

COMMUNICATION

Molecular Diagnostics: A Paradigm Shift

Asma Shabbir¹ and Syed Mehmood Hassan¹

How to cite this article: Shabbir A, Hassan SM. Molecular diagnostics: a paradigm shift. *Ann Jinnah Sindh Med Uni* 2019; 5 (1): 47-48

INTRODUCTION

Effective care given to the patient by a healthcare practitioner depends upon accurate 'diagnosis' and 'treatment' of the ailment. The first line laboratory diagnostic applications like gram staining, hematoxylin and eosin staining, complete blood count, and other biochemical tests have been available for decades. However, they have significant limitations. Molecular diagnostics has offered a dynamic transformation to the field of diagnostics. It is a branch which provides a set of techniques to analyze biological markers in the genetic code (genome) and protein expressed by the genes (proteome). Its role in human diseases widely tags in mutation and pathogen detection. Continuous availability of new methods and new applications has helped molecular diagnostics to be the most rapidly growing field in the laboratory medicine.

With ground breaking developments in molecular biology, we are trying to understand the ways in which diseases develop in an individual. Moreover, it also provides us an idea of disease resistance, immunity, and specific therapy to the patient. Molecular diagnostics is widely being used in hospitals, reference laboratories, and blood banks. The major technologies involved are polymerase chain reaction (PCR), blotting techniques, fluorescent in situ hybridization (FISH), microarray, and mass spectrometry amongst others. These tests are extensively being used in clinical applications including infectious diseases, genetic disease screening, pharmacogenomics, oncology and human leukocyte antigen typing¹.

PCR is the most commonly used technique in major laboratories. It has revolutionized the field of scientific research and medicine. Approximately more than twelve modified types of PCR are now available with

significant advantages over one another. It is playing an emerging role in infectious diseases like tuberculosis, HIV, hepatitis etc. Though the diagnosis of infectious agent can be done through traditional lab tests but PCR remains an exquisite choice as it is a more rapid, sensitive, and specific technique. Similarly, it has also gained importance in the field of histopathology particularly to understand the biology of lymphomas and leukaemia. Translocations in aggressive tumours like sarcomas are now being identified through RT-PCR. DNA profiling is also being utilized in forensic sciences, genetic, and paternity testing².

FISH technique has extended the field of cytogenetics, being more rapid and accurate as compared to the conventional method of karyotype analysis. It has become a vital tool in oncology to detect molecular markers and translocations in various cancers including leukaemia, breast carcinoma, prostate cancer, cholangiocarcinoma, and melanoma. However, literature shows that developing countries have limited use of FISH which might be due to lack of knowledge or unavailability of the technique³. Microarray is another upcoming technique which is not yet routinely used in diagnostics due to its cost and because it also requires skillful hands to work. But, it promises to be as reliable and sensitive as other molecular diagnostic facilities.

Other than the medical field, molecular techniques are also being utilized in agricultural and industrial applications. Moreover, research in gene therapy is ongoing and in the limelight. Approximately 19,000 to 20,000 protein coding genes are estimated to be present in a human genome⁴. In this era of personalized medicine, it is expected that complete human genome will be recognized soon through varieties of molecular techniques. Development and new insights in the field of molecular biology has provided a paradigm shift from routine diagnostic tests to the advancement in diagnosis and treatment of a number of diverse diseases.

¹ Jinnah Sindh Medical University, Karachi, Pakistan

Correspondence: Dr Asma Shabbir, Assistant Professor, Department of Pathology, Sindh Medical College, Jinnah Sindh Medical University, Karachi, Pakistan

Email: drasma52@gmail.com

CONCLUSION

Hence, we conclude that knowledge about molecular biology and diagnostics has become imperative for every medical practitioner. In this period of modern medicine, medical students as well should obtain awareness of molecular diagnostics. Moreover, focus should be on including molecular diagnostics in the medical curriculum.

Authors' contributions: Asma Shabbir conceived the idea and wrote the manuscript. Syed Mehmood Hasan did the critical review. Both the authors approved the manuscript.

References

1. Grody WW, Nakamura RM, Strom CM, Kiechle FL(eds). *Molecular Diagnostics: Techniques and Applications for the Clinical Laboratory*. Academic Press, Inc; Boston, MA. 2010.
2. Kumar A, Rani R, Goel A, Role of PCR in diagnostics. *Bio Evolution*, 2014; 23-24
3. Ratan ZA, Zaman S, Mehta V, Haidere MF, Runa NJ, Akter N, Application of Fluorescence In Situ Hybridization (FISH) Technique for the Detection of Genetic Aberration in Medical Science. *Cureus*, 2017; 9(6):1325, doi:10.7759/cureus.1325, PMID:28690958
4. Ezkurdia I, Juan D, Rodriguez JM, Frankish A, Diekhans M, Harrow J, et.al. Multiple evidence strands suggest that there may be as few as 19,000 human protein-coding genes. *Hum Mol Genet*. 2014; 23(22):5866–78. doi: 10.1093/hmg/ddu309.