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# The Use of AI in Detecting Rare Diseases

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## ABSTRACT

Rare diseases, affecting fewer than 1 in 2,000 individuals, present significant challenges in diagnosis and treatment due to their diversity, limited clinical data, and lack of awareness among healthcare professionals. Artificial intelligence (AI) is emerging as a valuable tool in rare disease detection, leveraging large datasets and machine learning algorithms to analyze symptoms, genetic data, and medical literature. This paper investigates how AI enhances diagnostic accuracy, shortens the diagnostic process, and addresses the complexities associated with rare diseases. Additionally, we examine case studies, discuss the ethical considerations of AI-based diagnosis, and highlight the potential for AI to revolutionize rare disease care.

**Keywords:** Artificial Intelligence (AI), Rare Diseases, Machine Learning, Diagnosis, Genetic Testing.

## INTRODUCTION

Rare diseases are defined as those that affect fewer than 1 in 2,000 people. These diseases are also referred to as orphan diseases because the pharmaceutical industry has little incentive to develop treatments for them. Currently, there are approximately 7,000 rare diseases that have been identified globally, of which 80% have a genetic basis. Rare diseases are dispersed over a wide range of clinical symptoms and human body functions. Some are cognitively degenerative, some lead to loss of muscle functions, and some prevent a person from building a proper body structure. Because of this variety of disorders, medical professionals find it challenging to curate enough information about the symptoms of a rare disease that might help in mass diagnosis and patient detection. While there have been a few efforts to explore artificial intelligence systems for the detection of commonly known diseases, there have been very few attempts to develop AI systems that can help detect rare diseases [1, 2]. Diagnosing rare diseases is a lengthy process with low success rates. It takes about five years due to a lack of knowledge in the medical field. AI systems can help detect some rare diseases by curating medical evidence data. With over 40 million biomedical articles, AI can mine this information to align diseases with symptoms. By scraping and pre-processing articles, a symptom-disease knowledge base can be created using natural language processing algorithms. Machine learning algorithms can then be used to develop a symptom-disease model [3].

## OVERVIEW OF RARE DISEASES

Rare diseases, also known as orphan diseases, affect only a small portion of the population. In the US, a rare disease is defined as one that affects fewer than 200,000 people, while in Europe, it is less than 1 in 2,000 people. There are approximately 7,000 recognized rare diseases globally, with around 95% lacking

approved drug treatments. Challenges include imprecise diagnostics, limited clinical evidence, difficulties in patient identification, and high costs of developing treatments. Consequently, many rare diseases go misdiagnosed or undiagnosed, leading to inadequate medical care. Most rare diseases are genetic disorders inherited from parents, while others can be sporadic or caused by environmental factors [4, 5]. Most diseases introduced in the 18th to 19th centuries acted as catalysts for medical thinking. Young patients with similar conditions initiated rare disease studies, leading to the discovery of new diseases. Today, a catalog of rare diseases is investigated worldwide. Popular examples include galactosemia, osteogenesis imperfecta, and Tay-Sachs disease. Understanding their mechanisms is essential for developing therapies, such as gene therapies [6].



## CHALLENGES IN DIAGNOSING RARE DISEASES

People with rare diseases face multiple issues in their condition management. There is usually a long interval between noticing symptoms and receiving a diagnosis, resulting in a disease-confirming time of 5 years to more than 30 years. Patients with rare diseases usually encounter misdiagnosis or delays in diagnosis due to the lack of knowledge and awareness of healthcare professionals. As a result, wounds are frequently treated, totally inappropriate medicines are prescribed, and the patient's condition may worsen with time. A patient with Cystic Fibrosis may be treated for asthma or bronchitis initially. Moreover, properly categorizing and acknowledging a rare disease, may take even longer due to the variability of its presentation and symptomatology. This is because patients with the same disease may not always have the same issue and some unique symptoms may be linked to other health issues. This increases the complexity of disease recognition, resulting in misdiagnosis or delayed diagnosis. Such a situation further complicates the further analysis of a rare condition and decreases the probability of how a correct rarity index is assigned to a newly introduced condition [7, 8]. Genetic testing for rare diseases is often inaccessible due to limited machinery or knowledge. Consequently, this frequently leads to patients and their relatives enduring arduous trials, expending substantial amounts of time and financial resources, all while remaining without any comprehensive resolution to their affliction. Furthermore, the prevailing lack of funding allocated towards rare disease research further compounds the issue, exacerbating the challenges faced by affected individuals. Moreover, the restricted scope of collaboration and communication further hinder the timely identification and acknowledgment of these exceptionally uncommon ailments [9].

## ROLE OF ARTIFICIAL INTELLIGENCE IN RARE DISEASE DETECTION

Advancements in AI technology are revolutionizing rare disease detection by analyzing patient data and assisting in accurate diagnoses. AI can analyze diverse data from health and non-health sources to aid in the diagnosis and treatment of rare diseases. AI in personal health care can locate genetic anomalies and provide initial diagnoses, while also helping specialists plan treatment by analyzing data on drug effectiveness. Overall, AI enhances decision-making for healthcare professionals [10, 11]. Machine learning algorithms are implemented in clinical workflows. Decision support systems enhance decision-making. AI systems aggregate feedback and improve performance. AI is used to diagnose diseases and analyze lab results. Patient involvement is important. AI supports analysis bottlenecks in genetic diagnostic testing. AI models rare conditions and identifies affected pathways. Drugs are determined through in silico modeling. Analysis of image and transcriptome data is performed. Rare diseases are disregarded due to limited samples [12].

## CASE STUDIES AND SUCCESS STORIES

Several case studies of AI being used in the diagnosis of rare diseases have shown significant improvements in patient care and return on investment. For example, AI was used to diagnose a child with seizures and other symptoms, allowing for early treatment. In another case, AI helped identify the genetic cause of a baby girl's developmental delay. These successes demonstrate how AI can enhance physicians' abilities and expand diagnostic options [11]. A significant proportion of rare diseases are undiagnosed, with most taking 3-5 years and 5-7 medical examinations to diagnose. Collaboration between tech companies and healthcare is crucial for earlier diagnosis and better outcomes. A pilot study in Israel used AI to analyze phenotypic data, facial photos, and genetic variants. AI suggested a probable diagnosis in 46% of 138 undiagnosed cases, improving the overall diagnostic yield to over 70%. This highlights the potential for AI in clinical settings, offering speed, accuracy, and cost-effectiveness. Challenges include identifying, training, and recruiting personnel, and establishing a beneficial relationship with healthcare providers [13].

## ETHICAL CONSIDERATIONS IN AI-BASED DIAGNOSIS OF RARE DISEASES

In recent years, there has been increased awareness of ethical challenges in AI usage, particularly in rare disease detection and diagnosis. By implementing ethical principles, society can benefit from AI while minimizing adverse consequences. An important ethical challenge is biases in AI-based medical technology due to flawed and biased training datasets, leading to inaccurate diagnoses. This is particularly concerning for patients in poor regions who are underrepresented in training datasets. To ensure accuracy, medical imaging data from poor regions should be added to the training set. Another challenge is the model's inability to make accurate predictions on data from different populations. Therefore, finding datasets from various populations is crucial to enhancing model generalizability [14, 3]. Trusting AI systems in diagnosis, especially deep neural networks, poses ethical dilemmas due to the lack of interpretability. The opaque decision process of these models makes it difficult to judge their appropriateness and ethical standards. This raises concerns about deploying AI models in healthcare as irreversible actions may be taken based on their predictions. Additionally, privacy issues arise with the growing use of cloud platforms, as patient data can be exposed to cyberattacks. Privacy-preserving frameworks are needed to protect private medical data while AI models operate on it [15, 16].



## CONCLUSION

AI holds significant promise in transforming rare disease detection by providing faster, more accurate diagnoses, especially in cases where traditional methods struggle due to a lack of clinical data. By leveraging machine learning, natural language processing, and genetic analysis, AI systems can integrate vast biomedical data, offering innovative solutions for early diagnosis and personalized treatment. However, ethical challenges such as data bias, privacy concerns, and model transparency must be addressed to ensure equitable and responsible use of AI in healthcare. As AI continues to evolve, it can potentially bridge the gaps in rare disease diagnosis, leading to better outcomes for patients globally.

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