**Case Studies of Novel Diagnostics in Clinical Practice**

Emmanuella Amara Ofoka Research Associate Texas A&M University,

 Department of Veterinary Physiology and Pharmacology College Station,

Texas, USA

emmanuella.amara@tamu.edu

ABSTRACT

The advent of novel diagnostics has revolutionized clinical practice by enhancing the speed, accuracy, and personalization of disease detection and management. This paper provides some associated case studies and explores the transformative potential of advanced diagnostic tools including liquid biopsies and point-of-care testing (POCT). Liquid biopsy, exemplified through a case of cancer, demonstrates the utility of noninvasive genetic mutation analysis in enabling targeted therapy and dynamic treatment monitoring. Similarly, the application of POCT in infectious disease management highlights its efficacy in providing rapid results, improving accessibility in resource-limited settings, and facilitating timely medical interventions. Despite these advancements, challenges such as high implementation costs, training requirements, and regulatory hurdles persist. There is a need for collaborative efforts among stakeholders to address these issues and leverage innovations, such as artificial intelligence and bioinformatics, for greater diagnostic precision. Ultimately, these advancements are pivotal in reshaping healthcare delivery, enhancing patient outcomes, and fostering an environment that prioritizes equity and efficiency. By embracing personalized medicine, we can ensure that each patient receives tailored care, ultimately leading to a more effective healthcare system. As we look to the future, the integration of innovative diagnostic tools will play a crucial role in advancing medical practices and improving the overall health of populations. Keywords- case study, novel diagnosis, point-of-care, liquid biopsy.

# INTRODUCTION

Novel diagnostics have emerged as a crucial aspect of clinical practice, aiming to enhance disease detection and treatment accuracy, efficiency, and speed [1]. These diagnostic tools are vital in improving patient outcomes across various medical fields by leveraging advanced technologies and innovative methodologies [2]. Molecular diagnostics have gained immense traction due to their ability to analyze genetic and molecular markers associated with diseases [3]. These techniques enable the identification of specific pathogens or genetic mutations, leading to more accurate diagnoses. For instance, next-generation sequencing (NGS) enables the identification of a broad range of genetic conditions and cancers, significantly improving diagnostic success rates in rare genetic diseases and complex tumors [4]. The advent of accessible technologies has enabled the application of molecular diagnostics at the point of care, thereby expediting the diagnostic process. Technological advancements have transformed diagnostics [5]. Tools such as portable diagnostic devices, like the GeneXpert Omni system, provide fast results for various conditions, including tuberculosis and HIV, within hours [6]. These devices have made conducting tests in smaller clinics and remote areas feasible, increasing accessibility to critical diagnostic services [6]. Furthermore, technologies such as microfluidics and smartphone dongles have facilitated rapid testing with high sensitivity and specificity, allowing for effective field diagnostics [7].

Despite the advancements, integrating novel diagnostics into clinical practice faces several challenges. Issues such as the need for regulatory approval, cost-effectiveness, and training for healthcare providers remain pertinent [1]. Moreover, ensuring the accuracy and reliability of new diagnostic tools is crucial. For example, while rapid tests for infectious diseases have led to quicker diagnoses, the potential for false negatives or positives must be managed [2]The future of diagnostics in clinical practice is promising, with ongoing research aimed at refining existing technologies and developing new methodologies [1]. Efforts are being made to enhance bioinformatics capabilities to interpret complex datasets generated by advanced diagnostic tools. This will allow for better integration of data around patient care, ultimately aimed at personalized medicine [3]. Additionally, emerging technologies in AI are expected to contribute to improved efficiency and accuracy in diagnostic decision-making, opening new avenues for patient management [2]. ​In this chapter, we discussed how novel diagnostics reshape clinical practice, provide timely disease detection methods, and state some case studies of novel diagnostics in clinical practice: the novel clinical challenges and the potential for significantly improved patient care through these innovations.

# Acceleration of Infectious Disease Diagnosis by Molecular Methods

The advent of molecular diagnostics has significantly accelerated the detection of infectious diseases by enabling rapid, sensitive, and specific identification of pathogens directly from clinical specimens. Compared to traditional microbiological methods, such as culture and immunoassays molecular platforms has markedly enhanced clinical decision making and improving patient outcomes.

Core Elements Accelerating Diagnosis

1. Direct Detection of Pathogen Genetic Material

 Unlike culture-based methods, which require time for microbial growth, molecular diagnostics detect pathogen-specific DNA or RNA directly from patient samples[4]. Techniques such as polymerase chain reaction (PCR) and isothermal amplification amplify target nucleic acids exponentially, delivering actionable results within hours. This capability eliminates the delays inherent in cultivation, especially for fastidious or slow-growing organisms [4].

1. Rapid Amplification Technologies

 Advanced PCR modalities, including quantitative PCR (qPCR) and digital PCR (dPCR), enable real-time detection of nucleic acid targets [5], often yielding results in under two hours. Isothermal amplification techniques such as loop-mediated isothermal amplification (LAMP) and recombinase polymerase amplification (RPA) offer further time efficiencies by operating at constant temperatures and generating results in as little as 15 to 60 minutes [6] [5]. These approaches also reduce equipment complexity, making them suitable for broader clinical deployment [5].

1. Point-of-Care and Automated Platforms

 Molecular point-of-care (POC) systems integrate key diagnostic steps including sample lysis, nucleic acid extraction, amplification, and detection into compact, user-friendly, and often cartridge-based platforms [7]. These systems can deliver results in less than one hour, even outside traditional laboratory environments, reducing delays due to specimen transport and centralized processing[8].

1. Multiplex and High-throughput Testing

 Multiplex PCR and microarray-based assays enable the simultaneous detection of multiple pathogens, virulence factors, or antimicrobial resistance genes in a single reaction [9]. This reduces the need for serial testing and expedites the diagnostic process, particularly in syndromic presentations such as respiratory infections, sepsis, or meningitis [9].

# Applications of novel diagnostics in infectious disease management

Incorporating novel diagnostics into clinical practice is crucial because it allows for earlier disease detection, more accurate diagnoses, personalized treatment plans, and improved patient outcomes. It can lead to more efficient healthcare delivery by enabling faster diagnosis and targeted therapies, minimizing unnecessary interventions, and improving overall patient care [10]. For instance, advanced molecular diagnostics can identify pathogens or genetic mutations quickly and accurately, allowing healthcare providers to make informed decisions rapidly [11]. Timely diagnoses can significantly reduce delays in treatment and improve prognostic outcomes, particularly in acute conditions where every moment counts. Providing accurate results in a shorter time frame directly influences the effectiveness of clinical interventions [12]. However, misdiagnosis is a substantial concern in healthcare, often leading to inappropriate treatment and adverse patient outcomes [13]. Incorporating advanced diagnostics can significantly mitigate this issue by providing more sophisticated tools for assessment [11]. These innovations have improved infectious disease detection, identification, and treatment. For instance, systems like the GeneXpert assay have revolutionized Tuberculosis (TB) diagnostics by allowing rapid detection of TB as well as rifampicin resistance within just a few hours, significantly outpacing traditional culture methods that take weeks to yield results [14]. Many people have been diagnosed with streptococcal (GAS) pharyngitis; this led to the development of rapid antigen detection tests (RADTs). Unfortunately, physical examination and clinical findings are inadequate to diagnose GAS pharyngitis. As its symptoms closely resemble those of other infectious causes of pharyngitis [15]. The Infectious Diseases Society of America (IDSA) recommends examination of this infection with RADTs as it is swift and accurate (Shulman et al., 2012). However, integrating novel diagnoses in clinical practice is essential in impacting patient outcomes. Recent studies have shown that multiplex PCR can identify multiple pathogens simultaneously from clinical samples, such as respiratory secretions during outbreaks of viral infections [9]. This also permits more effective surveillance programs, facilitating quick containment measures during outbreaks [17] Also, rapid identification of pathogens allows timely targeted treatment, which is crucial in scenarios where empirical treatment could lead to adverse outcomes, such as sepsis, where misdiagnosis can be life-threatening [18]. With the aid of novel diagnostics, healthcare professionals can initiate appropriate treatment plans without unnecessary delays that typically arise from laboratory testing [18].Thus, novel diagnostics have helped mitigate the effects of antimicrobial resistance by accurately identifying the causative agent of infections and their sensitivities to antimicrobial agents, thereby promoting better stewardship of existing antibiotics [19].

# Case study diagnosis through rapid diagnostic technology.

1. Case Study: Diagnosis of Cancer through Liquid Biopsy Testing

 Liquid biopsy has emerged as a minimally invasive and highly informative tool in oncology, particularly for patients in whom tissue biopsies are not feasible and patients present with non-specific symptoms, including persistent cough, weight loss, and fatigue. Initial imaging revealed a suspicious pulmonary nodule, but bronchoscopy failed to yield adequate tissue for histological evaluation [20]. However, a liquid biopsy performed identified circulating tumor DNA (ctDNA) harboring an EGFR exon 19 deletion mutation commonly associated with non-small cell lung cancer (NSCLC) [20]. Based on these findings, the patient started on first-line targeted therapy with Osimertinib, leading to significant clinical improvement and a reduction in tumor burden within eight weeks. This case highlights the clinical utility of liquid biopsy, not only for early cancer detection but also for molecular profiling to inform personalized treatment. Beyond lung cancer, liquid biopsies have shown promise in detecting mutations, monitoring minimal residual disease, and tracking treatment response in breast, colorectal, and prostate cancers. Unlike conventional biopsies, which carry procedural risks and may not capture tumor heterogeneity, liquid biopsy provides a real-time, systemic snapshot of the tumor's genetic landscape [21]. As such, it represents a paradigm shift in cancer diagnostics and precision oncology.

1. Case Study of a Patient Diagnosed with an Infectious Disease Through Point-of-Care Testing.

 This case exemplifies the application of point-of-care (PoC) HIV testing at birth within a pragmatic clinical trial conducted in Mozambique and Tanzania. In the study, out of 6602 enrolled neonates, 125 were diagnosed with HIV by 12 weeks of age. Notably, 55.1% of infants in the intervention group who received birth PoC testing were diagnosed at birth, enabling immediate initiation of antiretroviral therapy (ART) with a median age at the start of just 6 days. In contrast, infants diagnosed later in the control group had a median age at ART initiation of 33 days[22]. Early detection through point-of-care (POC) testing enabled rapid treatment initiation, which is crucial for reducing early mortality. Although the study showed that birth test-and-treat did not significantly improve clinical outcomes at 18 months, it was associated with a substantial 68% reduction in mortality within the first 6 months [22]. The trial showed that birth POC test-and-treat strategies are feasible and can have a meaningful impact on neonatal outcomes, demonstrating the importance of early diagnosis and treatment in controlling infant HIV infections in resource-poor settings [22].

1. Case Study 3: Diagnosing Syphilis in Antenatal Care

 In a study conducted in a South American country, a pregnant woman was screened for syphilis using a rapid point-of-care test during her first prenatal visit. The RDT returned a positive result, leading to immediate treatment with benzathine penicillin. This timely intervention helped prevent potential congenital syphilis [23]. Additionally, the García et al., 2013 study highlighted the importance of integrating POCT into routine antenatal care, significantly increasing the detection rate of syphilis and ensuring better maternal and neonatal health outcomes [23]. These case studies collectively illustrate the transformative impact of point-of-care testing in diagnosing infectious diseases.​ Delivering rapid results facilitates immediate treatment, enhances patient care, and plays a vital role in controlling outbreaks. As healthcare systems adopt POCT, the potential for better health outcomes, especially in resource-limited settings, becomes increasingly clear.



Figure 1: Comparative overview of amplification-based molecular diagnostic

Figure 1 illustrates how clinical samples, such as nasal swabs, blood, or saliva, undergo amplification to detect pathogen nucleic acids. Three key technologies are commonly used for this purpose: Polymerase Chain Reaction (PCR), Loop-Mediated Isothermal Amplification (LAMP), and Nucleic Acid Detection Platforms (NADP). PCR is known for its high-throughput capabilities and automation; however, it is limited by the complexity of primer design and risks of contamination. LAMP, on the other hand, offers portability and speed without the need for a thermocycler, but it can be susceptible to nonspecific amplification and inhibition from substances present in tissue. NADP targets RNA-specific sequences, allowing for functional analysis, but it also faces similar specificity limitations. These methods are employed to produce diagnostic outputs for diseases such as HIV, HPV, respiratory viruses, and tuberculosis. Key challenges that persist across all platforms include variable sample quality, the presence of biological inhibitors (such as maternal DNA), and the dependence on manual steps in traditional workflows (Diagram created by E. A. Ofoka using BioRender)

# Importance of incorporating novel diagnostics into routine clinical practice.

Incorporating novel diagnostics into clinical practice is crucial because it allows for earlier disease detection, more accurate diagnoses, personalized treatment plans, and improved patient outcomes. It can lead to more efficient healthcare delivery by enabling faster diagnosis and targeted therapies, minimizing unnecessary interventions, and improving overall patient care [10]. For instance, advanced molecular diagnostics can identify pathogens or genetic mutations quickly and accurately, allowing healthcare providers to make informed decisions rapidly [11]. Timely diagnoses can significantly reduce delays in treatment and improve prognostic outcomes, particularly in acute conditions where every moment counts. Providing accurate results in a shorter time frame directly influences the effectiveness of clinical interventions [24]. However, misdiagnosis is a substantial concern in healthcare, often leading to inappropriate treatment and adverse patient outcomes [13]. Incorporating advanced diagnostics can significantly mitigate this issue by providing more sophisticated tools for assessment. For example, molecular diagnostics can utilize biological markers specific to diseases, thus enhancing differentiation between similar conditions [24]. Employing techniques such as genomic sequencing, clinicians can identify specific variants in patients and customize therapeutic strategies accordingly[25]. This personalization can result in more effective treatment plans, minimizing trial and error in medication selection and increasing the likelihood of favorable outcomes [13]. Although the initial investment in advanced diagnostic technologies can be high, they tend to save costs in the long run by reducing the need for unnecessary tests and treatments associated with misdiagnosis [26]. Effective diagnostics lead to targeted interventions, translating to better resource allocation and potentially lower overall healthcare costs when considering the long-term management of chronic conditions [26].

# Challenges in implementing rapid diagnostics

Rapid diagnostic technologies have demonstrated significant potential in improving infectious disease management, their implementation remains constrained by several key challenges. One of the primary limitations involves the complexity and cost associated with many molecular and nucleic acid–based assays [19]. Despite their high sensitivity and specificity, these platforms often necessitate advanced laboratory infrastructure, specialized personnel, and costly instrumentation, limiting their accessibility in outpatient settings, emergency departments, and low-resource environments. Integration of diagnostic results into clinical workflows also presents a substantial barrier [27]. The clinical utility of rapid diagnostics hinges on the timely communication of results to guide antimicrobial therapy and inform infection control measures[28]. However, in many healthcare systems, the absence of robust electronic medical record systems and automated reporting tools impedes prompt delivery of results to clinicians or antimicrobial stewardship teams, thereby reducing the potential impact of these diagnostics on patient outcomes and antimicrobial use[29]. Regulatory and reimbursement frameworks further complicate implementation. Lengthy and demanding regulatory approval processes can delay the clinical availability of novel diagnostics, while inconsistent or inadequate reimbursement policies across regions may disincentivize adoption by laboratories and healthcare institutions[30]. Addressing these regulatory and economic barriers is critical to fostering innovation and ensuring long-term sustainability of rapid diagnostic deployment. Moreover, specific populations, such as pediatric and immunocompromised patients present unique diagnostic challenges. Limited validation of assays in children, due in part to difficulties in specimen collection and a smaller commercial market, has resulted in a scarcity of pediatric-specific diagnostic tools [31]. In immunosuppressed patients, atypical clinical presentations and a broader spectrum of potential pathogens necessitate more sensitive and comprehensive diagnostic approaches, adding further complexity to test design and clinical interpretation [32]. These tests may detect colonization or nonviable organisms, raising the risk of overtreatment and unnecessary isolation. Differentiating active infection from colonization or residual nucleic acids particularly in respiratory virus detection and sepsis diagnostics remains a critical need [33]. Advancements in biomarker discovery, the establishment of clinically relevant quantitative thresholds, and targeted clinician education are essential to enhance the accuracy and utility of rapid diagnostics in routine clinical practice. The cost-effectiveness of novel infectious disease diagnostics remains a critical, yet often underappreciated, component in the discussion of implementation challenges. Rapid and accurate diagnostics have the potential to significantly reduce healthcare expenditures while improving clinical outcomes. By facilitating timely and targeted antimicrobial therapy, these tools help minimize unnecessary antibiotic use, reduce hospital length of stay, and prevent complications arising from delayed or inappropriate treatment.

For instance, nucleic acid amplification tests (NAATs) used in the diagnosis of viral central nervous system infections, such as enteroviral meningitis and herpes simplex virus encephalitis, have demonstrated both clinical benefit and economic value by decreasing hospitalization duration and reducing the need for additional diagnostic workup [34]. Similarly, rapid molecular identification of antimicrobial resistance genes allows clinicians to promptly tailor therapy, avoiding prolonged ineffective treatment and associated costs [18]. Point-of-care (POC) molecular diagnostics, particularly those with turnaround times under one hour, have shown utility in emergency and outpatient settings by enabling immediate clinical decisions and reducing inappropriate antimicrobial prescriptions. This not only improves patient management but also supports antimicrobial stewardship and reduces broader healthcare costs through optimized resource utilization [8]. Advanced multiplex PCR panels that detect a wide array of pathogens simultaneously further contribute to economic efficiency by increasing diagnostic yield and enabling early, targeted treatment [6]. These panels have been associated with decreased hospital admissions, shorter ICU stays, and fewer ancillary tests factors that together offset the higher initial investment required for these platforms [9]. Importantly, rapid diagnostics have also demonstrated utility in infection control by facilitating timely isolation of contagious individuals and enabling swift responses to outbreaks, thereby avoiding nosocomial transmission and extended hospitalizations [35]. Despite these benefits, challenges related to cost remain. The initial investment required for equipment, consumables, and skilled personnel can be substantial, especially in resource-limited settings. Moreover, inconsistent reimbursement policies and regulatory uncertainties hinder broader adoption. Even in high-income healthcare systems, the lack of integrated cost-effectiveness models within diagnostic evaluations complicates decision-making for health administrators and policymakers [36]. There is a growing need for standardized economic evaluations that incorporate patient outcomes, antimicrobial usage, length of stay, and infection prevention metrics to provide a more comprehensive assessment of value. Training and education of clinicians and laboratory personnel on the appropriate interpretation and use of these diagnostics are essential to maximizing their cost-saving potential. Furthermore, declining equipment costs and increasing automation may improve affordability and scalability soon, enhancing accessibility across diverse clinical settings [37]. Overall, while the initial expenditures for novel diagnostic platforms may be significant, the downstream savings achieved through improved clinical outcomes, reduced unnecessary treatments, shorter hospitalizations, and better infection control strongly support their cost-effectiveness.

# Discussion

Integrating novel diagnostics into clinical practice represents a significant leap forward in the early detection, accurate diagnosis, and effective management of diseases. As demonstrated in this chapter, the development and application of advanced diagnostic tools, such as liquid biopsies and point-of-care testing (POCT), have had transformative impacts across diverse medical fields. These technologies bridge the gap between traditional diagnostic methods and the growing demand for precision medicine, offering advantages in speed, accessibility, and personalization[38]. As showcased in the cancer case study, liquid biopsy underscores the power of noninvasive techniques in identifying actionable genetic mutations, such as the EGFR mutation in bronchopulmonary adenocarcinoma [39]. The rapid initiation of targeted therapy, as illustrated, can lead to remarkable clinical improvements and better prognostic outcomes. This case study highlights the role of novel diagnostics in diagnosing complex diseases and monitoring treatment responses effectively. By reducing the need for invasive procedures and enabling dynamic disease tracking, liquid biopsies provide a compelling argument for their integration into routine oncology practice[40]. Similarly, using POCT in infectious disease management exemplifies the role of rapid diagnostics in improving healthcare delivery, particularly in resource-limited settings. Malaria and HIV case studies emphasize how timely diagnosis facilitated by RDTs can drastically reduce morbidity and mortality [41] [42]. POCT's ability to provide immediate results at or near the point of care enables healthcare professionals to make informed decisions swiftly, reducing delays often accompanying traditional laboratory methods. These advancements are particularly impactful in regions with limited access to centralized healthcare infrastructure, where they support timely interventions and enhance patient outcomes. Despite these successes, challenges remain, such as the high initial costs of implementing novel diagnostic technologies and the need for specialized training and regulatory approvals, significant barriers to widespread adoption. Ensuring accuracy and reliability while minimizing false positives or negatives is another critical aspect requiring attention [43]. Addressing these challenges will necessitate collaboration among stakeholders, including healthcare providers, policymakers, and technology developers, to create an ecosystem conducive to innovation and equitable access. The continued evolution of diagnostics, particularly with advancements in artificial intelligence and bioinformatics, holds immense potential ([3]. These technologies enhance data interpretation, facilitate personalized medicine, and streamline the integration of diagnostic tools into clinical workflows.

# Reference

[1] R. W. Peeling, D. L. Heymann, Y.-Y. Teo, and P. J. Garcia, “Diagnostics for COVID-19: moving from pandemic response to control,” *The Lancet*, vol. 399, no. 10326, pp. 757–768, Feb. 2022, doi: 10.1016/S0140-6736(21)02346-1.

[2] D. Gala, H. Behl, M. Shah, and A. N. Makaryus, “The Role of Artificial Intelligence in Improving Patient Outcomes and Future of Healthcare Delivery in Cardiology: A Narrative Review of the Literature,” *Healthcare*, vol. 12, no. 4, p. 481, Feb. 2024, doi: 10.3390/healthcare12040481.

[3] Q. Liu, X. Jin, J. Cheng, H. Zhou, Y. Zhang, and Y. Dai, “Advances in the application of molecular diagnostic techniques for the detection of infectious disease pathogens (Review),” *Mol Med Rep*, vol. 27, no. 5, p. 104, Apr. 2023, doi: 10.3892/mmr.2023.12991.

[4] L. Samuel, “Direct-from-Blood Detection of Pathogens: a Review of Technology and Challenges,” *J Clin Microbiol*, vol. 61, no. 7, Jul. 2023, doi: 10.1128/jcm.00231-21.

[5] I. of M. (US) C. on E. M. T. to H. in the 21st Century, M. S. Smolinski, M. A. Hamburg, and J. Lederberg, “Pathogen Discovery, Detection, and Diagnostics,” 2003, Accessed: Jun. 12, 2025. [Online]. Available: https://www.ncbi.nlm.nih.gov/books/NBK221492/

[6] P. Belgrader *et al.*, “PCR Detection of Bacteria in Seven Minutes,” *Science (1979)*, vol. 284, no. 5413, pp. 449–450, Apr. 1999, doi: 10.1126/science.284.5413.449.

[7] H. Chen, K. Liu, Z. Li, and P. Wang, “Point of care testing for infectious diseases,” *Clinica Chimica Acta*, vol. 493, pp. 138–147, Jun. 2019, doi: 10.1016/j.cca.2019.03.008.

[8] K. Gavina, L. C. Franco, H. Khan, J. P. Lavik, and R. F. Relich, “Molecular point-of-care devices for the diagnosis of infectious diseases in resource-limited settings – A review of the current landscape, technical challenges, and clinical impact,” *Journal of Clinical Virology*, vol. 169, p. 105613, Dec. 2023, doi: 10.1016/J.JCV.2023.105613.

[9] O. Lung, T. Furukawa-Stoffer, K. Burton Hughes, J. Pasick, D. P. King, and D. Hodko, “Multiplex RT-PCR and Automated Microarray for Detection of Eight Bovine Viruses,” *Transbound Emerg Dis*, vol. 64, no. 6, pp. 1929–1934, Dec. 2017, doi: 10.1111/tbed.12591.

[10] P. Croft *et al.*, “The science of clinical practice: disease diagnosis or patient prognosis? Evidence about ‘what is likely to happen’ should shape clinical practice,” *BMC Med*, vol. 13, no. 1, p. 20, Dec. 2015, doi: 10.1186/s12916-014-0265-4.

[11] P. Hunter, “Novel diagnostic technologies for clinical and frontline use,” *EMBO Rep*, vol. 18, no. 6, pp. 881–884, Jun. 2017, doi: 10.15252/embr.201744423.

[12] R. L. Zimmern, “Testing challenges: evaluation of novel diagnostics and molecular biomarkers,” *Clinical Medicine*, vol. 9, no. 1, pp. 68–73, Feb. 2009, doi: 10.7861/clinmedicine.9-1-68.

[13] G. Neale, H. Hogan, and N. Sevdalis, “Misdiagnosis: analysis based on case record review with proposals aimed to improve diagnostic processes,” *Clinical Medicine*, vol. 11, no. 4, pp. 317–321, Aug. 2011, doi: 10.7861/clinmedicine.11-4-317.

[14] C. C. Boehme *et al.*, “Feasibility, diagnostic accuracy, and effectiveness of decentralised use of the Xpert MTB/RIF test for diagnosis of tuberculosis and multidrug resistance: a multicentre implementation study,” *The Lancet*, vol. 377, no. 9776, pp. 1495–1505, Apr. 2011, doi: 10.1016/S0140-6736(11)60438-8.

[15] J. F. Cohen, N. Bertille, R. Cohen, and M. Chalumeau, “Rapid antigen detection test for group A streptococcus in children with pharyngitis,” *Cochrane Database of Systematic Reviews*, vol. 2016, no. 7, Jul. 2016, doi: 10.1002/14651858.CD010502.pub2.

[16] S. T. Shulman *et al.*, “Clinical Practice Guideline for the Diagnosis and Management of Group A Streptococcal Pharyngitis: 2012 Update by the Infectious Diseases Society of America,” *Clinical Infectious Diseases*, vol. 55, no. 10, pp. e86–e102, Nov. 2012, doi: 10.1093/CID/CIS629.

[17] “International experts explore diagnostics for infectious diseases – Global Health Press.” Accessed: Jun. 11, 2025. [Online]. Available: https://id-ea.org/international-experts-explore-diagnostics-for-infectious-diseases/

[18] N. Petrić Howe and E. Bates, “Rapid sepsis test identifies bacteria that spark life-threatening infection,” *Nature*, 2024, doi: 10.1038/D41586-024-02462-X.

[19] G. D. Kaprou, I. Bergšpica, E. A. Alexa, A. Alvarez-Ordóñez, and M. Prieto, “Rapid Methods for Antimicrobial Resistance Diagnostics,” *Antibiotics*, vol. 10, no. 2, p. 209, Feb. 2021, doi: 10.3390/ANTIBIOTICS10020209.

[20] J. C. M. Wan *et al.*, “Liquid biopsies come of age: towards implementation of circulating tumour DNA,” *Nat Rev Cancer*, vol. 17, no. 4, pp. 223–238, Apr. 2017, doi: 10.1038/nrc.2017.7.

[21] E. Heitzer, I. S. Haque, C. E. S. Roberts, and M. R. Speicher, “Current and future perspectives of liquid biopsies in genomics-driven oncology,” *Nat Rev Genet*, vol. 20, no. 2, pp. 71–88, Feb. 2019, doi: 10.1038/s41576-018-0071-5.

[22] I. V Jani *et al.*, “Impact of Point-of-Care Birth Test-and-Treat on Clinical Outcomes Among Infants With HIV: A Cluster-Randomized Trial in Mozambique and Tanzania,” *Clinical Infectious Diseases*, vol. 80, no. 5, pp. 1114–1124, Jun. 2025, doi: 10.1093/CID/CIAE530.

[23] P. J. García *et al.*, “Rapid Syphilis Tests as Catalysts for Health Systems Strengthening: A Case Study from Peru,” *PLoS One*, vol. 8, no. 6, p. e66905, Jun. 2013, doi: 10.1371/journal.pone.0066905.

[24] R. L. Zimmern, “Testing challenges: evaluation of novel diagnostics and molecular biomarkers,” *Clinical Medicine*, vol. 9, no. 1, pp. 68–73, Feb. 2009, doi: 10.7861/clinmedicine.9-1-68.

[25] A. M. Caliendo *et al.*, “Better Tests, Better Care: Improved Diagnostics for Infectious Diseases,” *Clinical Infectious Diseases*, vol. 57, no. suppl 3, pp. S139–S170, Dec. 2013, doi: 10.1093/cid/cit578.

[26] G. D. Schiff, “Diagnostic Error in Medicine,” *Arch Intern Med*, vol. 169, no. 20, p. 1881, Nov. 2009, doi: 10.1001/archinternmed.2009.333.

[27] J. M. Miller *et al.*, “Clinical Infectious Diseases A Guide to Utilization of the Microbiology Laboratory for Diagnosis of Infectious Diseases • CID 2018:67 (15 September) • e1 for Microbiology a,” 2018, doi: 10.1093/cid/ciy381.

[28] A. M. Peri, A. Stewart, A. Hume, A. Irwin, and P. N. A. Harris, “New Microbiological Techniques for the Diagnosis of Bacterial Infections and Sepsis in ICU Including Point of Care,” *Curr Infect Dis Rep*, vol. 23, no. 8, pp. 1–11, Aug. 2021, doi: 10.1007/S11908-021-00755-0/METRICS.

[29] D. S. Mouliou, “Managing Viral Emerging Infectious Diseases via current Molecular Diagnostics in the Emergency Department: the Tricky Cases,” *Expert Rev Anti Infect Ther*, vol. 20, no. 9, pp. 1163–1169, Sep. 2022, doi: 10.1080/14787210.2022.2089653;WGROUP:STRING:PUBLICATION.

[30] T. A. O. (m Ison, N. Theodoropoulos And S Pergam, and S. Editors, “Molecular Diagnostic Advances in Transplant Infectious Diseases”, doi: 10.1007/s11908-019-0704-7.

[31] J. Chen, Z. Qin, and Z. Jia, “The application status of sequencing technology in global respiratory infectious disease diagnosis,” *Infection 2024 52:6*, vol. 52, no. 6, pp. 2169–2181, Aug. 2024, doi: 10.1007/S15010-024-02360-4.

[32] B. Gu, C. Zhuo, X. Xu, and K. El Bissati, “Editorial: Molecular diagnostics for infectious diseases: Novel approaches, clinical applications and future challenges,” *Front Microbiol*, vol. 14, p. 1153827, Mar. 2023, doi: 10.3389/FMICB.2023.1153827/BIBTEX.

[33] “Infectious Disease Diagnostics: Clinical Trials with Diagnostics CROs.” Accessed: Jun. 12, 2025. [Online]. Available: https://www.lindushealth.com/blog/infectious-disease-diagnostics

[34] Y. Zhang *et al.*, “Clinical application and evaluation of metagenomic next-generation sequencing in suspected adult central nervous system infection,” *J Transl Med*, vol. 18, no. 1, p. 199, May 2020, doi: 10.1186/S12967-020-02360-6.

[35] V. Turbé *et al.*, “Towards an ultra-rapid smartphone- connected test for infectious diseases,” *Sci Rep*, vol. 7, no. 1, p. 11971, Sep. 2017, doi: 10.1038/s41598-017-11887-6.

[36] A. Awad *et al.*, “Connected healthcare: Improving patient care using digital health technologies,” *Adv Drug Deliv Rev*, vol. 178, p. 113958, Nov. 2021, doi: 10.1016/j.addr.2021.113958.

[37] “Importance of Disease Surveillance in Public Health - SSG, LLC.” Accessed: Jun. 11, 2025. [Online]. Available: https://www.ssg-llc.com/the-importance-of-disease-surveillance-in-public-health/

[38] J. Y. Zhang, A. T. Bender, D. S. Boyle, P. K. Drain, and J. D. Posner, “Current state of commercial point-of-care nucleic acid tests for infectious diseases,” *Analyst*, vol. 146, no. 8, pp. 2449–2462, Apr. 2021, doi: 10.1039/D0AN01988G.

[39] P. Ulivi *et al.*, “Case Report: The Added Value of Liquid Biopsy in Advanced Colorectal Cancer From Clinical Case Experiences,” *Front Pharmacol*, vol. 12, Nov. 2021, doi: 10.3389/fphar.2021.745701.

[40] Q. D. Thomas *et al.*, “Case report: Liquid biopsy, the sooner the better?,” *Front Oncol*, vol. 12, Dec. 2022, doi: 10.3389/fonc.2022.1089108.

[41] J. Cunningham *et al.*, “A review of the WHO malaria rapid diagnostic test product testing programme (2008-2018): Performance, procurement and policy,” *Malar J*, vol. 18, no. 1, Dec. 2019, doi: 10.1186/S12936-019-3028-Z.

[42] H. Zhang, G. Fink, and J. Cohen, “Malaria Rapid Tests, Febrile Illness Management, and Child Mortality Across Sub-Saharan African Countries,” *JAMA*, vol. 332, no. 15, p. 1270, Oct. 2024, doi: 10.1001/jama.2024.12589.

[43] J. Wambani and P. Okoth, “Impact of Malaria Diagnostic Technologies on the Disease Burden in the Sub-Saharan Africa,” *J Trop Med*, vol. 2022, pp. 1–8, Mar. 2022, doi: 10.1155/2022/7324281.