

## Commentary

# Would Artificial Intelligence Improve the Quality of Care of Patients With Rare Diseases?

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Rare diseases are characterized by low prevalence, they affect 4–5 patients per 10,000 people,<sup>[1]</sup> which results in a lack of knowledge, guidelines, training, and diagnostic complexities among clinicians. This significantly burdens the healthcare systems and negatively hinders the quality and patient safety.<sup>[2]</sup> There are 5000–8000 identified rare diseases affecting more than 350 million people worldwide; of these, 75% affect children, and 80% are genetically based.<sup>[3]</sup> Most patients with rare diseases have experienced a diagnostic odyssey through the prolonged journey of late diagnosis, which can take several years<sup>[4]</sup> and incur substantial cost estimated at \$US 1 trillion.<sup>[5,6]</sup> Nevertheless, the conclusion of their search for a diagnostic journey is usually followed by the beginning of another one, the “therapeutic odyssey”! A few examples of rare diseases are Huntington’s disease, Sickle cell disease, Gaucher disease, and hemophilia.<sup>[7,8]</sup> Neoplastic diseases are the most prevalent rare diseases (59%), followed by developmental, neurological, and circulatory diseases.<sup>[2]</sup> Proper management of these diseases leads to early intervention, prevention of deterioration, and enhances patients’ quality of life. This commentary aims to enlighten quality-focused clinicians on the substantial quality gap experienced by patients with rare diseases, and to explore the transformative power of artificial intelligence (AI) applications to improve the quality of healthcare services.

The substantial gap in diagnosing, treating, and monitoring patients with rare diseases profoundly impacts the quality of patient care and healthcare services.<sup>[7]</sup> In a study from EURODIS, the average time of diagnosis is between 5 and 30 years, and 40% were misdiagnosed during that period<sup>[4]</sup>; in most cases, patients who have been appropriately diagnosed lack access to appropriate medications, for which currently 90% of rare diseases have no effective treatments.<sup>[7]</sup> Thus, an examination through the lens of AI is warranted. AI can transform the quality of patient care for rare diseases by assisting

in early diagnosis, improving treatment effectiveness, and monitoring patients.

By leveraging AI in rare disease diagnosis, it becomes feasible to fulfill the 2027 goal set by the International Rare Diseases Research Consortium, which aims to diagnose all known rare diseases within a year of seeking medical advice on symptoms for the first time.<sup>[9]</sup> Recent advancements in AI for diagnosing rare diseases highlight an increasing use of supervised machine learning techniques; a recent systematic review of AI approaches for rare disease revealed that 86.3% of studies used supervised machine learning methods, with random forest being used most frequently in this group in 54.5% of the studies.<sup>[6]</sup>

In the realm of therapeutic treatment, AI tools have offered a glimmer of hope. The Hugh Kaul Precision Medicine Institute introduced “mediKanren,” an AI platform that has already benefited more than 500 families affected by rare diseases; it is easily searchable and assists healthcare professionals and researchers in comprehending rare diseases and pinpointing potential therapeutic avenues.<sup>[10]</sup> Second-generation AI was developed to rectify the gap in diagnostic and therapeutic monitoring; unlike first-generation tools and platforms, it continually monitors disease progression, treatment effectiveness, and environmental factors as part of a continuous dynamic feedback loop.<sup>[7]</sup> The integration of AI in therapeutic treatment also presents several challenges. Data privacy and security concerns persist, and the high costs of AI technology can limit accessibility. Furthermore, the lack of transparency in AI decision-making processes can hinder trust and understanding among healthcare professionals.<sup>[11]</sup>

Several studies have shown that AI-based approaches can be used to transform hospital-based quality improvement initiatives, especially for rare diseases. By leveraging AI, healthcare systems can improve early rare disease diagnosis, effective treatment, and monitoring, thereby improving patient care and quality of life.

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