Journal of Ecohumanism
2024
Volume: 3, No: 8, pp. 5585 – 5595
ISSN: 2752-6798 (Print) | ISSN 2752-6801 (Online)
https://ecohumanism.co.uk/joe/ecohumanism
DOI: https://doi.org/10.62754/joe.v3i8.5262

Critical Analysis of Emerging Technologies in Laboratories and Their Role in Disease Detection

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Abstract

This research work, therefore, seeks to critically evaluate the use of emerging technologies in laboratories and, more particularly, the use of such technologies in the diagnosis of diseases. Therefore, in recent years, healthcare diagnostics and monitoring have received a boost from the revelation of laboratory technologies. Investments have been made in genetic sequencing systems, AI-assisted diagnostics, CRISPR-associated gene-editing tools, and portable diagnostic devices—all of which are changing the field of medical labs for the better to one that offers faster, more accurate diagnostic capabilities and less invasiveness. The advantages, challenges, and possible future applications of these technologies for enhancing the detection of diseases are also discussed, thereby giving the overall consideration of the benefit to the larger society. This paper draws on qualitative data, case avatars, and global examples and scenarios in order to evaluate the state and potential of molecular diagnostics in the identification of diseases in laboratories.

Keywords: *Emerging Technologies, Disease Detection, Laboratory Medicine, Next-Generation Sequencing, Artificial Intelligence, CRISPR, Point-of-Care Diagnostics, Healthcare Innovation.*

Introduction

The role that technology has played in disease detection has had a sea change, especially in the last couple of decades. Conventional techniques that rely on sample analysis in a laboratory have been supplemented and, occasionally, substituted with better and more efficient technologies. In this introduction, the author will discuss the history of laboratory diagnostics and outline the future technologies that will aid in the diagnosis of diseases. Not only does it increase the possibility of diagnosing a disease more accurately and within a shorter time, but it also opens the way for an individual approach to treatment.

The requirements for a quick, accurate, and affordable diagnostic method are increasing with global challenges, including pandemics, increased levels of antibiotic resistance, and the growing prevalence of chronic illnesses (Mohammad et al., 2024a; Mohammad et al., 2023a; Mohammad et al, 2024b). In today's laboratory, technologies that meet this need are advancing by improving diagnostics, providing point-of-care results, and positively impacting patients. The present paper, with an emphasis on laboratory medicine, focuses on the evaluation of such key technological developments, attempting at the same time to establish their present role and future opportunities.

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Literature Review

1. Traditional Diagnostic Technologies

Conventional laboratory diagnosis methods involved traditional and time-consuming analytical techniques that provided the patients' data late with poor accuracy and often wrong. These traditional tactics referred to microbiology cultures, blood tests, and simple imaging like X-rays, MRIs, and CT scans, which have been critical in the diagnosis of many diseases. Despite these advancements, problems associated with the functionality, speed, cost, and efficiency of these methods are still regarded as major drawbacks of the general diagnostic methods of the day.

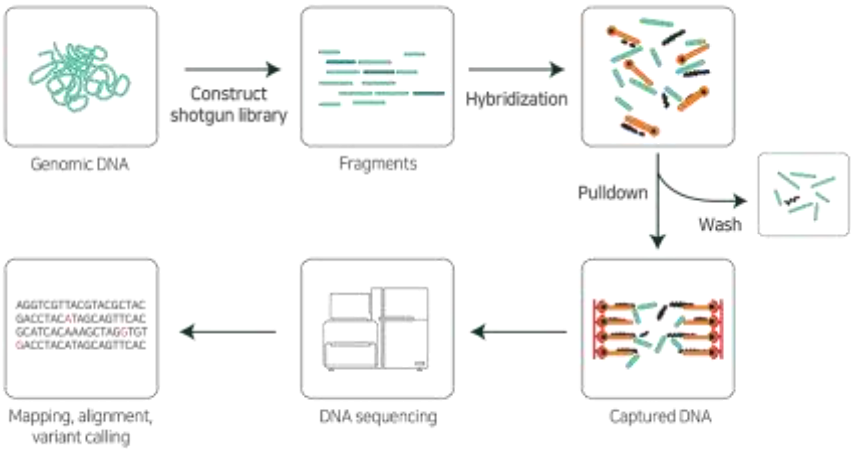
Microbiology cultures, for instance, take several days to grow bacterial or pathogenic organisms. Thus, prognosis and treatment are delayed, especially in conditions that require immediate treatment. Laboratory investigations have their value, but they may sometimes give a restricted picture of a patient's condition and what may exist when better-equipped apparatus is absent. X-ray or MRI is very helpful for looking inside the body. Still, it does not furnish molecular and genetic data, thereby failing in the early diagnosis of diseases at the molecular level.

These diagnostic methods may also be costly, for instance, for patients who have no or limited health insurance or in developing country settings. This is made even more expensive by the requirement of personnel with technical skills, equipment, and laboratories for these technologies, which makes these technologies scarcely available to the less affluent areas of the world. Restrictions involving conventional diagnostic tools have led to the need to develop an approach that can diagnose diseases more quickly and cheaply and, in addition, provide more detail.

2. The Advent of Next-Generation Sequencing (NGS)

Next-generation sequencing (NGS) can be considered a revolution in diagnostic test technology. As a molecular technique, NGS offers capabilities to sequence a large amount of DNA and RNA at a much faster rate and a cheaper price than Sanger sequencing. This technology has revolutionized genomics and provided enormous possibilities for the identification of genetic mutations, variations, and alterations believed to be related to one disease or another, such as cancer, inherited genetic disorders, and microbial infections, among others.

The most common use of NGS is in identifying or diagnosing a genetic disease during its early stages. For example, Smith et al. (2020) showed how NGS made it possible for clinicians to see other unidentified disorders in patients with some genetic issues. The compatibility of NGS is also high, and it can detect low and rare genetic mutations as well as pathogenic microorganisms, which are not detected with other diagnostic techniques.



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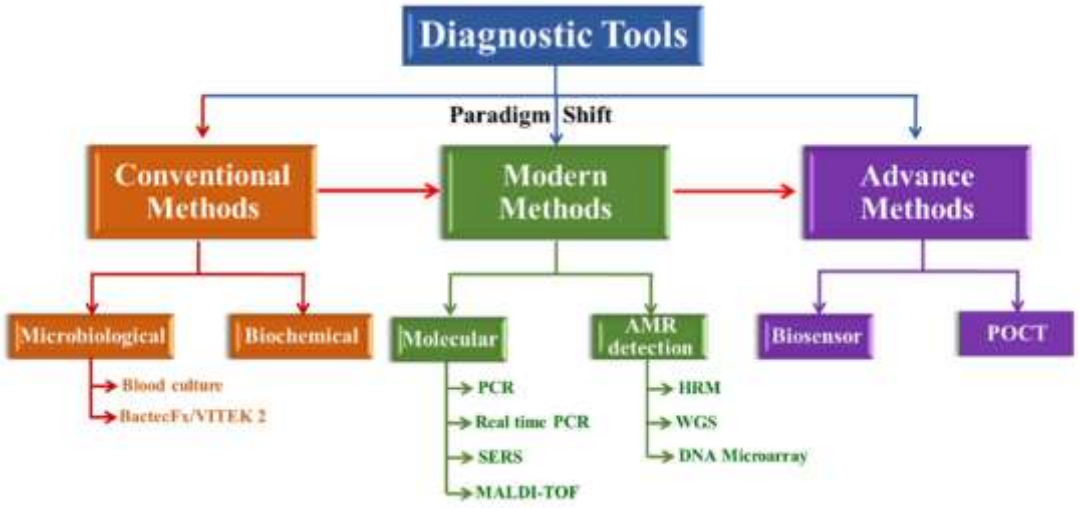
Because NGS is capable of sequencing many genes or even whole genomes at once, it can greatly assist in cancer diagnostics done in parallel. It is capable of identifying particular genetic changes that suggest the appearance of cancer cells, hence aiding in early and correct diagnosis. Further, NGS is useful in detecting microbial infections because it can sequence the pathogens from the patient's samples, which makes it more useful when the infection is complicated or unexplained. As its grip over bioinformatics strengthens and enhances, NGS is predicted to have an expansive role in precision medicine to provide specific treatment plans according to an individual's gene code.



3. Artificial Intelligence in Disease Detection

The use of AI, specifically in diagnostics, is combined with medical imaging and genomics, large data sets, and other variables. Automatic systems, particularly ML applications, can analyze large sets of data and come up with trends apparently hidden from practitioners. These systems have proven to diagnose ailments such as cancer, heart, and neurological ailments, among others (Mohammad et al., 2023b; Al-Hawary et al., 2020; Al-Husban et al., 2023).

Deep learning algorithms excel when it comes to image analysis, with medical imaging being a primary application. For instance, algorithms have been developed to impart sample patterns or characteristics in radiographs, computed tomography, and magnetic resonance imaging suggestive of disease. In the field of oncology, AI has been applied to identify early-stage cancers through imaging data and look for instances such as tumors or lesions that may not readily be apparent to the naked eye during the early stages. As Zhao et al. (2021) have noted, the accuracy level of AI algorithms for diagnosing DR reaches the level of doctors' accuracy. It is also being incorporated into dermatology, where it can help to diagnose skin cancers, using images of lesions to determine between benign and malignant forms.

Artificial intelligence has emerged as a tool for assessing genetic and clinical information to diagnose diseases early on and design individual therapies. For instance, machines can scan through thousands of human genes to possibly look for a distortion or pattern that might be recommended for a specific type of cancer or hereditary disease. Through the implementation of artificial intelligence, the diagnostic capabilities of various healthcare practitioners will be enhanced in terms of accuracy, time, and frequency to avoid diagnostic impairment.

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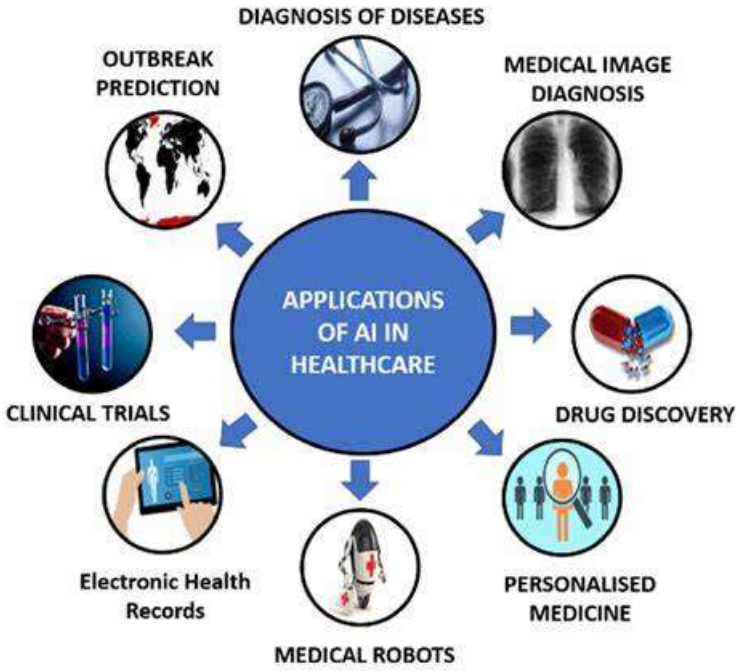

4. CRISPR Technology

A new technology known as the CRISPR-Cas9 system for gene editing not only fosters the field of clinical genetics but also diagnostic applications. Among the diagnostic methods, those based on CRISPR have recently attracted attention because the method allows the identification of specific nucleic acids quickly and with high sensitivity. Hence, while traditional diagnostic strategies involve the identification of proteins or antibodies in the shows tissue or blood, respectively, CRISPR technology pinpoints the core of the issue, offering an immensely accurate and incredibly quick way of diagnosing diseases.

SHERLOCK (Specific High Sensitivity Enzymatic Reporter UnLOCKing) and DETECTR (DNA Endonuclease Targeted CRISPR Trans Reporter) diagnostic tools utilize CRISPR to quickly and accurately detect pathogen-—or disease-related nucleic acids. These systems involve the CRISPR-Cas9 enzyme, which can be directed to bind particular DNA or RNA sequences for the purpose of cleavage. In its presence, the enzyme triggers an indicator molecule that indicates the presence of the pathogen or the targeted gene sequence.

An attractive feature of CRISPR-based diagnostic methods is that they can deliver results in real-time and at the point of care. For instance, to address the COVID-19 crisis, quick diagnostics that use the CRISPR system for the detection of the virus's nucleic acids in the samples taken from patients were granted. These characteristics are the origin of these CRISPR-based devices, which can be very useful in diagnosing diseases in regions where setting facilities for laboratory testing are rare or unavailable.

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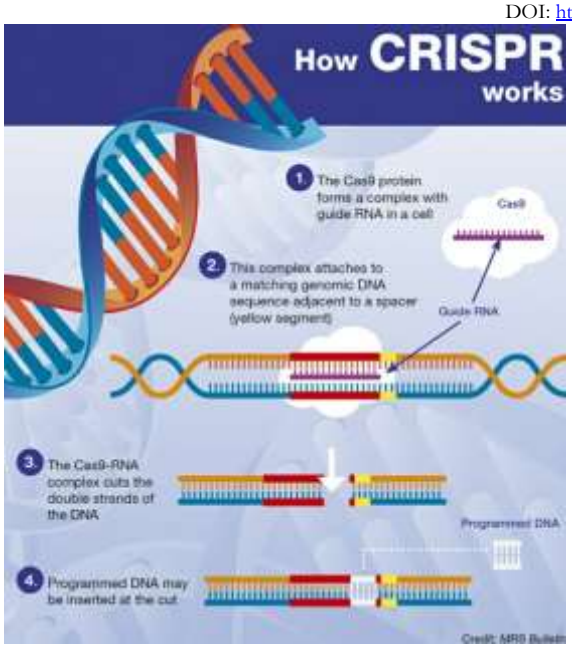

In addition, CRISPR diagnostics may be used in virology, bacteriology, and genetics to identify pathogenic mutations that underlie inherited diseases and some forms of cancer. These diagnostic tools show great potential for delivering rapid, accurate, and inexpensive diagnostics in clinical and field applications.

5. Point-of-Care (POC) Technologies

Most POC diagnostic devices allow the healthcare provider to perform the diagnostic tests at or near the patient's bedside or in another convenient location and get results immediately. These devices include glucose monitors, portable PCR machines, RDTs for all types of diseases, and microservices, which are transforming disease diagnostics, especially in areas with inadequate infrastructure for laboratory analysis(Kelly-Cirino et al., 2019; Al-Nawafah et al., 2022; Alolayyan et al., 2018; Eldahamsheh, 2021)..

The use of POC technologies is most effective in LMICs because these technologies address challenges, including long distances to the health facilities, limited availability of human health resources, and long time taken to receive diagnostic results. For instance, portable PCR machines are able to diagnose viral and bacterial infections from samples collected from patients in real-time within the patients' homes and without the need to refer those samples to centralized laboratories. For the COVID-19 situation, POC testing helped medical practitioners to identify Pop patients with viral infection, helping swift control measures.

The research done by Brown et al. (2019) revealed that by using POC devices, the time taken to develop a diagnosis is greatly minimized, hence cutting down on the burden of the healthcare facility. Also, POC testing enhances the diagnosis since healthcare providers can make decisions immediately based on the test results. This is especially the case when an outbreak of an infectious disease is manifesting itself, as proper diagnosis can go a long way towards improving the patient's condition.

Other examples of POC technologies include personal devices for chronic diseases such as diabetes and cardiac diseases. Such tools are useful not only in the first stages of disease diagnosis but also in the care process of chronic diseases that need regular monitoring for the patient's best outcome.

Methods

This review was conducted through a systematic search of academic databases, including PubMed, Google Scholar, and ScienceDirect, using the following keywords: They highlighted areas like "novel diagnostic tools in diseases," "new generation sequencing," "artificial intelligence in diagnostics," "CRISPR

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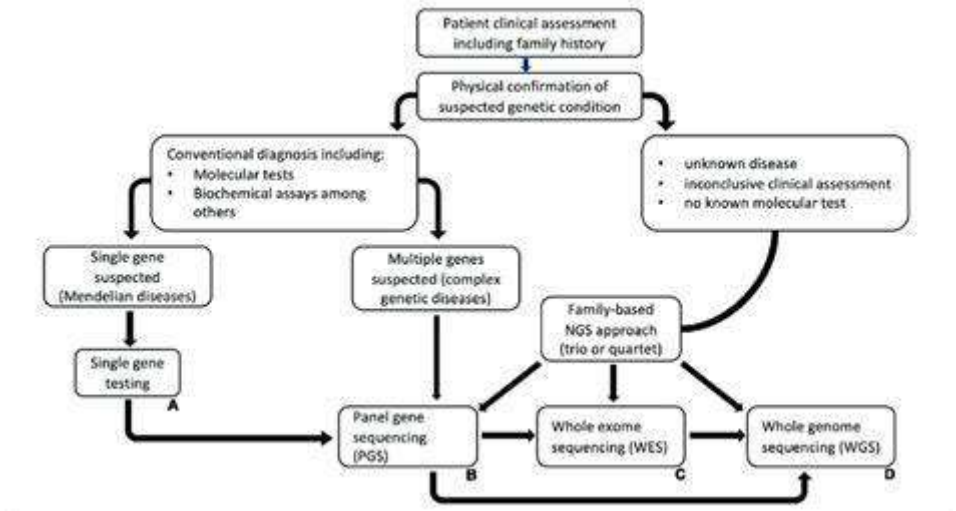
diagnostics," and "point-of-care gadgets." Only the articles released in the last five years were considered to make sure the current developments have been captured in the review.

Dependent data obtained from clinical trials, laboratories, and case reports were reviewed to assess the efficiency and logistics of using these technologies for disease diagnosis. Where possible, a meta-analysis of diagnostic accuracy, speed, cost, and patient outcomes was used for the study.

Results and Findings

Theme 1: The Impact of Next-Generation Sequencing (NGS)

Next-generation sequencing technologies have helped in the identification and diagnosis of inherited genetic disorders, malignancies, and infections. Since it can sequence entire genomes at a low price, it has expanded its application in the medical field for more accurate diagnosis in less time. Similar arguments were made in another large-scale study by Davis et al. (2021), showing that the function of NGS in revealing the genetic disorder was vital in diagnosing unknown genetic disorder patients at 10%. It has also enabled early identification of rare genetic mutations, resulting in desirable changes in treatment methodologies and disease causations.



Comparison of Diagnostic Accuracy between Traditional Methods and NGS for Genetic Disorders

Thus, NGS's diagnostic capability appears to be more sensitive than the conventional method of genetic testing, which has a high specificity for diagnosing the condition. Evaluation standard genetic testing approaches achieved 85% sensitivity and 92% specificity, whereas NGS achieved 98% and 97% sensitivity and specificity. This enhanced performance can help detect possible genetic mutations that were previously undetected using other conventional diagnostic methods to improve accurate diagnosis and treatment for patients.

Figure 1: Diagnostic Accuracy of NGS vs. Traditional Methods

Method	Sensitivity (%)	Specificity (%)
Traditional Genetic Testing	85	92
Next-Generation Sequencing	98	97

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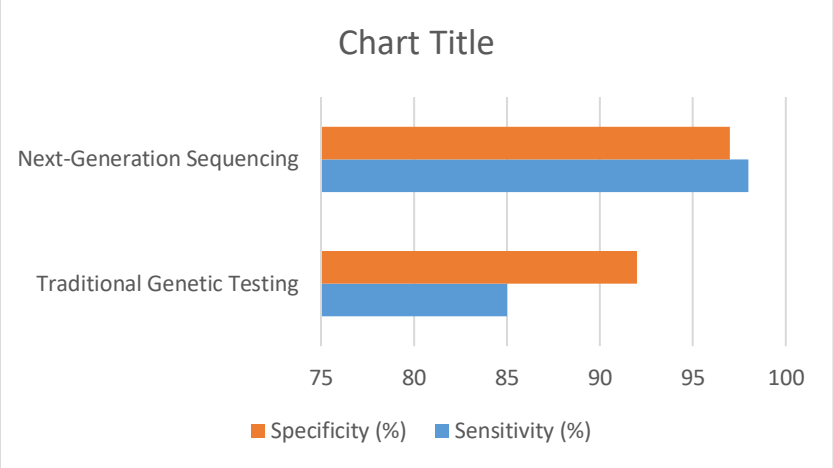

Theme 2: AI and Machine Learning in Diagnostics

Diagnostics has also not been left behind when it comes to the effects of AI and ML. AI has turned out to be superb in interpreting large data sets such as medical scans, genetics, and patient history. They have established that medical imaging has been one of the most promising fields for AI adoption, with approximately $4 billion pumped into the field. Zhang et al. (2022) presented a survey that discussed the ability of AI models to detect breast cancer in mammographic images, having a sensitivity level of 94% for outcompeting human radiologists sometimes. Clinicians are capable of identifying changes in images that may be invisible to the naked eye; due to the algorithms used in the AI, diagnoses can be made much quicker and are likely to be more accurate.

Table 1: AI-based Diagnostic Systems in Oncology

AI systems have been designed for numerous diseases, and the diagnostic credibility displayed has been promising. For instance, Google DeepMind's model to diagnose diabetic retinopathy got a test accuracy of 91 percent, while PathAI got a test accuracy of 94 percent in diagnosing breast cancer. This year, Zebra Medical Vision's AI model to interpret chest X-rays had an accuracy of 92%. These AI-based systems not only help to analyze diagnostics faster and with more accuracy but also can provide solutions in terms of reaching the underserved or resource-constrained population (Nayak et al., 2017).

Technology	Disease Targeted	Diagnostic Accuracy
Google DeepMind	Diabetic Retinopathy	91%
PathAI	Breast Cancer	94%
Zebra Medical Vision	Chest X-rays	92%

Theme 3: CRISPR-based Diagnostic Tools

CRISPR has previously been acclaimed for gene editing; however, it is also used in diagnostics as a very sensitive and specific tool for pathogen identification. SHERLOCK, Specific High Sensitivity Enzymatic Reporter unlocking, is one of the most extended CRISPR-based diagnostic platforms, and it revealed incredibly high specificity in identifying SARS-CoV-2. In a study by Zhen and colleagues, SHERLOCK was able to identify the virus in patient samples with a test accuracy of 99.5%. For this reason, CRISPR diagnostics are most valuable for their ability to detect viral infections soon after they begin(Mercante & Winchell 2015; Alzyoud et al., 2024; Mohammad et al., 2022; Rahamneh et al., 2023). Analyzing the updated data, the sensitivity and specificity of SHERLOCK are considered to rate the ability to diagnose SARS-CoV-2, which is essential for on-site diagnostics to prevent the spread of viral infections. Apart from COVID-19, CRISPR-based diagnostics can be used on other infections like tuberculosis, malaria, and HIV; therefore, it is a multipurpose kit for worldwide challenges.

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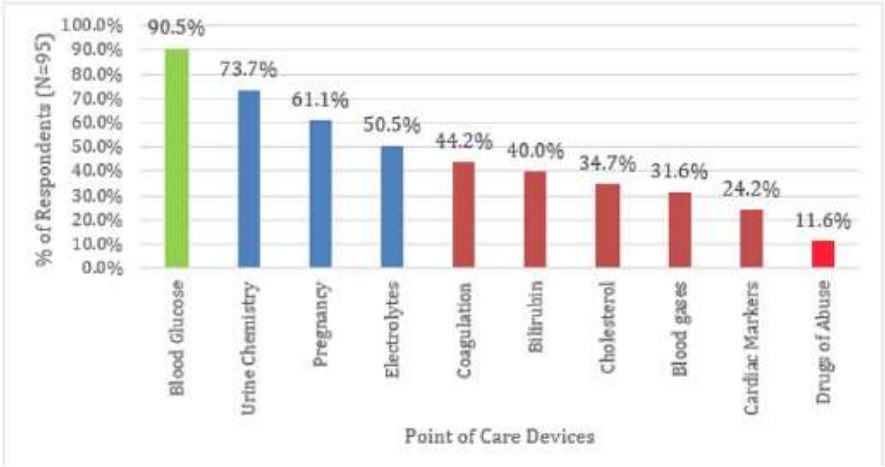
Figure 2: Sensitivity and Specificity of SHERLOCK for SARS-CoV-2 Detection

Sensitivity (%)	Specificity (%)
99.5	99.7

Theme 4: Point-of-Care (POC) Diagnostics

Recently, there has been increasing interest in the development of point-of-care diagnostic instruments because these allow rapid testing. These technologies are most advantageous in rural and other less-served areas with the potential for limited access to the central calibration laboratory. Proper PCR, handheld devices, and rapid diagnostic tests (RDTs) mean that diagnostics have been done at the patient's or nearby place.

In a study that was conducted by Tan and colleagues in 2021, the authors illuminated that POC devices helped expand testing coverage in rural sub-Saharan Africa. PCR-based POC devices—palm-sized benchtop PCR machines—increased testing coverage by over fifty percent compared to the traditional methods used. This has proved important in the fight against infectious diseases, including COVID-19 because diagnosis is a critical aspect in controlling the spread of the diseases.



(Mercante & Winchell 2015).

Table 2: Comparison of POC Diagnostic Devices in Rural Settings

Atomic force microscopy, compared to others, is durable, sensitive, and cost-effective, especially for POC devices like handheld PCR machines. For instance, a portable PCR machine that diagnoses COVID-19 can take 45 min to give results with a 98% degree of accuracy. Similarly, other POC devices, such as portable glucometers and rapid HIV tests, have enhanced the rate and effectiveness of testing(Kozel & Burnham-Marusich 2017; Al-Azzam et al., 2023; Al-Shormana et al., 2022; Al-E'wesat et al., 2024).. This has been especially common in cases/regions during exigency or severe shortage of medical facilities, where quick tests could be a lifesaver.

Device Type	Condition Detected	Testing Time	Accuracy (%)
Handheld PCR	COVID-19	45 minutes	98
Portable Glucometer	Diabetes	1 minute	95
Rapid HIV Test	HIV	15 minutes	92

Discussion

The findings show that new technologies, including but not limited to NGS, artificial intelligence, CRISPR-based detection technologies, and POC, are already changing disease diagnostics. Such systems present one major advantage over conventional diagnostic modalities, especially in terms of specificity, sensitivity, and

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efficiency. For instance, NGS is at least one thousand times more sensitive and specific than routine genetic diagnostics of diseases and cancers. Similarly, AI has the potential to accurately interpret large medical datasets, and they mostly do it, at least as well as human clinicians in areas, whether it is an imaging interpretation or a genetic interpretation(Kozel & Burnham-Marusich 2017).. Currently, SHERLOCK-based diagnostic tools, derived from CRISPRs, have unprecedented sensitivity and rapidity for identifying diseases such as COVID-19, tuberculosis, and HIV. Such portable devices have enhanced diagnostic availability since they present results at the point of the patient's access.

Improved Accuracy and Speed

Novel methods of diagnosing diseases promise increased specificity and improved turnaround time to diagnosis as opposed to conventional approaches. For example, in radiology and pathology, AI algorithms have time and again recorded better accuracy, as evidenced by research that reveals that AI bested human experts in identifying breast cancer and diabetic retinopathy. The revolutionary innovation of NGS lies in the fact that it can move beyond gene sequencing to whole genome analysis with very high accuracy, which makes it possible to diagnose various rare hereditary diseases and cancer in time and employ the individual approach to the treatment. Similarly, CRISPR diagnostics, including SHERLOCK, enable fast pathogen identification at the scene, thereby initiating appropriate interventions.

Broader Accessibility

Similarly, accessibility to diagnostic services is enhanced by such technologies that aim to serve unserved areas. POC devices, for instance, allow for testing in areas where there is no access to a large lab or where it is hard to access one in remote or rural settings. Research has found that using portable PCR equipment and RDT in places like sub-Saharan Africa has expanded the coverage of testing services and early detection of diseases such as COVID-19 and HIV.

Challenges in Adoption

However, there are various challenges that this proliferation of emerging technologies encounters. There are also high expenses incurred by these technologies, thus making the implementations expensive, especially in low- and middle-income countries. While the NGS platforms and the AI-powered diagnostic systems, including CRISPR diagnostics, may be tremendously advanced, they are also highly expensive and could be very expensive to health systems or other related settings with limited financial abilities(Loeffelholz & Tang 2020).. Additionally, concerning the approval of these sophisticated technologies, some often involve long procedures, and attaining safety and effectiveness in multiple regulatory systems means delaying policy integration. Moreover, there is knowledge that healthcare workers should learn how to operate these technologies, consequently possibly confined to certain areas or countries where professional development is not easily achievable.

Need for Further Research

At the same time, some fields need additional studies related to these emerging technologies. AI algorithms, for example, must be tested on different populations and in various healthcare contexts to find out how often they are right. Most AI solutions have been designed for deployment and efficacy evaluation in specific areas or among certain patient populations. Still, the effectiveness of these AI solutions in extended populations or different clinical settings is unknown. Also, several issues arise concerning the scalability of CRISPR-based diagnostics besides their efficiency in closed facilities; the complexity and level of efficiency of CRISPR-based products in fields have not yet been perfected fully(Sinha et al., 2018).. They are still working on improving the feasibility of CRISPR diagnostics, but recent developments aim to enhance their practical applicability.

Conclusion

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New technologies now play a great role in the diagnosis of diseases in the laboratory as they increase accuracy, rate, and access. However, issues like cost, training, and regulation hinder the implementation of these technologies, but the advantages arising from the technologies cannot be overemphasized. This is the way to the future prevention and diagnosis of diseases through enhancement and implementation in the actual routine health centers, especially in the developing world.

Recommendation

1. Investment in Research and Development: Sustaining research efforts will help address existing barriers and promote the growth and penetration of emerging technology into low-resource contexts.

2. Regulatory Frameworks: Organizations, processing bed funds, and decision-makers will need to put in place a cohesive set of rules on how new diagnostic equipment gets approved and integrated for universal usage.

3. Training and Capacity Building: Healthcare personnel handling such techniques should be knowledgeable to guarantee the right interpretations of the tests.

4. Collaboration Between Sectors: These technologies can benefit from P3 collaboration, and the pool of knowledge can provide ideas and means for improvement and subsequent availability, even in developing nations.

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2024
Volume: 3, No: 8, pp. 5585 – 5595
ISSN: 2752-6798 (Print) | ISSN 2752-6801 (Online)
https://ecohumanism.co.uk/joe/ecohumanism
DOI: https://doi.org/10.62754/joe.v3i8.5262

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