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From the desk of Editor-In-Chief

Editorial: Ensuing importance of quality control for genetic tests in laboratories

With the progress of life science and molecular biology, increasing number of diseases are being found to be linked to genetic abnormalities. Till now almost 1,50,000 diseases have been linked to genetic defects. These genetic changes may be due to established mutations or mutations in evolution in a particular community. To find out a genetic defect at the earliest opportunity is the most proficient way to detect the possibility of a genetically linked disorder at the earliest. As prevention is always better than cure, detection of a disease causing mutation at an early age can prevent a lot of diseases in adult life or at least delay their appearance significantly by appropriate management procedures. This necessitates a more frequent laboratory services for carrying out genetic tests so that the reports of genetic defects may be made available to a wider section of population as well as their treating physicians with accuracy and validity.

However, the present scenario about genetic tests in common laboratories are not that much hopeful and satisfactory. Most laboratories don't have a robust quality control set up for genetic tests. As genetic tests are supposed to generate a permanent gene setup, any erroneous false positive result may provide an unnecessary significant mental trauma in the recipient. In the same way an erroneous false negative result may delay the management process for crucial life threatening disorders like cancers. Hence, an accurate and robust quality control along with quality assurance system should be given prior importance in the laboratories before they start generating reports for genetic disorders.

It is also difficult to delineate a gene defect as a biomarker for a particular disease as most of the genetic disorders are multigenetic with several candidate and principle genes. Laboratories throughout the world face challenges in developing or identifying genetic biomarkers for the diseases which have already been suggested to have multiple genetic risk factors.

In spite of all these challenges, the efforts are going on for validating different genetic markers with their cognate disorders, individual drug responses and drug toxicities. A deep understanding and analysis using the modern whole genome sequencing and next generation gene sequencing tools are needed to establish the relative relevance of each biomarker with a particular genetic disorder. Advancement in the molecular biology techniques and genetics has now enabled the scientist to the process of individualisation of disease diagnosis, their treatment and their monitoring by incorporating the six well established important components of generating a good repertoire of genetic biomarkers1. They are: i. Identification of the candidate for a particular set of genes against a particular disorder in a particular population group, ii. Qualification of the individual or group of genetic biomarkers for suitability for a significant linkage with the concerned disorder, iii. Verification of the observed data, iv. Optimization of the research methods and laboratory methods for the assay procedure, v. Validation of the biomarker using robust quality control methods, and finally vi. Commercialization for making the biomarkers available to the common population and physicians as needed.

In this context, efforts are being made throughout the world including our country for enabling persons to know their risks for future disorders including cancers, allergies and proneness to infective and metabolic diseases, drug tolerability, drug response variations including the addictive ones and most interestingly their probable longevity in the holistic way. Genetic tests are going to hold most important laboratory diagnostic and preventive tests in near future and so, the potential laboratories should start equipping themselves with good, accurate and robust techniques for these tests. Most importantly the process should be monitored by precise quality control and quality assurance systems that can gain significant trust and support from both the common population and their treating physicians or counsellors.

1. Novelli G, Ciccacci C, Borgiani P, Papaluca Amati M, Abadie E. Genetic tests and genomic biomarkers: regulation, qualification and validation. Clin Cases Miner Bone Metab. 2008;5(2):149-54Available from: https://www.ncbi.nlm.nih.gov/pubmed/22460999.

Prof Dr. Anindya Dasgupta

Editor-In-Chief Journal of Applied Biochemistry and Laboratory Medicine