

How AI Has Changed the World of Healthcare Diagnoses

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INTRODUCTION: THE CURRENT ISSUES WITH MEDICAL DIAGNOSES

Imagine experiencing excruciating symptoms for many years but receiving little to no medical support—not even a definitive diagnosis. This is a frustrating reality for many, especially those with rare diseases, where diagnoses can take anywhere from six months to two decades. A qualitative study, “Time to diagnosis for a rare disease: managing medical uncertainty,” explains that many patients’ diagnoses are delayed due to the lengthy process of reaching a final medical diagnosis.¹ Due to the arduous procedure of reaching a conclusive medical conclusion, many patients experience prolonged delays. Around 8,000 diseases fall under the category of rare diseases, affecting between 263 and 446 million people globally, demonstrating the crucial need for accurate and swift diagnoses. With Artificial Intelligence (AI), a diagnosis can be provided faster by quickly analyzing genetic databases to find discrepancies that a practitioner may have missed.

The medical diagnostic process is often slow, complex, and error-prone, raising important questions: What are the biggest challenges in medical diagnosis, and how can AI address them?

DESCRIPTION: THE PROCESSES TO REACH A MEDICAL DIAGNOSIS

As the National Institutes of

Health (NIH) describes, medical diagnoses are made using pre-existing categories, in which medical professionals decide an outcome to describe a specific condition. A clinical decision is then reached by analyzing the patient’s underlying health issues, communicating to the patient their entire recommended treatment plan, and determining the viability of a positive health outcome from their treatment.² Initially, a doctor makes a provisional diagnosis, or

an educated guess, about the cause of the patient’s symptoms. Provisional diagnoses may also be given when the patient does not meet the diagnostic criteria for a condition due to inconsistent symptoms. On the other hand, in a differential diagnosis, a doctor concludes that there are multiple possible diagnoses.³ Doctors must then rule out malingering and factitious disorders (fabricated or exaggerated information provided by the patient), drug-related causes, and other medical conditions to determine the primary disorder.⁴

Overall, doctors must review the patient’s history, conduct a physical exam,

perform diagnostic testing, and consult with other clinicians to gather all the information they need to make an accurate clinical decision. Still, according to Johns Hopkins Medicine, the error rate across diseases was estimated at 11.1% and ranged to almost 62% for spinal abscesses—a serious inflammation around the spinal cord.⁵ In another study, general practitioners participating in the study could not even establish a specific diagnosis for 36% of the patients with health

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problems.⁶

Other issues in extracting a diagnosis come from obtaining a clear patient history due to limitations in communication and self-reporting. In some cases, clinicians may overdiagnose or incorrectly identify a disease, leading to unnecessary treatment, increased healthcare costs, and stress for patients.⁷ Medical diagnoses are hindered by inaccuracy, communication gaps, uncertainty, and lengthy delays, highlighting the need for improved diagnostic tools and processes.

THE SOLUTION: AI TO THE RESCUE

The many problems that emerge from the inefficient process of medical diagnoses make it essential to find enhanced methods. One solution is the integration and usage of AI. Recently, the integration of AI in healthcare has demonstrated excellent potential for improving disease diagnoses, treatment selection, and clinical laboratory testing. AI tools can process large sets of data and identify patterns faster than humans. The article “Trust and Medical AI: The Challenges We Face and the Expertise Needed to Overcome Them” details how AI can convert a patient’s entire medical record into a clear and simple number representing a diagnosis.⁸ It can increase accuracy while minimizing human errors, thus reducing unnecessary costs. The rapid progression of AI technology shows its influential usage in clinical processes, equipping healthcare providers with the necessary knowledge and tools to implement it into patient care.⁹ In fact, more than half of AI/machine learning-based medical devices have been approved

for radiological use.¹⁰ In this study, AI has demonstrated its immense capabilities by meeting and even outperforming human experts in image-based diagnoses for several medical specialties. The high computing power of AI models allows them to operate rapidly while producing accurate results. In cardiovascular diseases, AI models were able to identify and classify the data sets to a specificity rate of 93%, as described in a study involving machine learning-based models.¹¹

However, the purpose of using predictive AI in healthcare diagnoses is to transform the industry, not directly replace traditional diagnostic processes. Through the human-centered AI approach, the user identifies the key issues, and then AI is implemented to evaluate them by operating within existing norms and practices.¹⁰ Because AI tools provide binary answers without revealing the underlying process that led to the conclusion, they are not reliable in all cases. According to the article, “Hidden flaws behind expert-level accuracy of multimodal GPT-4 vision in medicine,” AI

made errors when explaining and describing medical images despite correctly identifying the patient’s symptoms.¹² AI has also shown tendencies to make biased decisions while focusing on clinically irrelevant findings. The study “Measuring the Impact of AI in the Diagnosis of Hospitalized Patients” elaborates on the potential issue of the worsening spread of biases in healthcare if left unchecked.¹³ Figure 1 shows the differences between the standard and biased model in accuracy when providing a medical diagnosis and the algorithm for reaching it.

Due to AI’s biases and inability to provide transparent reasoning, clinicians are enhancing diagnostic processes by integrating AI’s capabilities while maintaining necessary oversight. The continuous monitoring of AI tools is essential for preserving accuracy and fairness in medical applications.¹⁴ This includes AI’s use in cross-referencing patient records and validating them against established medical standards. Additionally, training datasets must represent diverse populations to prevent misdiagnosis or underdiagnosis in underrepresented communities. Addressing ethical, regulatory, and trust-related concerns is essential for patient protection and accountability in healthcare when integrating AI.¹⁴

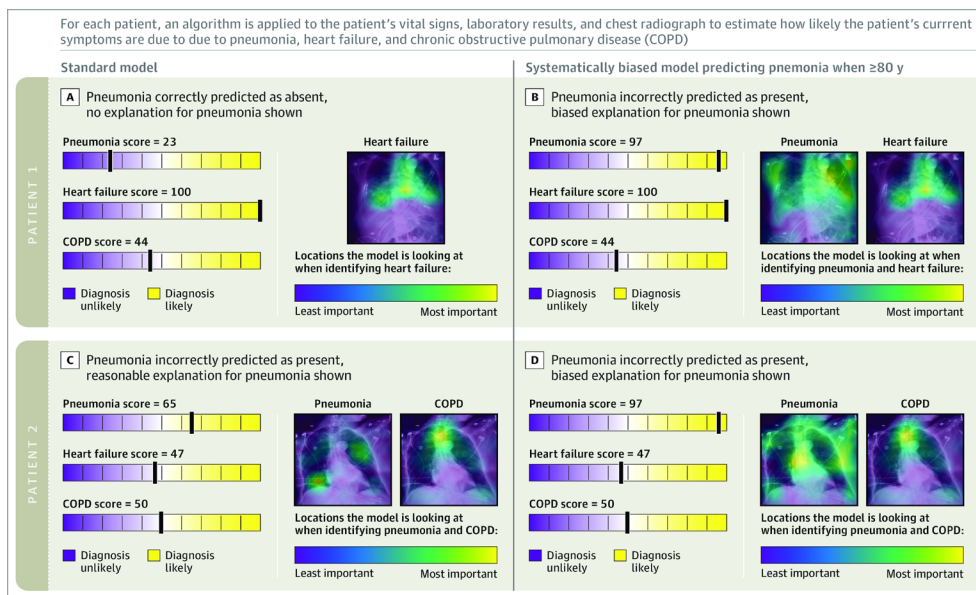


Figure 1: The AI model is analyzing the medical data from Patient 1 and Patient 2. A. The standard AI model correctly diagnosed heart failure as the cause of acute respiratory failure for Patient 1, an 81-year-old male with respiratory failure from heart failure, and provided an explanation, highlighting the regions in the chest radiograph used to make the prediction. **B.** On the contrary, the biased AI model diagnosed pneumonia as the cause of acute respiratory failure, failing to reach the right conclusion. This was due to the AI model taking in irrelevant information about the patient’s age and providing an explanation highlighting irrelevant features in the chest radiograph.

Patient 2’s data is from an 88-year-old female with respiratory failure from chronic obstructive pulmonary disease (COPD). **C.** The standard AI model incorrectly diagnosed pneumonia but correctly diagnosed COPD as the cause of respiratory failure and provided reasonable explanations. **D.** The biased AI model incorrectly diagnosed pneumonia as the cause of respiratory failure due to analyzing patient age and giving an explanation based on irrelevant areas in the radiograph.¹³

CONCLUSION: THE FUTURE OF MEDICAL DIAGNOSES WITH AI ADVANCEMENTS

In the future, AI advancements could lead to personalized diagnostic assessments that consider an individual’s genetic, lifestyle, and environmental factors. This shift towards medical diagnoses that are more precise and patient-specific could transform disease detection and treatment planning, making healthcare more efficient. With the exponential improvement of AI, medical uncertainties may be entirely eradicated, and accurate diagnosis may become the norm. Instead of enduring years of painful symptoms with no foreseeable diagnosis, someone with rare disease conditions may be able to receive their diagnosis in hours, thanks to Artificial Intelligence.

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