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Editorial

Molecular Diagnosis: A boon to healthcare

A V Moideen Kutty

Diagnostic procedures are one of the essential components in healthcare system. These tests provide cardinal information to enable clinicians to make accurate medical diagnosis, decide on management and treatment of diseases. The field of molecular diagnostics owe a great deal to the developments in molecular biology which reached newer vistas in the early part of twenty first century. The applicability of molecular diagnostics have gathered momentum subsequent to the accomplishments of the human genome project.

Laboratory diagnostic approaches are dynamic and transforming areas on account of the tremendous interest in translational medical research. Consequently, there is a paradigm shift in the approaches of clinicians and the healthcare system is critically and crucially dependent upon laboratory findings in the diagnosis of the diseases. Molecular diagnosis is one of the components of the laboratory diagnostic approaches and broadly it describes a class of diagnostic tests that assesses an individual's health status at molecular levels particularly at the genome level. Molecular diagnostics encompass large number of molecules expressed by genes. Precisely, it would include the identification of genes, its sequencing and expression. The variations observed specify on the possibility of an individual's predisposition for a disease or ability to resist it. Further, it would be of use in the assessment of the suitability and effectiveness of the treatment.

Molecular diagnostics also include detection of the presence of specific viruses, bacteria or different types of cells , which appear in the body as causes or consequences of diseases. This capability, generated integrated molecular diagnostics which enabled targetting diseases like tuberculosis, human immunodeficiency virus and identification of drug resistance. One of the important areas where molecular diagnostics play crucial role is virology where this methedology helped to hasten the process of diagnosis by replacing time consuming culturing methods to more safe, faster and reliable diagnosis.

Precise identification of the microbes causing diseases, the accuracy in locating the specific genes in genetic diseases and expression pattern of specific proteins causing diseases are all integral to molecular diagnositcs. The technologies involved in molecular diagnostic approaches aim to detect and quantify DNA/RNA and their expression products. The methodologies and tools involved are complex and Polymerase Chain Reaction (PCR) is the most fundamental of these techniques, which is used to amplify specific sequences of DNA/RNA.

The advances in technology linked to dealing with the genetic material DNA / RNA lead to furthering of the discovery of additional tests / methods which include FISH, a technique that unzips DNA or RNA in a sample using a labelled fragmant of DNA / RNA as probes that hybridizes with the target sequence . Chips and microarrays are technologies which could simulataneously measure DNA/ RNA, the expression of large number of genes or DNA Single Nucleotide Polymorphism (SNPs) or genome regions. Sequencing of DNA is evolving in a rapid pace and it is one of the most accurate methods either done through Capillary Electrophorosis (CE) or through Next Generation Sequencing (NGS) methods. Mass spectroscopy is another technique which is used to analyze protein biomarkers. Technologies in molecular diagnosis are briefed intentionally in a transitory manner as it's beyond the scope of this note.

Molecular diagnostics comprehensively help clinicians optimally manage cancer patients through risk assessment, screening, diagnosis, staging and progression, therapy selection and monitoring. There are large numbers of molecular diagnostics tests to cater to these requirements such as BRCA1 testing for assessment of risk of breast cancer, HPV test for screening, MTB for definitive diagnosis of tuberculosis, Oncotype Dx to assess severity and risk of recurrence of breast cancer, while Mamma Print is a prognostic and predictive diagnostic test for early stage breast cancer patients that evaluates the risk that the tumor will metastasize . It would also help in staging as well as prognosis. Her2 is used for predic-

tion and efficacy or safety response to specific treatment.

Though molecular diagnostics provide high degree of sensitivity and specificity which are integral for reliable diagnosis, majority of such tests are time consuming. Therefore, the next step of the revolutionary state in molecular diagnostics could be focus on the development of tests which are either faster or rapid. Loop mediated isothermal amplification (LAMP) method is a commendable leap in this direction and a number of methods based on this amplification process have been developed particularly for faster detection of viral agents responsible for diseases.

Molecular diagnosis thus provides a wide spectrum of choices for clinicians to effectively utilize its potential in healthcare. However, these modalities are yet to gain access to the healthcare system primarily on account of the rapid development of specialized tests and point of care devices for early detection as well as management of diseases. The global diagnostic market has been valued at over 65 billion USD with a compound annual growth rate of 6.5%. The United State and European Union account for about 65% of this with Asia- Pacific region recording around 30%. Though, the diagnostic market accounts for a huge economic transaction the molecular diagnostics contribute about 10% with a potential for annual growth estimated at 12% well above the growth of general diagnostic market. This clearly indicates its relevance in healthcare scenario and its utility in personalized medicine.

Molecular diagnostics have gained momentum in various situations of healthcare system including screening, diagnostics, staging, prognosis, therapy selection and monitoring of recurrence. For the clinicians, it is expected to provide vital information for initiating early treatment to prevent the disease from progressing and to avoid trial error strategies in treatment and control disease progression. Despite the fact that molecular diagnosis provide diverse scope in terms of clinical implications in healthcare service, the scope of these diagnostic modalities are highly dependent on technology and involves enormous cost. However, the full potential of molecular tests is yet to be realized but for the advances in sequencing modalities which paved way for development of a spectrum of disease specific diagnostic panels. Further, the directions in which the molecular diagnostics is progressing offer promise for point of care testing to help the clinicians hasten treatment decision.

This editorial has touched upon the surface of the advancements in molecular diagnostics today. The power of technology with clear links to clinical outcomes are gathering momentum day by day and this transitional research platform has tremendous potential of clinical applications.

***Corresponding Author**
Dr. A.V.M.Kutty
Professor,
Department of Cell Biology and Molecular Genetics,
Sri Devaraj Urs Medical College,
Sri Devaraj Urs Academy of Higher Education and Research,
Tamaka, Kolar -563101, Karnataka, India.
Mobile No : 9449767550
E-mail: kuttyavm@gmail.com