

Can AI Diagnose Rare Diseases? Shortening the Diagnostic Odyssey

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Abstract

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Can AI Diagnose Rare Diseases? Shortening the Diagnostic Odyssey

The journey to a rare disease diagnosis is often described as a "diagnostic odyssey"—a prolonged, frustrating, and emotionally taxing process that can span years or even decades. With over 7,000 known rare diseases, affecting an estimated 300 million people globally, the challenge for clinicians is immense. The complexity of these conditions, coupled with the scarcity of patient data and a lack of specialized expertise, makes accurate and timely identification a formidable task. In this landscape, **Artificial Intelligence (AI)** is emerging not just as a tool, but as a potential co-pilot in the quest to end the diagnostic odyssey.

The Formidable Challenge of Rare Disease Diagnosis

Rare diseases, by definition, affect a small percentage of the population, leading to several systemic hurdles in diagnosis. Clinicians may encounter a rare condition only once or twice in their entire career, making recognition difficult. Furthermore, the symptoms are often non-specific, mimicking more common ailments. The core issues are:

- Data Scarcity:** Limited patient cohorts mean the data needed to train diagnostic models is sparse and fragmented.
- Clinical Heterogeneity:** The same genetic mutation can manifest in wildly different ways, complicating pattern recognition.
- Diagnostic Delay:** The average time to diagnosis for a rare disease can be over five years, during which time the disease may progress and treatment options narrow [1].

AI's Role: From Genomics to Phenotyping

AI, particularly machine learning and deep learning, offers a powerful new

lens through which to examine the subtle, complex patterns characteristic of rare diseases. Its application is primarily focused on two critical areas:

1. Genomic Analysis and Interpretation

The majority of rare diseases have a genetic origin. AI excels at processing the massive datasets generated by whole-genome and whole-exome sequencing. Traditional analysis can be slow and prone to overlooking subtle variants. AI algorithms can rapidly sift through millions of data points to identify pathogenic mutations, prioritize candidate genes, and correlate genetic findings with clinical symptoms [2]. This capability significantly accelerates the process of pinpointing the underlying cause of a patient's condition.

2. Image-Based Phenotyping

AI is transforming how clinicians interpret medical images and patient photographs. Deep learning models can be trained to recognize subtle facial dysmorphism, skeletal abnormalities in X-rays, or specific patterns in MRI scans that are indicative of a rare syndrome. For instance, AI tools can analyze patient photos to suggest potential genetic syndromes with high accuracy, providing a crucial starting point for genetic testing [3]. This is a powerful example of how AI can translate visual data into actionable diagnostic hypotheses.

The Promise and the Pitfalls

The potential of AI to shorten the diagnostic timeline is transformative. Early successes, such as the use of Large Language Models (LLMs) to analyze electronic health records and suggest diagnoses, show promising initial diagnostic rates [4]. By integrating data from genomics, imaging, and clinical notes, AI systems can generate a comprehensive, holistic view of the patient that no single human clinician could synthesize as quickly.

However, the path is not without obstacles. The primary challenge remains the **quality and quantity of training data**. AI models are only as good as the data they learn from. For rare diseases, data is often siloed, inconsistent, and lacks the necessary diversity. Ethical concerns also loom large, particularly regarding data privacy, algorithmic bias, and the need for a "human guarantee" to ensure that AI recommendations are validated by expert clinicians [5].

The Future of Digital Health and Expert Insight

The future of rare disease diagnosis lies in a symbiotic relationship between advanced AI systems and human expertise. AI will handle the heavy lifting of data processing and pattern recognition, while clinicians will provide the critical judgment, empathy, and final diagnostic authority. This partnership is essential to ensure that the technology serves to augment, not replace, the physician's role.

For more in-depth analysis on this topic, including the ethical frameworks and implementation strategies for AI in clinical settings, the resources at www.rasitdinc.com provide expert commentary

and professional insight into the evolving landscape of digital health.

The integration of AI into the diagnostic workflow is not a distant dream; it is a rapidly evolving reality. By addressing the challenges of data and ethics, AI promises to turn the years-long diagnostic odyssey into a manageable journey, offering hope and timely treatment to millions of patients worldwide.

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References

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Conclusion

The convergence of AI and digital health represents a pivotal moment in the fight against rare diseases. While challenges in data standardization and ethical governance persist, the sheer potential of AI to analyze complex, multi-modal data—from a patient's genome to their facial phenotype—offers a powerful mechanism to drastically reduce the time to diagnosis. This technological leap is not merely an incremental improvement; it is a fundamental shift that promises to transform the lives of millions, moving them from the uncertainty of the diagnostic odyssey to the clarity of a timely and accurate diagnosis. The ongoing collaboration between technology developers, clinicians, and researchers will be key to realizing this future.