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The Rules of Molecular Technique in Diseases Identification

Sara Ahmed Zulkifli

Kulliyyah of Allied Health Sciences (KAHS), International Islamic University Malaysia (IIUM),
Kuantan, Malaysia

zulkifli_sarah@yahoo.com

Article Info.

Article history:

Received 2 April 2022

Revised 26 May 2022

Published 15 June 2022

Keywords:

Molecular techniques, RT-PCR,
Disease Identification

How to cite:

Sara Ahmed Zulkifli, The Rules of
Molecular Technique in Disease
Identification. Aca. Intl. J. Med.
Sci., 2022;1(1):23-27:

<https://doi.org/10.59675/M2022-03>

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Abstract

Molecular techniques have revolutionized the identification and diagnosis of various diseases, providing unprecedented accuracy and speed. This article explores the fundamental rules governing molecular techniques in disease identification, focusing on their applications in infectious diseases, oncology, neurodegenerative disorders, and allergies. It discusses the evolution of these techniques, their clinical implications, and the challenges faced in their implementation. By synthesizing recent advancements and recommendations from the literature, this article aims to provide a comprehensive overview of the current landscape of molecular diagnostics.

1. Introduction

The advent of molecular diagnostics has transformed the landscape of disease identification, enabling clinicians to detect pathogens, genetic mutations, and biomarkers with high specificity and sensitivity. Molecular techniques, including polymerase chain reaction (PCR), next-generation sequencing (NGS), and microRNA profiling, have become integral to modern medicine, particularly in the fields of infectious diseases, oncology, and neurodegenerative disorders [1–5]. This article delineates the rules and principles underlying these molecular techniques, emphasizing their significance in clinical practice.

2. Molecular Techniques in Infectious Disease Diagnosis

Molecular diagnostics have significantly enhanced the detection of infectious diseases. Techniques such as quantitative RT-PCR and multiplex PCR panels allow for the rapid identification of pathogens, including bacteria, viruses, and fungi, directly from clinical specimens [2,4–6]. The sensitivity of these methods often surpasses traditional culture techniques, which can be time-

consuming and may fail to detect certain pathogens [7,8]. For instance, the use of ribosomal RNA-based PCR has shown promise in detecting periprosthetic infections, highlighting the potential of molecular techniques to improve diagnostic accuracy [9].

Moreover, advancements in point-of-care testing, facilitated by smartphone-based genetic analyzers, have made it possible to perform molecular diagnostics in real-time, thus expediting patient management [10]. However, the implementation of these technologies is not without challenges. Issues such as the need for specialized training, the cost of equipment, and the interpretation of complex data must be addressed to optimize the use of molecular diagnostics in clinical settings [6,8].

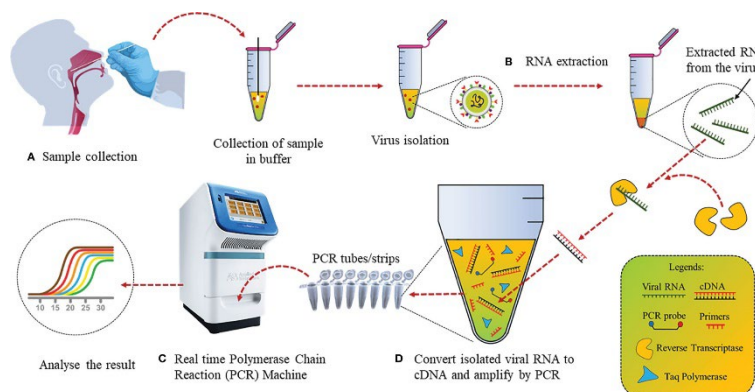


Figure 1 Steps of infectious disease detection by conventional RT-PCR [11]

3. Molecular Diagnostics in Oncology

In oncology, molecular diagnostics play a crucial role in the early detection and characterization of tumors. Techniques such as NGS enable the identification of genetic mutations associated with various cancers, facilitating personalized treatment strategies [12,13]. For example, the identification of IDH1 mutations and 1p/19q codeletion in gliomas has significant prognostic implications, guiding therapeutic decisions [14,15]. Furthermore, molecular tests for circulating tumor DNA (ctDNA) are emerging as valuable tools for monitoring treatment response and detecting minimal residual disease [1,3].

The integration of molecular diagnostics into histopathology training has also been emphasized, as it allows pathologists to utilize genetic information alongside traditional histological assessments [16]. This multidisciplinary approach enhances the accuracy of cancer diagnoses and supports the development of targeted therapies.

4. Neurodegenerative Disorders and Molecular Diagnostics

Molecular techniques are increasingly being employed to diagnose neurodegenerative disorders such as Alzheimer's disease, Parkinson's disease, and amyotrophic lateral sclerosis. Biomarkers identified through molecular diagnostics can facilitate early detection and monitoring of disease progression [1,17]. For instance, the analysis of specific microRNAs has shown potential as non-invasive micromarkers for these conditions, although further research is needed to fully understand their roles in disease pathophysiology (Saikumar et al., 2014).

The challenges in diagnosing neurodegenerative diseases using molecular techniques include the complexity of the diseases themselves and the need for standardized protocols for biomarker validation [1,17]. Nonetheless, the promise of molecular diagnostics in this field is substantial, with ongoing research aimed at refining these techniques for clinical application.

5. Molecular Techniques in Allergy Diagnosis

Molecular diagnostics have also found applications in the diagnosis of allergic diseases. The use of molecular allergenic components allows for a more precise understanding of patient sensitization profiles, which is crucial for effective management [19]. Recommendations for the use of these advanced diagnostic methods have been developed to guide clinicians in their application, ensuring that patients receive accurate diagnoses and appropriate treatment [19]

6. Challenges and Future Directions

Despite the advancements in molecular diagnostics, several challenges remain. The complexity of molecular data interpretation, the need for robust quality control measures, and the integration of these techniques into routine clinical practice are critical areas that require attention[3,20]. Additionally, the rapid evolution of technology necessitates continuous education and training for healthcare professionals to keep pace with new developments [16,21].

Future directions in molecular diagnostics include the exploration of novel biomarkers, the refinement of existing techniques, and the expansion of point-of-care testing capabilities. As the field continues to evolve, it is essential to establish standardized protocols and guidelines to ensure the reliability and accuracy of molecular diagnostic tests (Emmadi et al., 2011; Piñero et al., 2016).

7. Conclusion

Molecular techniques have fundamentally changed the approach to disease identification, offering enhanced accuracy and speed in diagnostics. The integration of these techniques into clinical practice holds great promise for improving patient outcomes across various medical fields. However, addressing the challenges associated with their implementation is crucial for maximizing their potential. Continued research and collaboration among clinicians, researchers, and technologists will be vital in shaping the future of molecular diagnostics.

References

- [1] Agrawal M, Cho W. Editorial: molecular diagnostics in the detection of neurodegenerative disorders. *Front Mol Biosci* 2017;4. <https://doi.org/10.3389/fmolb.2017.00010>.
- [2] Fairfax M, Salimnia H. Diagnostic molecular microbiology. *Clin Lab Med* 2013;33:787–803. <https://doi.org/10.1016/j.cll.2013.08.003>.
- [3] Biswas B. Clinical performance evaluation of molecular diagnostic tests. *Journal of Molecular Diagnostics* 2016;18:803–12. <https://doi.org/10.1016/j.jmoldx.2016.06.008>.

- [4] Almanseekanaa LH, Ali Alabbas AK, Kadhim NJ, Ogaili RH. MOLECULAR STUDY OF PSEUDOMONAS AERUGINOSA ISOLATED FROM DIFFERENT CLINICAL CASES. *Biochem Cell Arch* 2021;21.
- [5] Kadhim NJ, Al-Karawi NJ, Almanseekanaa LH, Ogaili RH. Molecular characterization and antibiotic pattern of the *Klebsiella pneumoniae*. *Veterinary Practitioner* 2021;22.
- [6] Emmadi R, Boonyaratanakornkit J, Selvarangan R, Shyamala V, Zimmer B, Williams L, et al. Molecular methods and platforms for infectious diseases testing. *Journal of Molecular Diagnostics* 2011;13:583–604. <https://doi.org/10.1016/j.jmoldx.2011.05.011>.
- [7] Wickes B, Wiederhold N. Molecular diagnostics in medical mycology. *Nat Commun* 2018;9. <https://doi.org/10.1038/s41467-018-07556-5>.
- [8] Kociolek L. Strategies for optimizing the diagnostic predictive value of *Clostridium difficile* molecular diagnostics. *J Clin Microbiol* 2017;55:1244–8. <https://doi.org/10.1128/jcm.00147-17>.
- [9] Bergin P, Doppelt J, Hamilton W, Mirick G, Jones A, Sritulanondha S, et al. Detection of periprosthetic infections with use of ribosomal rna-based polymerase chain reaction. *The Journal of Bone and Joint Surgery (American)* 2010;92:654–63. <https://doi.org/10.2106/jbjs.i.00400>.
- [10] Nguyen H, Nguyen V, Liu F, Seo T. An integrated smartphone-based genetic analyzer for qualitative and quantitative pathogen detection. *ACS Omega* 2020;5:22208–14. <https://doi.org/10.1021/acsomega.0c02317>.
- [11] Roberts A, Chouhan RS, Shahdeo D, Shrikrishna NS, Kesarwani V, Horvat M, et al. A Recent Update on Advanced Molecular Diagnostic Techniques for COVID-19 Pandemic: An Overview. *Front Immunol* 2021;12. <https://doi.org/10.3389/fimmu.2021.732756>.
- [12] Johnston J, Rubinstein W, Facio F, Ng D, Singh L, Teer J, et al. Secondary variants in individuals undergoing exome sequencing: screening of 572 individuals identifies high-penetrance mutations in cancer-susceptibility genes. *The American Journal of Human Genetics* 2012;91:97–108. <https://doi.org/10.1016/j.ajhg.2012.05.021>.
- [13] Gilissen C, Hoischen A, Brunner H, Veltman J. Unlocking mendelian disease using exome sequencing. *Genome Biol* 2011;12:228. <https://doi.org/10.1186/gb-2011-12-9-228>.
- [14] Tabatabai G, Stupp R, Bent M, Hegi M, Tonn J, Wick W, et al. Molecular diagnostics of gliomas: the clinical perspective. *Acta Neuropathol* 2010;120:585–92. <https://doi.org/10.1007/s00401-010-0750-6>.
- [15] Reifenberger G, Wesseling P. Molecular diagnostics of brain tumors. *Acta Neuropathol* 2010;120:549–51. <https://doi.org/10.1007/s00401-010-0752-4>.

- [16] Flynn C, James J, Maxwell P, McQuaid S, Ervine A, Catherwood M, et al. Integrating molecular diagnostics into histopathology training: the belfast model. *J Clin Pathol* 2014;67:632–6. <https://doi.org/10.1136/jclinpath-2014-202176>.
- [17] Agrawal M, Biswas A. Molecular diagnostics of neurodegenerative disorders. *Front Mol Biosci* 2015;2. <https://doi.org/10.3389/fmolb.2015.00054>.
- [18] Saikumar J, Ramachandran K, Vaidya V. Noninvasive micromarkers. *Clin Chem* 2014;60:1158–73. <https://doi.org/10.1373/clinchem.2013.216044>.
- [19] Villalta D, Tonutti E, Bizzaro N, Brusca I, Sargentini V, Asero R, et al. Recommendations for the use of molecular diagnostics in the diagnosis of allergic diseases. *Eur Ann Allergy Clin Immunol* 2018;50:51. <https://doi.org/10.23822/eurannaci.1764-1489.32>.
- [20] Heuvel L, Smeitink J, Rodenburg R. New approaches to diagnosing mitochondrial abnormalities: taking the next step. *J Pediatr Biochem* 2016;02:205–12. <https://doi.org/10.1055/s-0036-1586417>.
- [21] Vranic S, Damjanov I. Advances in diagnostic and molecular pathology. *Acta Med Acad* 2021;50:1. <https://doi.org/10.5644/ama2006-124.321>.
- [22] Piñero J, Berenstein A, González-Pérez A, Chernomoretz A, Furlong L. Uncovering disease mechanisms through network biology in the era of next generation sequencing. *Sci Rep* 2016;6. <https://doi.org/10.1038/srep24570>.