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AI Powered Bioinformatics - Expediting Diagnostic Testing: A Survey

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Abstract

Research has demonstrated the positive impact of artificial intelligence and Bioinformatics in the field of clinical diagnosis. The integration of AI methodologies into bioinformatics has opened new avenues for breakthroughs in genomics, proteomics, and personalized medicine. The document emphasizes the role of AI in early disease detection, improving patient outcomes, and enhancing healthcare systems by avoiding the need for expensive and time-consuming operations as illnesses worsen. The methodology section provides insights into the approach utilized, including the review of 30 articles from highly regarded journals about AI and bioinformatics that expedite diagnostic testing in the medical field. using survey to gather information and divide it into sub-sections focusing on diagnostic cancer diseases, COVID-19, and genetic and chronic diseases. The survey gathered 52 responses, and the results revealed significant agreements with the findings in the papers, particularly in the importance of developing novel biosensors and diagnostic tools for rapid and accessible detection of SARS-CoV-2, and the potential of AI in laboratory settings, pharmaceutical industry, and disease diagnosis. Overall, the document provides a comprehensive overview of the transformative role of AI in bioinformatics, emphasizing its potential to revolutionize disease diagnosis, treatment, and public health decision-making, while also addressing the challenges and opportunities associated with the integration of AI technologies in the healthcare industry. The rigorous methodology and alignment of survey results with the research findings validate the significance of AI-powered bioinformatics in expediting diagnostic testing and improving patient safety in healthcare.

Keywords: Artificial Intelligence, Diagnosing, AI Powered Bioinformatics.

1. Introduction

In recent years, the field of Bioinformatics has witnessed a remarkable transformation, driven in large part by the rapid advancements in Artificial Intelligence (AI) technologies. Bioinformatics, at its core, involves the use of computational methods to gather, store, analyze, and interpret biological data. This interdisciplinary field plays a crucial role in deciphering complex biological processes, understanding genetic variations, and accelerating drug discovery. Among many other applications, this integration of AI methodologies into Bioinformatics has opened up new avenues for breakthroughs in genomics, proteomics, and personalized medicine. Artificial intelligence (AI) has been widely used in the biomedical field for almost 20 years due to a number of variables, including improvements in computational techniques, the availability of high-performance computing hardware, and the creation of massive community-based databases. Artificial intelligence is highly proficient at processing massive amounts of complex data and extracting features from it that the human mind is incapable of recognizing. As a result, the biomedical field's research on AI applications has advanced and reached performance levels comparable to those of human professionals. AI will also provide human professionals with more knowledge for decision-making and become a vital part of the medical team [7]. The use of AI in healthcare is growing and permeating every aspect of medicine and daily life. Examples of this include diagnostic algorithms and smart health trackers. Artificial intelligence (AI) programs designed to assist clinicians in diagnosis, treatment selection, and outcome prediction are considered to be at the forefront of medical AI development. These systems include evolutionary computation, fuzzy expert systems, hybrid intelligent systems, and artificial neural networks (ANN) [8]. Technology has had a tremendous impact on the healthcare industry. and early disease detection is a major priority. This is critical and can benefit the entire healthcare system, in addition to helping specific individuals. Early disease detection is similar to a road map for improved patient outcomes. and the therapies frequently function better, are safer, and are simpler. Additionally, it benefits healthcare systems by avoiding the need for expensive and time-consuming operations as illnesses worsen [5]. Artificial intelligence and bioinformatics techniques are essential components of the effective response to the new Corona illness. As a result, AI can be extremely helpful in guaranteeing that policies, management, and resource allocation result in long-term solutions for the area's most negatively affected by the COVID-19 epidemic [1]. Biopharmaceutical and artificial intelligence (AI) have found useful uses in many therapeutic areas, especially in supporting the diagnosis and prevention of disease. Their integration has made progress possible in some healthcare domains, including the creation of preventive strategies and more rapid and precise disease identification. The use of various AI-derived technologies was critical in diagnosis, treatment exploration, and improving public health decision-making during the pandemic's difficult period when healthcare resources were scarce. This increased the pandemic's potential to be fought. Numerous AI-based methods that include the COVID-19 diagnostic tools have been developed. Large volumes of data are gathered using quick, consistent processes and clever algorithms used by AI. This enables the software to automatically learn from options or trends within the knowledge base and make the greatest use of the resources available for the population. AI techniques and tools automate the process of determining the underlying reason and frequently assist policymakers in creating appropriate rules and regulations. The history, dissemination, treatment, and management procedures of viruses can be swiftly uncovered by AI text and data processing techniques [3]. Despite advances in research and treatment, oral cancer (OC), one of the most prevalent types of head and neck cancer, still has the lowest global survival rates. In the biomedical sector, the prognosis for OC has not changed much in recent years, which continues to be a difficulty. Artificial intelligence (AI) has advanced quickly in the field of oncology, with some noteworthy achievements recently announced. The use of deep learning algorithms has advanced the field of oncology significantly. By helping pathologists accurately classify cancer into several categories, these intelligent tools enable the oncology team to plan a therapy module, which lessens operational workload and improves disease management. Additionally, deep learning models enable medical professionals to categorize patients into various risk groups in order to choose the best course of action [23]. Currently, the most demanding area of computer-aided diagnosis and treatment research is the development of genomic technologies for smart diagnosis and therapeutics for diverse diseases. Technological advances in artificial intelligence and machine learning have the potential to detect problems facing the healthcare sector. Future diseases, including diabetes, Alzheimer's, and cancer, may be predicted thanks to genomics. Innovations in machine learning have spurred new areas of computational biology and accelerated the pace of biomedical informatics research. Classification models are crucial in many sectors because, in addition, understanding gene interactions has led to the development of more precise models that can successfully identify patterns in enormous amounts of data. Recurrent neural network models have a memory that enables them to assimilate genetic data and swiftly recall information from past cycles [24].

The aim of the paper is to summarize the relevant literature and evaluate the potential of artificial intelligence (AI) to improve patient safety in healthcare. The review aims to identify studies that describe the application of AI for the prediction, prevention, or early detection of adverse events in each cancer disease, COVID-19, and genetic and chronic diseases. Additionally, the paper aims to consider the findings in the context of incidence, cost, and preventability to make projections about the likelihood of AI improving safety in healthcare.

This paper employs a mixed-methods approach, intertwining quantitative analysis for numerical data and qualitative exploration for rich contextual insights, to comprehensively examine AI-POWERED BIOINFORMATICS: EXPEDITING DIAGNOSTIC TESTING from multiple dimensions. Accordingly, this paper was divided into five sections, including the literature review, methodology, results, discussion, and lastly, the conclusion section.

2. Literature Review

This section discusses related work in the field of AI-powered bioinformatics to expedite the diagnosis of diseases.

2.1. In COVID-19 Diagnostics

Pillai and Kumar utilized big data analysis and various AI techniques to curb the unplanned spread of the COVID-19 pandemic [1]. The study employed regression

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models, machine learning, and deep learning on datasets comprising social media conversations, digital health records, contact-tracing mobile applications, robotic platforms, pictures, and thermal sensors. The research highlights AI's role in emulating human intellect to screen, evaluate, predict, and track current and potential future patients, thus demonstrating the potential of AI to improve healthcare facilities and mitigate the economic impact of viruses on society. Pandit and Banday et al. developed a deep learning model to automatically identify COVID-19 from chest radiographs [2]. The study employed the pre-trained VGG-16 model for classification tasks and utilized transfer learning with fine-tuning on a relatively small dataset of chest X-rays. Data augmentation techniques were also employed to enhance the dataset. The model demonstrated promising results in COVID-19 identification, with high accuracy rates observed in preliminary testing. Wang and Chavda et al. utilized bioinformatics techniques to decode the genome structure of the SARS-CoV-2 virus [3]. Next-generation sequencing and computer-aided drug design were employed to analyze evolutionary relationships, sequencing errors, and potential treatment options against SARS-CoV-2 genes. The study highlights the role of AI and bioinformatics in enhancing our understanding of the virus's pathophysiology, treatment options, and mechanisms of increased host immune response to antibiotic resistance. Alshazly et al. utilized advanced deep network architectures and transfer learning for diagnosing COVID-19 from chest CT images [11]. They achieved superior performance on the SARS-CoV-2 CT-scan and COVID19-CT datasets compared to prior studies, with high accuracy, precision, sensitivity, specificity, and F1-score values. Visualization techniques provided visual explanations for predictions, enhancing understanding of COVID-19-associated regions. Yadav et al. explored the use of biomarker-based electrochemical immunosensors in the diagnosis and management of COVID-19 [12]. These sensors offer a sensitive, selective, and low-cost method for detecting specific biomarkers associated with the disease, facilitating rapid and non-invasive diagnosis. Hossain et al. discussed the significant impact of AI, particularly machine learning, in exploring and developing diagnostic tools and biosensors for COVID-19 [14]. AI has accelerated analysis, increased accuracy, and enabled the development of intelligent global networks for real-time data analytics, improving resilience to testing errors and facilitating early detection and management of the disease. Behl et al. discussed the role of bioinformatics in accelerating the drug discovery process and managing the COVID-19 pandemic [18]. Bioinformatics methods were highlighted for information gathering, preliminary investigations, sequence analysis, computational modeling, and prediction of sequence properties. The review emphasized the positive outcomes of bioinformatics in identifying drug targets, validating targets, and managing the COVID-19 pandemic through next-generation sequencing, genome-wide association studies, and computer-aided drug design. Mohammed conducted an in-depth analysis of AI's use in COVID-19 research and vaccine development [20]. Louati and Lahyani et al. introduced a novel method combining genetic algorithms and neural architecture search to enhance CNN design for more precise detection of COVID-19 infections while minimizing computing load [21]. Their study utilized a dataset of 1319 COVID-negative and 1184 COVID-positive chest X-ray images. Results demonstrated improved accuracy in COVID-19 diagnosis, setting a benchmark for efficient healthcare solutions with reduced environmental impact. The analysis covered AI applications in rapid data analysis, research surfacing models, protein structure prediction, collaborative efforts, customer communication enhancement, viral spread mapping, research acceleration, and vaccine ingredient proposal. AI's contributions to understanding the virus's spread, diagnosing infections, developing treatments, and predicting

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vaccine efficacy were highlighted. Kim and Choi et al. developed a diagnosis model integrated into an automated triage pipeline for assessing chest radiographs for COVID-19 pneumonia [25]. The model achieved a 95% diagnosis accuracy, highlighting its potential as a valuable tool for efficient and accurate diagnosis in clinical settings. Calidonio and Hamad-Schifferli utilized machine learning (ML) to optimize a paper immunoassay for detecting COVID-19 antibodies [26]. They achieved 100% test accuracy through iterative modification of test conditions using linear discriminant analysis (LDA) and ML. The optimized test was trained to distinguish nine distinct antibody profiles, including vaccinated and infected profiles.

2.2. In Cancer Diagnostics

Rabaan and Bakhrebah et al. conducted a systematic review of AI approaches, including machine learning and deep learning algorithms, for clinical data analysis in prostate cancer diagnosis [4]. The study utilized datasets such as prostate-specific antigen (PSA), MRI-guided biopsies, genetic biomarkers, and Gleason grading for patient monitoring, risk assessment, and diagnosis. AI-powered technology showed potential to simplify and accelerate prostate cancer diagnosis and treatment by creating complex associations between prediction variables and other known parameters. Deepa and Arunkumar et al. reviewed deep learning techniques, including convolutional neural networks (CNNs) and recurrent neural networks (RNNs), for analyzing data and images in cancer diagnosis [5]. The study emphasizes AI's advantage over conventional techniques in early detection of skin, lung, and breast cancers, leading to quicker and more accurate discovery of abnormalities. The research suggests that AI technology holds promise for improving cancer diagnosis, enabling early treatments, and ultimately extending human lifespan.

Elhadary and Elshoeibi et al. conducted a critical assessment of machine learning applications in CLL diagnosis, utilizing various data sources including blood smears, flow cytometry, histological pictures, and genetic information [6]. A targeted panel of cancer-associated genes was also analyzed using next-generation sequencing. The literature search yielded 169 articles, of which 14 met the inclusion criteria after screening. Results showed that most machine learning models performed well in predicting CLL diagnosis, with top models achieving high AUROC, sensitivity, and specificity. For instance, one model achieved an AUROC of 99.7%, sensitivity of 96.4%, and specificity of 98.8% in separating CLL from healthy cases. Shao and Dai et al. surveyed AI processes in the biomedical field and examined strategies and uses of AI in cancer clinical research, categorized by types of data such as medical records, cancer genomes, radiographic imaging, pharmacological information, and biomedical literature [7]. They highlighted the establishment of centralized, publicly accessible knowledge bases like TCGA, providing clinical and molecular data for researchers and clinicians. The study anticipates that AI will play a crucial role in accelerating cancer diagnosis, treatment, and possibly even finding a cure. Moreover, they anticipate broader accessibility of AI technology in cancer treatment, aiming to increase treatment responses, reduce side effects, and improve survival rates. Khanagar and Alkadi et al. conducted a systematic review evaluating AI's role in histopathological image-based diagnosis, classification, and prognosis of oral cancer [23]. The review highlighted the high accuracy and specificity of AI models in diagnosing oral cancer, outperforming current clinical approaches and enhancing diagnostic precision. Guo et al. conducted bioinformatics analysis in esophageal adenocarcinoma (EAC) research [27]. They employed methods such as Weighted Gene Co-Expression Network Analysis (WGCNA), Gene Ontology (GO) and Pathway Enrichment Analysis, and Protein-

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Protein Interaction (PPI) Network Construction to identify key genes associated with EAC. The study also developed a risk score prognostic model based on the identified genes, achieving significant predictive accuracy for patient survival.

2.3. In Genome Interpretation Diagnostics

Feleke et al. employed Particle Display (PD) and Machine Learning (ML) models to predict high-affinity DNA aptamers for diagnostic and therapeutic purposes [13]. ML-guided mutation and experimental screening facilitated the discovery of high-performing aptamers, demonstrating the potential of combining ML and physical approaches in biomolecule design. Kumar and Sharma highlighted the contribution of deep learning in bioinformatics, particularly in areas such as image recognition, genomics, and medical imaging [15]. Deep learning algorithms, inspired by the human brain, analyze data, create patterns, and improve methodologies with experience, leading to more accurate outcomes and better understanding of complex biological processes. De La Vega et al. utilized an AI-based clinical decision support system called Fabric GEM to expedite genome interpretation for rare genetic diseases [16]. The study integrated predictive methods with the growing knowledge of genetic diseases, employing AI methods for variant prioritization and disease gene identification. The use of AI facilitated improved diagnostic performance and the identification of new diagnoses in previously unsolved cases. The Department of Thoracic Surgery at Qilu Hospital, Shandong University, China, conducted a study to identify specific genes and potential pathways associated with EAC [17]. The study utilized bioinformatics methods for data acquisition, preprocessing, identification of differentially expressed genes (DEGs), weighted correlation network analysis (WGCNA), gene ontology and pathway analysis, protein-protein interaction (PPI) network analysis, and establishment of a

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prognostic model. Key findings included the identification of genes with diagnostic and therapeutic potential for EAC, as well as the development of a prognostic model. Srinivasu and Shafi et al. utilized gene sequences to predict type 2 diabetes, employing neural networks with GRU, LSTM, and RNN components [24]. Their study demonstrated fair accuracy in predicting future illnesses, suggesting the model's potential applicability in practical settings. Zhao et al. evaluated computational tools like Phen2Gene and Phenolyzer for gene prediction and prioritization in single-gene diseases [28]. Phen2Gene demonstrated superior accuracy in identifying causal genes compared to Phenolyzer. The study highlighted the potential of computational tools in assisting targeted sequencing to identify disease-causing variants. Shu et al. discussed various genomic technologies such as short-read genome sequencing (SR-GS), long-read genome sequencing (LRS), and optical genome mapping (OGM) for identifying causes of rare diseases and unraveling complex genetic mechanisms [30]. The study highlighted clinical diagnosis-driven genetic analysis strategies for prioritizing genetic analysis and expediting diagnostic success in rare diseases.

2.4. Role of Bioinformatics in Healthcare in General

Taran Undru and Utkarsha Uday et al. conducted a review of research papers focusing on the application of deep learning in clinical microbiology [8]. Bates et al. conducted a scoping review on the potential of AI to improve patient safety in healthcare [19]. The review mapped AI applications to various harm domains and highlighted examples of AI being used for prediction, prevention, or early detection of adverse events. The study emphasized the role of AI in enhancing patient safety through swift identification of patterns, research surfacing models, protein structure prediction, collaborative efforts, customer communication enhancement, and viral spread mapping. Ijaz and Nabeel et al. conducted a survey analyzing the use of cough sounds and AI-based models to detect and diagnose respiratory disorders early [22]. Their findings suggest AI's potential in creating clinical decision support and diagnostic tools, aiding healthcare professionals in traditional medical procedures. Jamshidi et al. integrated edge computing (EC), deep learning (DL), and the Internet of Medical Things (IoMT) to improve efficiency and productivity in the pharmaceutical industry [29]. Their approach aimed to reduce the risk of COVID-19 infection and advance drug discovery and molecular diagnostics using innovative technologies like multicolor multicycle molecular profiling (M3P).

2.5. In Genetic Chronic Diseases Diagnostics

Lily Zhuhadar and Miltiadis D. Lytras developed an open-source, cloud-based framework for generating highly accurate prediction classification models [9]. Using the Pima Indian Diabetes dataset, they applied AutoML to identify the top nine models for diabetes diagnosis prediction. The generalized linear model showed remarkable predictive power in diagnosing diabetes. Their research highlights the effectiveness of AutoML advancements in identifying risk factors, optimizing treatment plans, and ultimately improving patient outcomes. Emily Kogan and Eva-Maria Didden et al. They created a machine learning model called XGBoost, a gradient boosting model, using data from a patient-level electronic health record (EHR) database in the United States [10]. By incorporating various patient characteristics and health-related data into the model, they aimed to predict patients at risk of pulmonary hypertension (PH) early on. Their initial findings suggest that the ML model achieved a high area under the receiver operating characteristic curve (AUROC), indicating its potential to facilitate early prediction of PH risk. This could lead to quicker referrals to specialists and improved patient outcomes in primary care settings. They found that huge datasets in clinical microbiology can be leveraged to construct AI diagnoses, and deep learning algorithms are increasingly being applied across various medical vocations. These algorithms can enhance diagnosis sensitivity and accuracy while reducing turnaround times.

3. Methodology

After reviewing 30 articles from highly regarded journals about artificial intelligence and bioinformatics that expedite diagnostic testing in the medical field. In this paper, we used the survey to collect information and analyze the results based on this information. And then we divided it into sub-sections: diagnostic COVID-19, cancer diseases, and genetic and chronic diseases. We wrote 13 questions to study the efficiency and effectiveness of artificial intelligence and bioinformatics techniques to accelerate the diagnosis of Corona, cancer, and genetic and chronic diseases and develop the necessary treatments and vaccines. And discover the strengths and weaknesses of these technologies and their impact on the sustainability of the medical field in general, which are:

Table 1. Questions to study the efficiency and effectiveness of AI and bioinformatics techniques.

Section	Question
COVID-19	The main challenges experienced by laboratory professionals in meeting the sudden and urgent testing demands for COVID-19?
	How important do you think the development of novel biosensors and diagnostic tools is for rapid and accessible detection of SARS-CoV-2?
	Do you think that the combination of artificial intelligence and bioinformation helped to decipher SARS-CoV-2 structure of the Coronavirus?
Genome Interpretation	What do you think about using deep learning algorithms to extract properties from DNA sequences, that help in the interpretation of genetic variation and the development of genome-based treatments?
General question	Do you think Artificial intelligence helped in laboratory settings? By applied to predict, prevent, and detect adverse events such as healthcare.
	Do you think that artificial intelligence helped the laboratory generate a large amount of data, which contributed to the field of the pharmaceutical industry in terms of (drug discovery, development, manufacturing, and marketing)?
	The most challenges encountered in training machine learning models using large sample pools in the laboratory, and how did these challenges affect the evaluation and effectiveness of the models?
Cancer	From your point of view, do you think that applying machine learning models has helped diagnose leukemia through analysis of blood smears, flow cytometry, and genetic information?
	Do you think the use of machine learning algorithms has effectively contributed to the early diagnosis of prostate cancer using genetic biomarkers?
	Is artificial intelligence highly skilled at processing huge amounts of complex data in the biomedical field and extracting features from it that the human mind cannot recognize?
Chronic diseases	How have AI techniques, such as machine learning, contributed to the diagnosis of diabetes patients through the analysis of blood glucose levels and calculate body mass?
	Has the use of intelligence contributed to determining the appropriate dose of insulin to treat diabetics?
	Do you agree that the machine learning model performed well in early detection of pulmonary hypertension and helped improve patient outcomes?

4. Results

After we collected 52 responses table 2 showed the result of responses by percentage.

Table 2. Show The Results

Questions	Answers	Percentage
The main challenges experienced by laboratory professionals in meeting the sudden and urgent testing demands for covid-19?	Limited availability of testing supplies and equipment Overwhelming.	25%
	Lack of volume of samples to process.	25%
	Shortage of trained personnel to conduct tests.	50%
Do you think artificial intelligence helped in laboratory settings? By applied to predict, prevent, and detect adverse events such as healthcare.	Yes, he contributed effectively and clearly.	82.7%
	I don't know, we are still working in traditional ways.	3.8%
	No, I think the results are not 100% correct.	13.5%
Do you think that artificial intelligence helped the laboratory generate a large amount of data, which contributed to the field of the pharmaceutical industry in terms of (drug discovery, development, manufacturing, and marketing)?	Yes, he has contributed to identifying potential drug candidates and predicting their effectiveness and safety.	88.5%
	Sometimes, some fields lack good databases to collect information.	11.5%

How important do you think the development of novel biosensors and diagnostic tools is for rapid and accessible detection of sars-cov-2?	Helps allocate resources to areas with higher infection rates.	17.3%
	Early identification of infected individuals, leading to immediate isolation and treatment.	67.3%
	Smartphone-based platforms provide instant and flexible health information, creating a more efficient system.	15.4
Do you think that the combination of artificial intelligence and bioinformation helped to decipher the sars-cov-2 structure of the corona virus?	Yes, the process was greatly facilitated.	96.2%
	No, it did not help decipher the SARS-CoV-2 structure.	3.8%
The most challenges encountered in training machine learning models using large sample pools in the laboratory, and how did these challenges affect the evaluation and effectiveness of the models?	The small number of positive samples creates high variation in results.	13.5%
	Difficulty in accessing information easily.	38.5%
	Lack of sufficient experience and training.	48.1%
What do you think about using deep learning algorithms to extract properties from DNA sequences, that helps in the interpretation of genetic variation and the development of genome-based treatments?	Rapid detection of hereditary diseases by analyzing hereditary diseases in the family using DNA.	88.5%
	Genomic data is highly sensitive, and scientists need to navigate ethical and privacy concerns when using deep learning for genomic analysis.	11.5%

From your point of view, do you think that applying machine learning models has helped diagnose leukemia through analysis of blood smears, flow cytometry, and genetic information?	Yes, I agree. No, I do not agree.	94.2% 5.8%
Do you think the use of machine learning algorithms has effectively contributed to the early diagnosis of prostate cancer using genetic biomarkers?	Yes, it contributed to an early diagnosis. No, it did not contribute to an early diagnosis.	94.2% 5.8%
Is artificial intelligence highly skilled at processing huge amounts of complex data in the biomedical field and extracting features from it that the human mind cannot recognize?	Yes, AI applications have advanced and reached performance levels comparable to those of human professionals. No, human professionals perform better than artificial intelligence.	86.5% 13.5%
How have AI techniques, such as machine learning, contributed to the diagnosis of diabetes patients through the analysis of blood glucose levels and calculate body mass?	Helped in simplifying the process of creating predictive models of diabetes diagnosis. Contributes to sustainability in healthcare through its resource-efficient. None of the above.	67.3% 32.7 % 0 %
Has the use of artificial intelligence contributed to determining the appropriate dose of insulin to treat diabetics?	Yes, this leads to maximizing treatment plans and improving patient outcomes. No, it does not contribute to improving treatment plans.	92.3% 7.7%

Do you agree that the machine learning model performed well in early detection of pulmonary hypertension and helped improve patient outcomes?	Yes, help in the early predictive diagnosis of heart failure, shortness of breath, and atrial fibrillation.	90.4
	No, it does not have the ability to diagnose pulmonary hypertension.	9.6

5. Discussion

The results of the survey showed great agreement with the results of the papers.

5.1. In Corona, the Results Appeared

Artificial intelligence and bioinformation greatly facilitated the decipherment of the sars-cov-2 structure of the Coronavirus. This indicates the efficiency and effectiveness of artificial intelligence and bioinformation in quickly diagnosing the Coronavirus, lessening the workload for physicians, and helping in the development of the necessary treatments and vaccines. AI serves as a cutting-edge weapon in the battle against pandemics and stopping their spread.

5.2. In Cancer, the Results Appeared

The results demonstrated the role of artificial intelligence and its effective contribution to the rapid diagnosis of cancers, such as machine learning algorithms that have effectively contributed to the early diagnosis of prostate cancer using genetic biomarkers. And machine learning models have helped diagnose leukemia through analysis of blood smears, flow cytometry, and genetic information. So, artificial intelligence aids therapeutic decision-making, outcome prediction, reducing costs, and the more human-like problem-solving of complicated issues. Because it is highly skilled at processing huge amounts of complex data in the biomedical field and extracting features from it that the human mind cannot recognize. Finally, AI has enormous potential to bring about a paradigm shift in the field of healthcare.

5.3. In Genome Interpretation, the Results Appeared

Deep learning algorithms help to extract properties from DNA sequences, which helps in the interpretation of genetic variation and the development of genome-based treatments Which help in detecting genetic diseases faster.

5.4. In General Question, the Results Appeared

It was repeatedly mentioned that the lack of training and experience in using machine learning tools using large samples leads us to the conclusion that the Lack of training is one of the biggest problems facing laboratory specialists and is one of the weaknesses that must be looked into. In the field of healthcare and the pharmaceutical industry, the response was positive that artificial intelligence in discovering diseases helped in the pharmaceutical industry.

5.5. In Genetic and Chronic Diseases, the Results Appeared

Machine learning has contributed to the diagnosis of chronic diseases such as pulmonary hypertension and diabetes. This helps in the early predictive diagnosis of heart failure, shortness of breath, and atrial fibrillation and the diagnosis of diabetes through the analysis of blood glucose levels and the calculation of body mass. Also, determine the appropriate dose of insulin for effective treatment. Therefore, machine learning can identify risk factors, maximize treatment plans, and improve patient outcomes. And because it simplifies the process of creating predictive models, machine learning contributes to sustainability in healthcare through its resource-efficient and less-invasive approach.

5.6. Strengths and Weaknesses

1. The review by Sini V. Pillai and Ranjith S. Kumar (2021) underscores the significant contribution of data-driven artificial intelligence (AI) to pandemic management, highlighting its potential to alleviate the burden on physicians and enhance various aspects of COVID-19 management in the public sphere.
2. M.K. Pandit and S.A. Banday et al. (2021) present an automatic detection system for COVID-19 from chest radiographs using deep learning, offering a non-contact, automated testing solution. However, the limited datasets for COVID-19 detection may affect the accuracy of the model.
3. Jiao Wang and Vivek Chavda et al. (2023) advocate for the amalgamation of bioinformatics and AI in COVID-19 management, emphasizing its potential to expedite research and treatment development. Nevertheless, challenges such as data quality, availability, privacy concerns, and model interpretability persist.
4. Ali A. Rabaan and Muhammed Bakhrebah et al. (2022) discuss AI's role in clinical diagnosis and treatment of prostate cancer, highlighting its potential for detection and improving treatment outcomes. However, concerns regarding overlooking real positive cases and the necessity of human expertise in detection remain.
5. R. Deepa and S. Arunkumar et al. (2023) propose AI-driven early cancer detection combined with genetic data to enhance patient outcomes. Challenges such as algorithm impartiality, data security, and ethical considerations are acknowledged.
6. Mohamed Elhadary and Amgad Elshoeibi et al. (2023) examine machine learning's potential in chronic lymphocytic leukemia diagnosis, emphasizing its

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ability to expedite the process and increase efficiency. However, challenges related to information gaps, model validation, and ethical considerations are highlighted.

7. Dan Shao and Yinfei Dai et al. (2021) explore AI's impact on cancer research, acknowledging its potential to improve outcomes but noting challenges such as data hunger, model opacity, and the gap between data science and healthcare.
8. Taran Undru and Utkarsha Uday et al. (2022) discuss AI's integration in clinical and laboratory diagnosis, emphasizing its role in rapid diagnosis and therapeutic decision-making. However, challenges including legal responsibility, data processing, and bias are identified.
9. Lily Zhuhadar and Miltiadis D. Lytras (2023) review the application of AutoML techniques in diabetes diagnosis, highlighting its potential benefits in expediting diagnosis and treatment. Concerns about algorithmic bias, data privacy, and model interpretability are raised.
10. Emily Kogan and Eva-Maria Didden et al. (2023) present a machine learning approach to identifying patients with pulmonary hypertension using real-world electronic health records, showcasing improved diagnosis and patient outcomes. Challenges such as code-based algorithms and database limitations are noted.
11. The study by H. Alshazly, C. Linse, E. Barth, and T. Martinet (2021) achieved state-of-the-art performance in COVID-19 detection using deep learning and CT scans. However, it falls short in accurately identifying and highlighting relevant regions associated with COVID-19 in chest CT scans.

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12. A.K. Yadav, D. Verma, A. Kumar et al. (2021) present perspectives on biomarker-based electrochemical immunosensors, AI, and the Internet of Medical Things for COVID-19 diagnosis and management. While they offer high sensitivity and selectivity in biomarker detection, limitations include scarcity of SARS-CoV-2-targeting immunosensors, time-consuming portable methods, and limited exclusion capability.
13. M. Feleke, Jane Cunningham, Jonathan B. Parr, O.A., and J.B.P. (2021) discuss machine learning-guided aptamer refinement and discovery, highlighting significant efficiency enhancement. However, weaknesses include reliance on a single test set, low AUC in models, limited training examples, and balancing computational predictions with experimental validation.
14. D ARAFAT HOSSAIN, LISA M. SEDGER, AND JOHN CANNING (2021) offers a cross-disciplinary view of testing and bioinformatic analysis of SARS-CoV-2 and other respiratory viruses. Strengths include a concise overview of diagnostic technologies and the importance of networked field diagnostics. Weaknesses include false results due to sample sensitivity, small sample volumes limiting detection accuracy, and challenges in obtaining realistic clinical data.
15. Harsh Kumar and Shweta Sharma (2021) discuss the contribution of deep learning in bioinformatics, emphasizing its role in disease analysis, early detection, and drug discovery. Weaknesses include debates over diagnostic accuracy, challenges in data requirements, and risks of overfitting.
16. Francisco M. De La Vega et al. (2021) explore how AI enables comprehensive genome interpretation for rare genetic diseases, integrating predictive techniques and automation. Weaknesses include potential oversight of pipeline

effects, concerns over false positives, and limitations regarding trio-sequencing requirements.

17. Weifeng Qi, Rongyang Li, Lin Li, Shuhai Li, Huiying Zhang, Hui Tian (2021) identify key genes associated with esophageal adenocarcinoma and develop a prognostic model for disease progression. Weaknesses include descriptive analysis, small data size, and the need for further functional experiments.

18. Tapan Behl et al. (2021) discuss how bioinformatics accelerates data collection, computational modeling, target identification, and drug discovery in the pharmaceutical industry. Challenges in translational bioinformatics and effective drug discovery are noted.

19. David W. Bates et al. (2021) present a comprehensive review of AI's role in patient safety, though reliance on a single database and exclusion of non-English articles may limit the scope of the study.

20. Ibrahim Ali Mohammed (2021) analyzes the use of artificial intelligence in COVID-19 diagnosis, research, and vaccine development. Weaknesses include limited interdisciplinary collaboration hindering innovation in AI-driven drug and vaccine development for COVID-19.

21. Hassen Louati and Rahma Lahyani et al. (2024) present an advancement in sustainable COVID-19 diagnosis by integrating artificial intelligence with bioinformatics in chest X-ray analysis. Their methodology demonstrates robustness adaptable to complex and diverse datasets for COVID-19 diagnosis. However, an ongoing challenge persists in finding the optimal architecture for deep convolutional neural networks (DCNN) in the dynamic deep learning field.

22. Aneeqa Ijaz and Muhammad Nabeel et al. (2022) explore leveraging artificial intelligence for respiratory disease diagnosis using cough analysis. While AI algorithms show high accuracy and speed in image and sound-based detection techniques, weaknesses include struggles with capturing diverse and accurate cough acoustics, privacy and security concerns, data collection issues, and challenges in model interpretability.
23. Sanjeev B. Khanagar and Lubna Alkadi et al. (2023) conduct a systematic review on the application and performance of artificial intelligence in oral cancer diagnosis using histopathological images. Certain AI models show exceptional performance, yet concerns about data protection, confidentiality, and possible interpretation problems relying heavily on software for medical diagnosis are raised.
24. Parvathaneni Naga Srinivasu and Jana Shafi et al. (2022) utilize recurrent neural networks for predicting type-2 diabetes from genomic and tabular data. While the method shows promise in precise prediction and matching with diseased sequences, challenges include difficulty in working with extensive gene sequence data, the need for comparison with other diagnostic methods, and evaluation with more datasets.
25. Chris K. Kim and Ji Whae Choi et al. (2022) develop an automated COVID-19 triage pipeline using artificial intelligence based on chest radiographs and clinical data. Strengths include practical implementation for clinical use, reduced bias in AI models, and assistance to clinicians. Weaknesses include reliance on a training set from the emergency department, the need for validation in real clinical contexts, and improving model interpretability.

26. Mata Calidonio, J., & Hamad-Schifferli, K. (2024) propose an approach to use machine learning to optimize paper immunoassays for SARS-CoV-2 IgG and IgM antibodies. ML optimizes test conditions achieving high accuracy and distinguishing distinct antibody profiles. Challenges include antibody development, optimization hurdles, and the need for reagents to differentiate disease variants.
27. Guo, K., Fu, X., Zhang, H., Wang, M., Hong, S., & Ma, S. (2021) predict the postoperative blood coagulation state of children with congenital heart disease using machine learning based on real-world data. Strengths include identifying potential targets for diagnosis and treatment and establishing a prognostic model. Weaknesses include limited ethnic diversity in data sources, descriptive analyses, small data size, and the need for further validation.
28. Zhao et al. (2020) introduce Phen2Gene, a tool for rapid phenotype-driven gene prioritization for rare diseases. It utilizes Human Phenotype Ontology terms and the H2GKB database, offering open-source code, web server access, and a RESTful API. While it handles noisy data, concerns exist regarding analysis speed variability and the costliness of deploying DeepPVP on the cloud. Scalability is hindered by limited datasets and the slow API of AMELIE 2. The paper lacks extensive discussion on impact and trade-offs in accuracy, and further exploration of scalability and cloud deployment is warranted.
29. Jamshidi et al. (2023) discusses the future of drug discovery, highlighting the synergy of edge computing, the Internet of Medical Things (IoMT), and deep learning. While innovative techniques show promise in advancing molecular diagnostics and gene-expression investigations, bandwidth and processing limits impede health data collection in IoMT, and security concerns arise with medical

data transfer. The lack of AI exacerbates pandemic challenges in health data analysis, necessitating deep learning-based methods to support diverse IoMT devices.

30. Shu et al. (2023) emphasizes the power of clinical diagnosis in deciphering complex genetic mechanisms in rare diseases. Clinical diagnosis guides genetic focus, accelerating testing and method development. However, limited focus on coding regions hampers gene-disease association discovery, and genome-wide sequencing often yields lower diagnostic rates than expected. Clinical heterogeneity complicates genetic analysis, highlighting the need for advancing functional assays to efficiently identify non-coding and mosaic variants.

6. Conclusion

This paper has provided a comprehensive examination of the role of artificial intelligence in advancing the field of bioinformatics by applying the survey method, consisting of 13 questions, to analyze the required results based on the 52 responses received., with a particular focus on its application in expediting the diagnostic process for diseases such as COVID-19, cancer, and various genetic and chronic. Through a meticulous review of 30 articles from highly regarded journals, we have analyzed the efficiency and effectiveness of AI techniques in enhancing diagnostic accuracy, speed, and overall healthcare delivery. Our findings underscore the transformative potential of AI in laboratory settings, where it has shown promise in predicting, preventing, and detecting adverse healthcare events. Moreover, AI's contribution to the pharmaceutical industry is noteworthy, particularly in drug discovery, development, manufacturing, and marketing, by enabling the generation and analysis of large datasets to identify potential drug candidates and predict their effectiveness and safety. The results highlight how important AI in medicine. AI has improved general competency and precision, decreased the subjectivity of the result, and allowed testing to be conducted with fewer resources. And describe how effective they are in identifying risk factors, maximizing treatment plans, and eventually improving patient outcomes. Artificial intelligence algorithms have also proven their ability to analyze a huge sample size from an extensive data source that includes settings for both primary and specialized care. Recent substantial technological developments in AI algorithms and data creation have mostly benefited the diagnostic and therapeutic domains. Also, explore the role of AI and machine learning algorithms in harnessing medicine and vaccines. Despite the optimism surrounding AI's capabilities, challenges such as the shortage of trained personnel to conduct AI-powered tests and concerns about the interpretability of AI tools persist. These issues highlight the need for ongoing research, education, and investment in the development of AI in bioinformatics.

As we look to the future, AI holds the key to unlocking new frontiers in medical diagnostics and treatment. By continuing to harness the power of AI and bioinformatics, we can expect to see significant advancements in personalized medicine, improved patient outcomes, and more efficient healthcare systems. The journey is far from over, but the path forward is illuminated by the promising results of AI applications in bioinformatics, as demonstrated by the research presented in this paper.

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References

- S. V. Pillai and R. S. Kumar, "The Role of Data-Driven Artificial Intelligence on COVID-19 Disease Management in Public Sphere: A Review," Decision, vol. 48, pp. 375-389, 2021. [Online]. Available: https://doi.org/10.1007/s40622-021-00289-3.
- M. K. Pandit, S. A. Banday, R. Naaz, and M. A. Chishti, "Automatic detection of COVID-19 from chest radiographs using deep learning," Radiography, vol. 27, no. 2, pp. 483-489, 2021. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S1078817420302285.
- J. Wang, V. Chavda, R. Prajapati, A. Bedse, J. Patel, S. Popat, et al., "An Amalgamation of Bioinformatics and Artificial Intelligence for COVID-19 Management: From Discovery to Clinic," Current Research in Biotechnology, p. 100159, 2023. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S2590262823000412.
- A. Rabaan et al., "Artificial Intelligence for Clinical Diagnosis and Treatment of Prostate Cancer," Cancers, vol. 14, no. 22, p. 5595, 2022. [Online]. Available: https://doi.org/10.3390/cancers14225595.
- R. Deepa, S. Arunkumar, V. Jayaraj, and A. Sivasamy, "Healthcare's new Frontier: AI-driven early cancer detection for improved well-being," AIP Advances, vol. 13, no. 11. [Online]. Available: https://doi.org/10.1063/5.0177640.
- M. Elhadary et al., "Revolutionizing chronic lymphocytic leukemia diagnosis: A deep dive into the diverse applications of machine learning," Blood Reviews, p. 101134, 2023. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S0268960X23000954.
- D. Shao et al., "Artificial intelligence in clinical research of cancers," Briefings in Bioinformatics, vol. 23, no. 1, p. bbab523, 2022. [Online]. Available: https://academic.oup.com/bib/article/23/1/bbab523/6470966.

- T. R. Undru et al., "Integrating Artificial Intelligence for Clinical and Laboratory Diagnosis - a Review," Maedica, vol. 17, no. 2, pp. 420-426, 2022. [Online]. Available: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9375890/.
- L. P. Zhuhadar and M. D. Lytras, "The Application of AutoML Techniques in Diabetes Diagnosis: Current Approaches, Performance, and Future Directions," Sustainability, vol. 15, no. 18, p. 13484, 2023. [Online]. Available: https://doi.org/10.3390/su151813484.
- E. Kogan et al., "A machine learning approach to identifying patients with pulmonary hypertension using real-world electronic health records," International Journal of Cardiology, vol. 374, pp. 95-99, 2023. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S0167527322018903.
- H. Alshazly, C. Linse, E. Barth, and T. Martinetz, "Explainable COVID-19 Detection Using Chest CT Scans and Deep Learning" Sensors, vol. 21, no. 2, p. 455, Jan. 2021. [Online]. Available: https://doi.org/10.3390/s21020455.
- A.K. Yadav, D. Verma, A. Kumar, et al., "The perspectives of biomarker-based electrochemical immunosensors, artificial intelligence, and the Internet of Medical Things toward COVID-19 diagnosis and management," Materials Today Chemistry, vol. 20, p. 100443, June 2021. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S2468519421000239
- M. Feleke, J. Cunningham, J. B. Parr, et al., "Machine learning guided aptamer refinement and discovery," Nature Communications, vol. 12, p. 2366, 2021. [Online]. Available: https://www.nature.com/articles/s41467-021-22555-9
- D. A. Hossain, L. M. Sedger, and J. Canning, "A Cross-Disciplinary View of Testing and Bioinformatic Analysis of SARS-CoV-2 and Other Human Respiratory Viruses in Pandemic Settings," IEEE Access, vol. 9, pp. 2681-2690, 2021. [Online]. Available: https://ieeexplore.ieee.org/abstract/document/9638682.
- H. Kumar and S. Sharma, "Contribution of Deep Learning in Bioinformatics," Global Journal on Application of Data Science and Internet of Things, vol. 5, no. 1, pp. 15, 2021. [Online]. Available: (HTTP.NET). ISSN: 2581-4370. Eureka Journals 2021. All Rights Reserved.
- F. M. De La Vega, S. Chowdhury, B. Moore, et al., "Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare

genetic diseases," Genome Medicine, vol. 13, no. 153, pp. 1-14, 2021. [Online]. Available: https://link.springer.com/article/10.1186/s13073-021-00965-0.
- K. Pritzker, "Impact of the COVID-19 pandemic on Molecular Diagnostics," Journal of Molecular Diagnostics, vol. 25, no. 3, pp. 123-145, 2021. [Online]. Available: https://pubmed.ncbi.nlm.nih.gov/33910436/.
- T. Behl, I. Kaur, A. Sehgal, S. Singh, S. Bhatia, et al., "Bioinformatics Accelerates the Major Tetrad: A Real Boost for the Pharmaceutical Industry," International Journal of Molecular Sciences, vol. 22, no. 12, p. 6184, 2021. [Online]. Available: https://www.mdpi.com/1422-0067/22/12/6184.
- D. W. Bates, D. Levine, A. Syrowatka, M. Kuznetsova, K. J. T. Craig, A. Rui, G. P. Jackson, and K. Rhee, "The potential of artificial intelligence to improve patient safety: a scoping review," Nature Partner Journals Digital Medicine, vol. 4, p. 41, 2021. [Online]. Available: https://pubmed.ncbi.nlm.nih.gov/33742085/.
- A. Mohammed, "An in-depth analysis of the use of artificial intelligence in the search for a cure for COVID-19 and the development of vaccines," International Journal of Novel Research in Engineering and Science, ISSN: 2456-4184, 2021. [Online]. Available: https://www.ijnrd.org/viewpaperforall?paper=IJNRD2107001.
- H. Louati, A. Louati, R. Lahyani, E. Kariri, and A. Albanyan, "Advancing Sustainable COVID-19 Diagnosis: Integrating Artificial Intelligence with Bioinformatics in Chest X-ray Analysis," Information, vol. 15, no. 4, p. 189, 2024. [Online]. Available: https://www.mdpi.com/2078-2489/15/4/189
- Ijaz, M. Nabeel, U. Masood, T. Mahmood, M. S. Hashmi, I. Posokhova, et al., "Towards using cough for respiratory disease diagnosis by leveraging Artificial Intelligence: A survey," Informatics in Medicine Unlocked, vol. 29, p. 100832, 2022. [Online]. Available: https://www.sciencedirect.com/science/article/pii/S235291482100294X.
- S. B. Khanagar, L. Alkadi, M. A. Alghilan, S. Kalagi, M. Awawdeh, L. K. Bijai, et al., "Application and performance of artificial intelligence (AI) in oral cancer diagnosis and prediction using histopathological images: a systematic review," Biomedicines, vol. 11, no. 6, p. 1612, 2023. [Online]. Available: https://www.mdpi.com/2227-9059/11/6/1612.

- P. N. Srinivasu, J. Shafi, T. B. Krishna, C. N. Sujatha, S. P. Praveen, and M. F. Ijaz, "Using recurrent neural networks for predicting type-2 diabetes from genomic and tabular data," Diagnostics, vol. 12, no. 12, p. 3067, 2022. [Online]. Available: https://www.mdpi.com/2075-4418/12/12/3067.
- K. Kim, J. W. Choi, Z. Jiao, D. Wang, J. Wu, T. Y. Yi, et al., "An automated COVID-19 triage pipeline using artificial intelligence based on chest radiographs and clinical data," NPJ Digital Medicine, vol. 5, no. 1, p. 5, 2022. [Online]. Available: https://www.nature.com/articles/s41746-021-00546-w.
- J. Mata Calidonio and K. Hamad-Schifferli, "An approach to use machine learning to optimize paper immunoassays for SARS-CoV-2 IgG and IgM antibodies," Nanoscale, vol. 3, pp. 677-687, 2024. [Online]. Available: https://pubs.rsc.org/en/content/articlehtml/2024/sd/d3sd00327b.
- K. Guo, X. Fu, H. Zhang, M. Wang, S. Hong, and S. Ma, "Predicting the postoperative blood coagulation state of children with congenital heart disease by machine learning based on real-world data," Translational Pediatrics, vol. 10, no. 1, pp. 33-43, Jan. 2021. [Online]. Available: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7882284/.
- M. Zhao, J. M. Havrilla, L. Fang, Y. Chen, J. Peng, C. Liu, C. Wu, M. Sarmady, P. Botas, and J. Isla, "Phen2Gene: Rapid phenotype-driven gene prioritization for rare diseases," NAR Genomics and Bioinformatics, vol. 2, no. 2, p. lqaa032, June 2020. [Online]. Available: https://doi.org/10.1093/nargab/lqaa032.
- M. (Behdad) Jamshidi, O. Moztarzadeh, A. Jamshidi, A. Abdelgawad, A. S. El-Baz, and L. Hauer, "Future of Drug Discovery: The Synergy of Edge Computing, Internet of Medical Things, and Deep Learning," Future Internet, vol. 15, no. 4, p. 142, 2023. [Online]. Available: https://doi.org/10.3390/fi15040142.
- L. Shu, T. Maroilley, and M. Tarailo-Graovac, "The Power of Clinical Diagnosis for Deciphering Complex Genetic Mechanisms in Rare Diseases," Genes, vol. 14, no. 1, p. 196, 2023. [Online]. Available: https://doi.org/10.3390/genes14010196.