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A Review of AI-powered Diagnosis of Rare Diseases

Shanavaz Mohammed[1], Dr. Talmeez Ahmed Syed DDS[2], Nasar Mohammed[3], Waseem Sultana[4]
[1]School of Computer and Information Sciences, University of the Cumberlands, Williamsburg, KY
[2]School of Dentistry, University of Minnesota, Minneapolis, MN
[3]Valparaiso University, Valparaiso, IN
[4]Canadore at Stanford – Scarborough, ON, Canada

ABSTRACT: The diagnosis of rare diseases presents significant challenges due to their low prevalence, complex symptomatology, and the scarcity of specialized knowledge. However, advancements in Artificial Intelligence (AI) offer promising solutions to these challenges. This review explores the current state of AI-powered diagnostic tools for rare diseases, focusing on the methodologies, algorithms, and platforms utilized in this emerging field. We examine how AI technologies, such as machine learning, deep learning, and natural language processing, are being integrated into clinical practice to enhance diagnostic accuracy and speed. The research also provides the examples that highlight the successes and limitations of AI in this domain, providing insights into how AI can be harnessed to improve patient outcomes in rare disease diagnosis and management.

KEYWORDS: Artificial Intelligence (AI), Rare Diseases, Machine Learning, Deep Learning, Diagnosis, Genomics, Personalized Medicine, Data Integration

INTRODUCTION

Orphan diseases or rather rare diseases are defined as those medical conditions that predominantly affect a very small percentage of the population. These diseases, according to the National Cancer Center, are those ones that affect less than 200,000 people in the United States, while below 1 in 2000 people is prescribed in the European countries. It is recorded that there are also at least 7100 rare known diseases which are majorly as a result of the changes in a person's chromosomal or genetic materials [4]. For instance, some form of cancer such as pancreatic cancer and Ewing Sarcoma, take years to diagnosis and one might have shortened life due to delays in treatment [11]. Other examples include infectious diseases such as plague, smallpox and yellow fever. The aspect or rarity is that there has not been enough data about the diseases and that it can be quite difficult to diagnose the disease. These diseases are complex, severe and chronic, which poses a great life threatening challenges to the patients. The diagnostic journey of these rare diseases is quite lengthy, and patients can wait for several years before getting an accurate diagnosis. This results to delays in treatment options which might lead to worsened conditions and also adds to the emotional and financial burden on patients and their families.

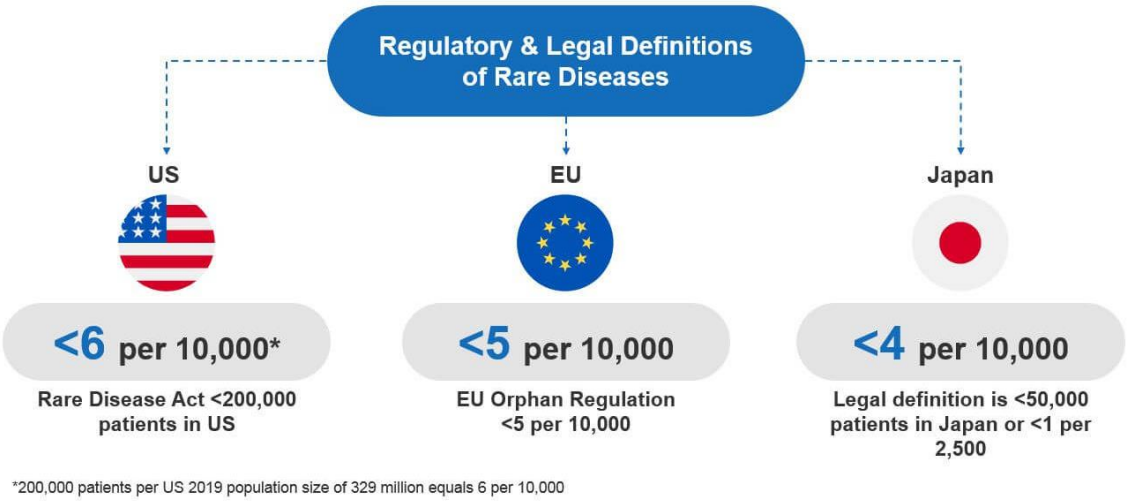

Figure 1: Diagram showing the legal definitions of rare diseases (Moritz. 20200

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Examples of rare diseases span a wide range of medical conditions, affecting various bodily systems. Some well-known examples include Huntington's disease, a neurodegenerative disorder; cystic fibrosis, which affects the respiratory and digestive systems; and Duchenne muscular dystrophy, a genetic disorder characterized by progressive muscle degeneration [19]. The global impact of rare diseases is profound, not only because of the number of people affected but also due to the significant healthcare resources required to manage these complex conditions. The burden of rare diseases is felt not just by patients but also by families, healthcare systems, and society as a whole, highlighting the need for improved diagnostic and treatment strategies.

The healthcare sector has greatly been revolutionized by the introduction of Artificial Intelligence (AI). AI has introduced several aspects of technology including machine learning, predictive analytics, Natural Language Processing (NLP), and deep learning [7]. All these technologies are meant to perform functions similar to human beings but with greater accuracy and less influence from the environment thus maintaining consistency. Therefore, it has been able to handle great amount of medical data from various sources such as Patients health data centers with less time and with greater accuracy. It has also been able to analyze huge data and identify patterns within the data set to make informed predictions about processes such as clinical trials. It has also come in handy when dealing with rare diseases where traditionally there is a limited amount of data that prevents proper diagnostics and treatment. AI has helped reduce the amount of time for research into the rare conditions which has led to increased data available for reference, reduced time for diagnostics and treatment which has in turn led to saving of human life [2]. By getting to know how AI powered research on the rare conditions is done, we will understand the benefits that patients and healthcare institutions can get in the short and long-term. We will also look at both the ethical and regulatory challenges that come with integrating AI into rare disease diagnosis.

CHALLENGES OF RARE DISEASES

As earlier mentioned, rare diseases are not common as they affect a small number of the population. Treating these disease is quite hectic and time consuming due to several factors. For instance, the diseases have complex and varied symptoms [20]. Most of the rare diseases have overlapping symptoms with a variety of the common diseases. This means that they will initially be mistaken or misdiagnosed for other common diseases as they continue to manifest into full stages [24]. For instance, most rare diseases will start off by giving out symptoms such as fatigue, neurological issues, muscle weaknesses and fevers. These symptoms can easily be mistaken for other common diseases such as malaria. For instance, Fabry disease is a rare genetic disorder that is characterized by the body being unable to break down a specific type of fat [11]. This leads to accumulation of fat on the major body organs which might lead to complications. Some of the common signs and symptoms of Fabry disease is dizziness, fever, fatigue, sweating and swelling, numbness and extreme pain during physical activity. These symptoms are non-specific and can resemble those of more common conditions like fibromyalgia, chronic fatigue syndrome, or even rheumatologic diseases [18]. For this reason, the patients may be treated for these symptoms, while not treating the underlying condition in itself. These signs and symptoms will be progressive and can also be unpredictable meaning that they might change with the disease advancements [23]. Complexity of these diseases mean that for correct diagnosis and treatment, a high level of expert advice is needed where the physicians must undertake numerous tests to write off any chances of serious infections and onset of the rare diseases.

Another issue with the correct diagnosis of the rare diseases and that hinders proper treatment is that there is a lack of clinical experiences. This means that due to the scarcity of patients with these rare conditions, physicians generally encounter one or two cases in their life time. They do not dwell too much on understanding too much information about the rare diseases leave alone studying their specific signs and symptoms [2]. The limited exposure to these rare diseases means that any diagnosis that they make, they cannot link the patients signs and symptoms to a rare disease but rather they connect the symptoms to common diseases. The medical trainings can also be blamed for this limited or lack of clinical experience since they focus more on the common illnesses which means that the clinical doctors are left with minimal knowledge about the rare conditions such that they are forced to seek more research if they encounter any rare disease [15]. This knowledge gap can result in patients undergoing numerous tests, seeing multiple specialists, and receiving incorrect diagnoses before the rare disease is finally identified, further delaying treatment.

Another reason it takes time to treat the diseases is that there is limited research on these conditions. It is clear that researchers put a lot of effort into areas that affect the largest population as it will yield the largest benefit in terms of treatment [3]. Therefore, the rare diseases are not accorded much time for research as they affect only a small portion of the community. Lack of research stems from the lack of proper financing into the course. This kind of research also lack the necessary funding to do a thorough dig into the

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causes and treatment of the rare diseases. Researching diseases is a capital intensive process which requires funds to conduct clinical trials, laboratory experiments, patient recruitment, research institutions, drug development among other key factors [1]. Without proper financing from institutions such as WHO will result to poor attention given to the rare diseases as well as poor management of patients. The lack of research into the rare conditions also results in lack of proper medication development from the pharmaceutical companies. This lack of proper medication can sometimes be costly as clinicians opt to use off the counter drugs to treat patients which might lack the efficiency. These issues combined lead to "diagnostic odyssey," which means patients endure a lengthy and often frustrating journey to obtain an accurate diagnosis and treatment [4].

AI TRANSFORMATION OF RARE DISEASES CASES

These challenges facing the rare diseases treatment showcases the gaps that exist in the medical research and treatment fields. Artificial Intelligence (AI) presents a variety of innovative approaches that have helped the industry to catch up and increase capabilities in the diagnostics and treatment options. AI's ability to increase speed of research, remove human error, analyze large data sets as well as perform repeat processes with ease, make it the best solution for majority of the issues with rare disease treatment and identification. Some of the critical areas that AI has been able to transform are discussed herein.

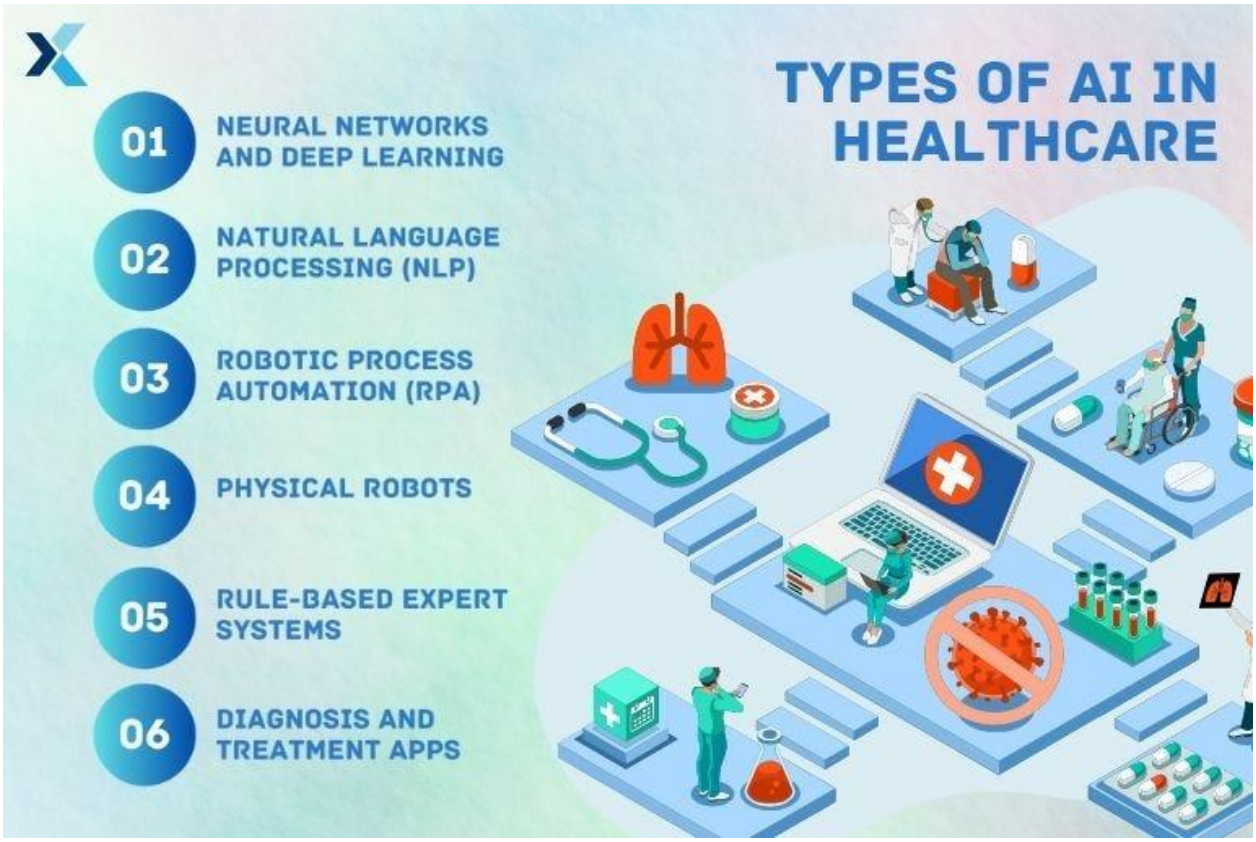

Figure 2: Diagram illustrating some of the AI systems in healthcare (AnalytixLabs, 2024).

i. Enhanced research

AI has the ability to comb through a large data set of information with ease thanks to its technology such as machine learning and Natural Language Processing (NLP). In this case it can peruse over the medical records archives, numerous clinical trials and genetic studies with a specific aim on rare diseases. It is able to filter off the numerous research items on the internet with ease and only focus on rare disease information. One area that AI has been able to assist is in the research of possible causes of the diseases such as the genetics and hereditary factors [8]. AI powered platforms have been able to research on the genetic sequences to identify mutations associated with rare genetic disorders. This research can be cumbersome to human since there is not a lot of information

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about that on the internet and would require a lot of time to comb through the numerous databases in order to find enough information. AI technology has helped in reducing the time and work needed to research on key components of rare disease diagnostics and treatment. For rare diseases, where patient data is often limited and scattered, AI's ability to aggregate and analyze data from multiple sources is crucial [25]. AI tools can uncover new genetic mutations, biomarkers, or disease mechanisms that may not have been previously identified, paving the way for new research avenues and potential treatments [16]. AI technologies are able to identify the rare and variable symptoms that are often under looked. As such this information can assist doctors be able to identify the signs earlier on.

ii. Better diagnostics

AI's capability to quickly analyze and interpret large data sets makes it ideal for increased speed and accuracy of rare diseases diagnostics. This advantage ensures that patients get earlier intervention and better patient outcomes. Aspects of AI technology such as machine learning, facilitates predictive decision making based on data present from medical platforms. Medical AIs can study the medical images from patients with rare diseases which enables identification of crucial patterns in genetic data and therefore predict the occurrence of certain diseases. Deep learning aspect of AI has enabled study of imaging data from MRI or CT scans with a keen eye which has enabled identification of underlying abnormalities which might be evade the human eye [21]. Natural language processing, another critical AI technology, allows computers to understand and interpret human language, making it possible to extract valuable information from unstructured data such as electronic health records (EHRs) and medical literature [17]. Moreover, AI can enhance the precision of diagnostics by reducing human error, which is particularly beneficial in complex cases involving rare diseases.

There are several AI systems that have been developed to facilitate efficient drug development and diagnostics in healthcare. For instance, Google health has been leading in developing AI systems that utilize deep learning to aid in diagnostics in radiology section [12]. IBM Watson Health uses AI to analyze vast amounts of medical data to support clinical decision-making. The Zebra Medical Vision focuses on providing informed clarity on the medical images especially for patients with rare conditions. PathAI is another technology used by big pharma where it is utilized for drug and diagnostic development in pathology [9]. Other AI technologies have been able to identify lung cancer by checking on the imaging data that would have otherwise been missed by radiologists. AI is used to tailor treatments to individual patients based on their genetic makeup, lifestyle, and other factors [22]. This approach not only improves the effectiveness of treatments but also reduces the likelihood of adverse reactions.

iii. Drug development

AI algorithm can also be programmed to scan through a large data set to identify potential drug targets which is the first step to drug development. This process can be cumbersome and can take many years but has been made easier and much faster thanks to AI integration into the healthcare system. Once potential drug candidates are identified, AI can also assist in designing and optimizing these molecules for better efficacy and safety. AI technology such as the natural language processing (NLP) is used to extract valuable information from medical literature, research papers, and case studies helping researchers to stay updated with the latest medical knowledge and to incorporate findings from new studies into their diagnostic algorithms. This data can be used to create a comprehensive personalized data for each patient with rare diseases which might help understand the issue better.

In the same manner, AI can be used for drug repurposing whereby existing drugs can be used for other treatments with higher efficacy levels. This can be done by examining existing patterns in how drugs interact with various biological targets and AI will be able to suggest new therapeutic applications for drugs that are already approved or in development [3]. This ultimately saves a lot of years which a new drug would take to develop. AI algorithms can also be used to identify potential harmful effects of drugs on the human body through research and data analysis [10]. This would help in identifying ways in which the drugs can be modified to ensure safety of the patients. This aspect has also been employed in designing personalized medicines with the help of analyzing clinical and genetic data of the patients. This approach not only improves patient outcomes but also makes drug development more efficient by targeting therapies to those who need them most.

iv. Better patient monitoring and care

AI technology has been instrumental in ensuring that patients get better healthcare from the comfort of their homes. For instance, AI has enabled personalization of treatment plans which are tailored to a patients' health profile. Personalized treatment plans have been enabled through careful analysis of a patient's health data such as genetics, treatment history, and lifestyle to ensure that the patient receives the best and most effective treatment specific to him/her [8]. For instance, AI algorithms like those used by Tempus

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analyze clinical and molecular data to tailor cancer treatment plans to individual patients, leading to more personalized and effective care. AI technology has also allowed for proper medication adherence by making sure that patients receive alerts and reminders for them to take their medications. It also sets reminders of when refills should be scheduled once the medicine is depleted. This is particularly useful for patients of the rare and chronic diseases. One particular technology that has enabled reminders for medications is the Medisafe mobile application that engages a patient by tracking adherence to medication as well as providing educative materials on how best to maintain a treatment plan and its benefits [11].

v. Collaboration and data sharing

One area that AI has ensured in promoting is the collaboration with professional medics as well as seamless sharing of data between the patient and the health organizations. Wearable technologies have revolutionized how patients communicate and share data with the healthcare practitioners. For instance, devices such as smartwatches and fitness trackers, continuously monitor various health parameters, including heart rate, blood pressure, and physical activity levels [5]. This data can then be transmitted to the healthcare institutions for monitoring by experts in real time to provide interventions when needed. AI supported telemedicine and telehealth technologies enable consultations with experts at the comfort of a patients' home without the need to physically visit an office or hospital [17]. This enhanced connectivity ensures that patients can easily share their health data with multiple healthcare providers, leading to more accurate diagnoses and more effective treatment plans.

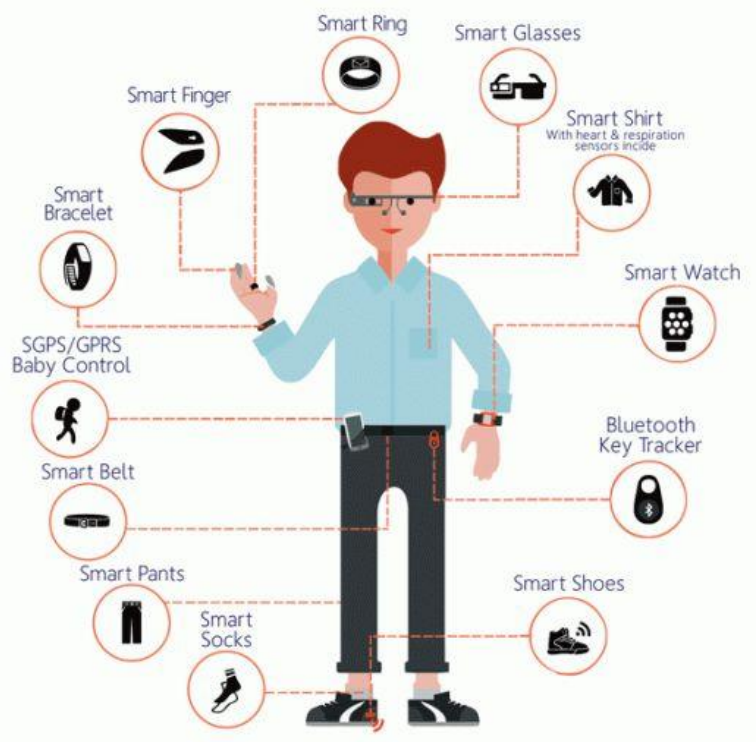

Figure 3: Figure showing some variety of wearable technology (Mishra, 2018)

The AI technology also assists in the integration of patient data with the Electronic Health Records (EHR) to provide healthcare providers with comprehensive views of their patients' health. This integration allows for more informed decision-making and ensures that all relevant data is considered when developing treatment plans. For instance, DeepMind Health is one of the Google Health technology which helps doctors have access to patient data across the EHR system where they simultaneously provide guidance and help patients improve their health [3]. The integration of patient data has been keen on diagnosing the rare diseases which rely heavily on the diversity of data from both medical literature and the genetic data. Genetic data, including DNA sequences and information about genetic mutations, provide critical insights into the underlying causes of many rare diseases.

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CONCLUSION

AI-powered diagnosis holds transformative potential for the field of rare diseases, offering solutions to some of the most persistent challenges in this area. By leveraging advanced algorithms and integrating diverse data sources, AI can significantly reduce the time to diagnosis, improve diagnostic accuracy, and identify conditions that might otherwise go unrecognized. However, realizing this potential requires overcoming several hurdles, including the "black box" nature of AI algorithms, ethical concerns surrounding patient data, and the need for robust regulatory frameworks. Additionally, global collaboration and data sharing are essential to ensure that AI models are trained on diverse and representative datasets, further enhancing their applicability and reliability. As AI technologies continue to evolve, their integration into healthcare promises to revolutionize the diagnosis and treatment of rare diseases, offering hope for earlier intervention, more personalized care, and ultimately, better patient outcomes.

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