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REVIEW
DERLEME

Redefining Differential Diagnosis in Chronic Diseases: The Emerging Role of Artificial Intelligence

Kronik Hastalıklarda Ayırıcı Tanının Yeniden Tanımlanması: Yapay Zekânın Gelişen Rolü

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ABSTRACT

Overlapping symptoms, involvement of several systems, and lengthy illness trajectories make differential diagnosis of chronic diseases extremely challenging in clinical practice. Disease progression, higher healthcare costs, and worse quality of life are consequences of incorrect or delayed diagnosis. Artificial intelligence (AI) has recently opened up new possibilities for improving clinical decision-making and diagnostic accuracy in complex chronic diseases. Focusing on their capacity to integrate multidimensional clinical data and support medical decision-making, this study aims to assess the performance of AI-based methods in improving the differential diagnosis of chronic diseases. A literature search was conducted in PubMed and Medline from 2000 to 2025. A narrative review of artificial intelligence (AI) applications on chronic diseases was conducted. Artificial intelligence (AI) diagnostic models outperformed human clinicians in distinguishing among inflammatory, autoimmune, metabolic, and neurological diseases that share similar symptoms. Compared with traditional clinical evaluations, these systems performed much better at identifying subtle disease-specific traits and recognizing complex patterns across large datasets. There was hope that AI-assisted tools could help shorten diagnostic wait times and bolster individualized treatment plans. By improving pattern identification, data integration, and diagnostic accuracy, artificial intelligence shows promise as an adjunct to the differential diagnosis of chronic diseases. Although AI systems can't replace human doctors, they could help diagnose patients faster and more accurately if used routinely. To ensure safe and effective adoption in chronic disease management, future research should focus on model transparency, clinical validation, and ethical implementation.

Keywords: Artificial intelligence, treatment, chronic diseases, predicting steps

ÖZ

Belirtilerin örtüşmesi, birden fazla sistemin tutulumu ve uzun hastalık seyirleri, klinik uygulamada kronik hastalıkların ayırıcı tanısını son derece güçleştirmektedir. Yanlış veya gecikmiş tanı; hastalık progresyonu, sağlık hizmeti maliyetlerinde artış ve yaşam kalitesinde bozulma gibi önemli sonuçlara yol açmaktadır. Son yıllarda yapay zekâ, karmaşık kronik hastalıklarda klinik karar verme süreçlerini ve tanısal doğruluğu artırmaya yönelik yeni olanaklar sunmuştur. Bu çalışmanın amacı, çok boyutlu klinik verileri entegre edebilme ve tıbbi karar verme süreçlerini destekleme kapasitelerine odaklanarak, yapay zekâ temelli yöntemlerin kronik hastalıkların ayırıcı tanısındaki performansını değerlendirmektir. 2000–2025 yılları arasında PubMed ve Medline veri tabanlarında literatür taraması yapılmıştır. Kronik hastalıklarda yapay zekâ uygulamalarını ele alan çalışmalar üzerinden bir inceleme gerçekleştirilmiştir. Yapay zekâ temelli tanı modellerinin; benzer semptomlar gösteren inflamatuvar, otoimmün, metabolik ve nörolojik hastalıkları ayırt etmede insan klinisyenlere kıyasla daha üstün performans sergilediği görülmüştür. Bu sistemler, geleneksel klinik değerlendirmelerle karşılaştırıldığında, hastalığa özgü ince özellikleri saptamada ve geniş veri setleri içerisindeki karmaşık örüntüleri tanımlamada belirgin şekilde daha başarılı bulunmuştur. Yapay zekâ destekli araçların, tanı süresini kısaltma ve bireyselleştirilmiş tedavi planlarını güçlendirme potansiyeline sahip olduğu düşünülmektedir. Yapay zekâ, ğörüntü tanıma, veri entegrasyonu ve tanısal doğruluğu artırma yoluyla kronik hastalıkların ayırıcı tanısında önemli bir yardımcı araç olma potansiyeline sahiptir. Yapay zekâ sistemleri insan hekimlerin yerini alamasa da, rutin klinik uygulamaya entegre edildiğinde daha hızlı ve daha doğru tanı konulmasına katkı sağlayabilir. Kronik hastalık yönetiminde yapay zekânın güvenli ve etkili biçimde kullanılabilmesi için, gelecekteki çalışmaların model şeffaflığı, klinik doğrulama ve etik uygulamalara odaklanması gerekmektedir.

Anahtar Kelimeler: Yapay zeka, tedavi, kronik hastalıklar, adımları tahmin etme

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INTRODUCTION

The integration of patient history, physical exam, laboratory results, imaging, and longitudinal follow-up is essential for differential diagnosis, a foundational step in clinical reasoning. Even for experienced doctors, a growing mountain of medical data, tighter deadlines, and increasingly complex patients are common challenges. Delays or incorrect diagnoses of chronic diseases can worsen the disease, cause permanent organ damage, and lower quality of life (1,2).

One powerful technique for addressing these issues is artificial intelligence (AI), which encompasses three divisions (3-5).

MACHINE LEARNING (ML)

The subfield of AI that enables computers to automatically learn new tasks and improve their performance by analyzing patterns in existing data. Machine learning (ML) has widespread applications in medicine, including risk stratification, classification, and prediction, using structured data derived from clinical parameters and laboratory findings (3-5)

• Deep Learning (DL)

A branch of machine learning that models complex, high-dimensional data using multilayer artificial neural networks. DL's ability to automatically extract useful features from raw data makes it well suited for image analysis (radiology, pathology), signal processing, and voice recognition (3-5)

• Natural Language Processing (NLP)

A subfield of AI concerned with teaching computers to read, comprehend, and create new text in the style of human speech. Medical reports, discharge summaries, and doctors' notes are examples of unstructured clinical material that can be analyzed using natural language processing (NLP) (3-5).

AI systems can sift through vast, diverse datasets to identify relationships and patterns that humans cannot. Here, we examine how AI is changing the game in chronic disease differential diagnosis and where we can go from here in terms of ethically integrating it into clinical practice (3-5).

CHALLENGES IN DIFFERENTIAL DIAGNOSIS OF CHRONIC DISEASES

Overlapping Clinical Presentations

The symptoms of many long-term illnesses are similar. Examples of conditions that frequently cause exhaustion and joint pain include rheumatoid arthritis, fibromyalgia, systemic lupus erythematosus, and persistent infections. As with asthma, COPD, GERD, and interstitial lung disease, other medical conditions can also present with a persistent cough. Such overlap lengthens the evaluation process and increases diagnostic uncertainty (6).

Multisystem and Dynamic Nature of Disease

Many organ systems are affected by chronic diseases, which tend to evolve over time. As the disease advances, new clinical symptoms appear, but early signs may be mild or unusual. This dynamic complexity can be too much for traditional diagnostic algorithms to handle, since they tend to be static and disease-specific (7,8).

Cognitive Bias and Human Limitations

Cognitive biases, such as anchoring, availability bias, and premature closure, may affect clinical thinking. When diagnosing chronic, rare, or complex diseases, these biases may lead to errors. Furthermore, doctors may struggle to process large amounts of longitudinal data within the allotted time for consultations. When computers can mimic human intelligence, we say they have artificial intelligence. These models are well-suited to complex diagnostic settings because, unlike rule-based decision support systems, they continuously improve through exposure to new data (3,9,10).

AI APPLICATIONS IN DIFFERENTIAL DIAGNOSIS OF CHRONIC DISEASES

Electronic Health Records and Pattern Recognition

Probabilistic differential diagnoses can be generated by AI models trained on EHRs by integrating demographic data, symptoms, laboratory trends, and comorbidities. There is evidence that ML-based systems can detect chronic renal disease, inflammatory bowel disease, and heart failure earlier than conventional scoring systems (3,11,12).

Imaging-Based Differential Diagnosis

Radiologic and histopathologic image analysis have been highly successful with deep learning. Subtypes of interstitial lung disease and neurodegenerative disorders on magnetic resonance imaging (MRI) are two examples of chronic diseases where AI-assisted imaging has helped distinguish between illnesses with similar radiographic appearances. By reducing interobserver variability, these techniques improve diagnostic accuracy (3,13,14).

Biomarker and Omics Integration

Chronic illnesses often involve intricate molecular processes. By integrating genomic, proteomic, and metabolomic data, AI can distinguish between the outward and internal manifestations of an illness. Because molecular signatures guide diagnosis and treatment in autoimmune and oncologic disorders, this approach has proven especially useful in these areas (14,15).

Symptom Clustering and Phenotyping

To identify new disease groupings, unsupervised ML methods can cluster symptom patterns and clinical trajectories. This approach's reimagining of illness categorization in diabetes, chronic rhinosinusitis, and asthma now enables more complex differential diagnosis and individualized treatment plans (15).

CLINICAL IMPACT AND BENEFITS

Improved Diagnostic Accuracy

When it comes to improving diagnostic sensitivity and specificity, AI systems really shine at spotting modest connections across different datasets. Artificial intelligence (AI)-powered solutions have shown promise in reducing diagnostic delays in chronic diseases such as systemic vasculitis and multiple sclerosis (3,16).

Reduction of Diagnostic Delays

Artificial intelligence (AI) can shorten the diagnostic journey for patients with chronic diseases by detecting high-risk patterns early and prompting additional examinations or referrals. Earlier diagnosis enables quicker intervention and better outcomes (15).

Decision Support and Physician Augmentation

Avoid treating AI as a substitute for human doctors; view it as a tool to support clinical decision-making. Well-integrated AI yields improved clinician confidence, reduced cognitive burden, and support for evidence-based decision-making (15).

Limitations and Challenges

The accuracy of AI models depends on the quality of the training data. Incomplete, biased, or non-representative datasets can lead to misleading or unfair diagnostic suggestions, disproportionately affecting marginalized groups. Many AI models are "black boxes," with no clear way to see how they make decisions. In high-stakes diagnostic settings, a lack of explainability could be a dealbreaker for clinicians. Data protection, responsibility, and informed consent are among the ethical issues raised by the use of AI in diagnosis. One outstanding legal challenge is determining culpability in cases of AI-related diagnostic errors (16,17). By taking a multidisciplinary approach to explainability in healthcare AI, we can make algorithmic models more interpretable, transparent, and clinically trustworthy rather than opaque. Explainable AI frameworks shed light on how forecasts are generated and on the variables that drive therapeutic decisions by integrating insights from medicine, data science, ethics, and human-computer interaction. Improved interpretability helps clinicians better understand and validate AI outputs, enabling more informed clinical judgment, greater responsibility, and patient-centered treatment. Detecting bias, ensuring regulatory compliance, and promoting the safe incorporation of AI into real-world healthcare procedures also depend on explainability. For AI-driven solutions to gain traction in clinical practice and remain sustainable over the long run, it is crucial to build trust through systems that are easy to understand and use (18).

CONCLUSION

There is hope that AI systems of the future will improve in areas such as openness, adaptability, and integration with healthcare workflows. Improved interpretability may be possible with hybrid models that combine expert knowledge with data-driven learning. Safety, efficacy, and cost-effectiveness must be demonstrated through prospective clinical trials and real-world validation studies.

Responsible use of AI requires multidisciplinary teams that include doctors, data scientists, ethicists, and lawmakers. The ultimate objective is to supplement clinical reasoning with intelligence rather than replace it. By addressing complexity, data overload, and diagnostic uncertainty, artificial intelligence is transforming the way chronic disease differential diagnosis is performed. Artificial intelligence (AI) can greatly augment human clinical knowledge through data integration and sophisticated pattern recognition, leading to faster diagnoses, shorter wait times, and more individualized treatment plans. Integrating AI into the management of chronic diseases will require openness, ethical leadership, and ongoing partnership between medical professionals and technological advancements as the technology continues to develop.

ETHICAL DECLARATIONS

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