnology.

The burden of genetic diseases can be estimated and lowered by launching, and incorporating biobanking, data mining, bioinformatics, establishment of stem cell banks (for treatment of fatal blood diseases), developing various databases and software to ease data storage, processing, and interpretation, and recommend accurate and precise mitigations through genomics data. Furthermore, implementation of Artificial Intelligence (AI) in precision agriculture, medicine, and in livestock sector are needed for various tasks. Proper legislations need to be implemented in order to address problems related to Intellectual Property Rights (IPR), Commercialization, and ethical issues. In addition to all these recommendations, genetic counseling needs to be implemented especially in the areas considered hotspots of consanguineous marriages.

Note: To compete in the international market, strict biosafety, biosecurity, biorisk, and bioethical protocols needed to be developed and adopted. Personnel working in Quarantine need to be trained and equipped to be able to identify genetically modified organisms (GMO) and its products.

CONCLUSION

The incorporation of all the proposed strategies will help the country to meet the current international high-tech based demands in Medical and Agricultural Biotechnology. The vision of Biotechnology policy for Pakistan should be to harness the vast potential of Biotechnology as key contributor to the development of the country. Biotechnology strategies, which have been proposed under several focus areas, if implemented, will usher a new era in the development of the country. Biotechnology implementation in all sectors including crops, livestock, microbes, fungus, and viruses will lead to not only self-sufficiency but also help to eliminate malnutrition and earn much needed foreign revenue.

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