Is AI in Diagnostics Applicable to Neuromuscular Diseases?

Artificial intelligence (AI) tools are being increasingly utilized in diagnostics. A 2024 American Medical Association (AMA) survey of physicians found that 51% of respondents use or plan to incorporate AI as an assistive diagnostic tool in their practice.

According to Tina Duong, MPT, PhD, a research scientist and physical therapist at Stanford University, AI tools potentially could be useful for helping clinicians diagnose neuromuscular diseases, which encompass a broad range of conditions. Machine learning excels at synthesizing multi-modal data and recognizing subtle patterns in heterogeneous symptoms.

“With the appropriate learning models, you would have the ability to integrate a lot of complex data that are currently siloed in unstructured patient charts,” she says. She notes that clinical data may not be integrated into a single chart, and charts often contain a combination of quantitative (e.g., biomarkers) and subjective (e.g., clinical notes) data. “There’s a possibility of integrating all of this information; we just don’t know how yet.”

Currently, data scarcity in neuromuscular diseases remains a barrier for machine learning models. “One of the things to address is some kind of federated learning model, a decentralized approach where AI learns from data across multiple institutions without compromising patient privacy. The model is only as good as we train it,” Dr. Duong says. She emphasizes that it will take collaborative work to build up datasets, “similar to all the work that’s being done toward building disease registries.”

Clinicians must understand the advantages and pitfalls of AI diagnostic tools. “AI is your assistant, not your answer,” Dr. Duong says. Trust must be built by making AI explainable and interpretable. She notes that clinicians must understand the reasoning behind its conclusions to effectively communicate with patients.

In addition, patients should be informed when AI is used and must consent to the processing and storage of their data. “I think it’s just like consent for any treatment or drug,” she says. “Patients have to be able to consent for their data to go into anything, and they have to know who has access to the data.”

Dr. Duong currently uses AI primarily in neuromuscular clinical research. In her own research, she uses digital health technologies such as OpenCap, a markerless video-based motion capture system developed at Stanford, to objectively assess disease-specific changes in movement. Her goal is to track progression more accurately than just using traditional timed tests.

Dr. Duong believes that AI has the potential not only to reduce the diagnostic odyssey for neuromuscular diseases but also to be used in care management. This could help with a more individualized approach, particularly for ultra-rare diseases, and aid in precision medicine.

Resources

AMA EdHub: Governance for Augmented Intelligence
American College of Physicians: Artificial Intelligence (AI) Resource Hub
Joint Commission: AI, Data Analytics & Research
PATH: AI for Health Toolkit