

Can AI Accelerate Rare Disease Diagnosis?

Rasit Dinc

Rasit Dinc Digital Health & AI Research

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Abstract

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The Diagnostic Odyssey: A Challenge for Modern Medicine

For individuals with rare diseases, the path to a diagnosis is often a long and arduous journey, commonly referred to as a “diagnostic odyssey.” This journey can involve numerous specialist visits, extensive and often inconclusive tests, and an average of five to seven years before a definitive diagnosis is reached. The rarity of these conditions, with over 7,000 distinct rare diseases identified, means that most clinicians will have limited, if any, experience with a specific rare disease. This lack of familiarity, coupled with the often complex and heterogeneous presentation of symptoms, contributes significantly to diagnostic delays. The consequences of these delays are profound, leading to prolonged suffering, delayed treatment, and increased emotional and financial burdens on patients and their families. In this context, the advent of artificial intelligence (AI) offers a beacon of hope, promising to revolutionize the diagnostic landscape for rare diseases.

AI: A Paradigm Shift in Rare Disease Diagnosis

Artificial intelligence, particularly its subfield of machine learning (ML), is emerging as a transformative force in healthcare. By leveraging the power of complex algorithms to analyze vast and diverse datasets, AI can identify patterns and insights that may be imperceptible to human observers. This capability is particularly well-suited to the challenges of rare disease diagnosis. AI-powered tools can integrate and analyze a wide range of data sources, including electronic health records (EHRs), genomic data, and medical imaging, to provide a more holistic and accurate view of a patient's condition.

The Role of Machine Learning and Deep Learning

Machine learning algorithms can be trained on large datasets of diagnosed cases to recognize the subtle patterns associated with specific rare diseases. Supervised learning models, for instance, can be trained to classify patients based on their symptoms and genetic information, while unsupervised learning can help identify novel disease subtypes. Deep learning, a more advanced form of machine learning, has shown remarkable success in analyzing medical images, such as MRIs and CT scans, to detect the tell-tale signs of rare genetic syndromes that might otherwise be missed. Furthermore, natural language processing (NLP), another branch of AI, can be employed to extract critical information from unstructured clinical notes and patient histories, providing valuable clues for diagnosis.

From Theory to Practice: AI in Action

A compelling example of AI's potential is the development of the popEVE model by researchers at Harvard Medical School. As detailed in their 2025 publication in *Nature Genetics*, popEVE is an AI model designed to predict the likelihood that a specific genetic variant will cause disease [1]. By analyzing a patient's genome, popEVE can rank genetic variants by their potential severity, providing clinicians with a prioritized and clinically meaningful list of potential culprits. This innovative approach has already demonstrated its utility by identifying over 100 novel genetic alterations responsible for previously undiagnosed rare diseases. The success of popEVE and similar AI-driven tools underscores the immense potential of these technologies to shorten the diagnostic odyssey and improve patient outcomes.

Navigating the Challenges and Embracing the Future

Despite the significant promise of AI in rare disease diagnosis, several challenges must be addressed to ensure its widespread and equitable adoption. These include the need for large, high-quality datasets for training and validation, the development of robust and transparent algorithms to avoid bias, and the establishment of clear regulatory frameworks to ensure patient safety and data privacy. Furthermore, the integration of AI tools into existing clinical workflows will require careful planning and training to ensure that clinicians can effectively utilize these new technologies.

In conclusion, artificial intelligence represents a new frontier in the diagnosis of rare diseases. By harnessing the power of AI to analyze complex data and identify subtle patterns, we can significantly accelerate the diagnostic process, reduce the burden on patients and their families, and ultimately, improve the lives of millions of people affected by rare diseases. While challenges remain, the continued development and refinement of AI-powered diagnostic tools, coupled with a collaborative approach between researchers, clinicians, and patients, will undoubtedly pave the way for a brighter future in the world of rare diseases.

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