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EDITED BY Francesco Napolitano. University of Sannio, Italy

REVIEWED BY Francisco Tustumi, University of São Paulo, Brazil

Ailin 7hao ☑ irenez20@outlook.com Yijun Wu ⊠ wuyj01029@wchscu.cn

[†]These authors have contributed equally to this work and share first authorship

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Rare and complex diseases in focus: ChatGPT's role in improving diagnosis and treatment

Yue Zheng^{1†}, Xu Sun^{2†}, Baijie Feng³, Kai Kang¹, Yuqi Yang³, Ailin Zhao^{2*} and Yijun Wu^{1*}

¹Cancer Center, West China Hospital, Sichuan University, Chengdu, Sichuan, China, ²Department of Hematology, West China Hospital, Sichuan University, Chengdu, Sichuan, China, ³West China School of Medicine, Sichuan University, Chengdu, Sichuan, China

Rare and complex diseases pose significant challenges to both patients and healthcare providers. These conditions often present with atypical symptoms, making diagnosis and treatment a formidable task. In recent years, artificial intelligence and natural language processing technologies have shown great promise in assisting medical professionals in diagnosing and managing such conditions. This paper explores the role of ChatGPT, an advanced artificial intelligence model, in improving the diagnosis and treatment of rare and complex diseases. By analyzing its potential applications, limitations, and ethical considerations, we demonstrate how ChatGPT can contribute to better patient outcomes and enhance the healthcare system's overall effectiveness.

ChatGPT, artificial intelligence, rare and complex diseases, diagnosis, treatment

1 Introduction

Rare and complex diseases, often referred to as orphan diseases, affect a relatively small number of individuals but can have devastating effects on those afflicted. Due to the rarity and complexity of these conditions, they are frequently misdiagnosed or remain undiagnosed for extended periods, leading to delayed or inadequate treatment. However, the emergence of artificial intelligence (AI) and natural language processing (NLP) technologies has opened up new possibilities for addressing the diagnostic and therapeutic challenges associated with these diseases (Krishnan et al., 2023). One such example is idiopathic pulmonary arterial hypertension (IPAH), a rare and progressive lung disorder. AI-driven tools have been harnessed to scrutinize medical imaging data, such as echocardiograms, enabling early detection of IPAH and facilitating prompt intervention (Kiely et al., 2019). This demonstrates how the synergy of AI and NLP technologies provides promising solutions for tackling the diagnostic and therapeutic hurdles presented by rare and complex diseases, ultimately enhancing the quality of life for patients.

2 Discussion

In the realm of healthcare, ChatGPT, as an advanced language model, has demonstrated remarkable potential (Liu et al., 2023; Vaishya et al., 2023). AI and NLP tools empower healthcare professionals to rapidly analyze extensive volumes of medical literature and patient records, possibly aiding in the diagnosis and comprehension of rare diseases like Zheng et al. 10.3389/frai.2024.1338433

TABLE 1 Example of ChatGPT's possible contributions in helping diagnosing and treating rare and complex diseases.

| Disease | Possible contribution of ChatGPT |
|---|---|
| Idiopathic pulmonary Arterial hypertension | Early detection through artificial intelligence-driven analysis of medical imaging data |
| Klippel-trenaunay syndrome | Rapid analysis of extensive medical literature for better understanding |
| Early-onset Parkinson's disease | Analyzing patient symptoms, even vague or unconventional ones |
| Rett syndrome | Access to latest research findings and personalized treatment plans |

NLP, Klippel-Trenaunay syndrome, Early-Onset Parkinson's Disease, and Rett Syndrome (Table 1). ChatGPT, as a powerful language model, serves as a valuable assistant to physicians and researchers, facilitating access to relevant information, exploration of cutting-edge treatment modalities, and improved communication with patients (Figure 1).

One noteworthy capability of ChatGPT is its ability to analyze patient symptoms, even when they are vague or unconventional. By processing textual descriptions from patients or medical professionals, ChatGPT can generate relevant diagnostic suggestions and highlight potential indicators of rare diseases. Additionally, ChatGPT's proficiency in quickly accessing and summarizing vast amounts of medical literature helps physicians stay abreast of the latest research findings related to rare diseases, potentially leading to more accurate diagnoses (Lapidus, 2023). Additionally, AI technology holds promise in disease prediction and early screening (Au and Yang, 2023; Haver et al., 2023), facilitating the identification of pre-symptomatic signs of rare diseases by analyzing genetic data, biomarkers, and clinical information. This approach expedites early intervention and treatment implementation, potentially improving patient outcomes.

Furthermore, ChatGPT can contribute significantly to personalized treatment plans by considering individual patient data, medical history, and the latest clinical guidelines. It assists in tailoring treatment strategies that account for the unique characteristics of each rare disease case. Moreover, ChatGPT aids in expediting drug discovery for rare diseases by analyzing scientific literature and identifying potential drug candidates or existing medications with repurposing potential, thus reducing the time and cost associated with drug development (Alves et al., 2022).

However, it is important to note that the effectiveness of ChatGPT is contingent on the quality and diversity of the data it has been trained on. Bias in training data can lead to inaccurate recommendations and reinforce healthcare disparities (Aronson, 2023). Besides, ChatGPT's reliance on patterns learned from training data may pose challenges in comprehending the nuanced contextual aspects of individual patient situations. This limitation underscores the necessity for human involvement in interpreting and communicating decisions to patients, ensuring

a more accurate and empathetic understanding of their unique circumstances (Tustumi et al., 2023).

It's crucial to emphasize that while ChatGPT provides valuable assistance, it should not replace the expertise of medical professionals. Collaboration between AI and healthcare providers remains essential to ensure patient safety and the provision of quality care (Derevianko et al., 2023). Moreover, in recognizing the pivotal collaboration between AI and human expertise, the realistic scenario involves humans managing and collaborating with chatbots, rather than seeking outright substitution. Human oversight is indispensable for ensuring accuracy, ethical adherence, and a personalized touch in patient interactions. Human intervention plays a crucial role, especially in addressing complex or unconventional cases where ChatGPT's limitations in understanding context, emotions, and unique patient scenarios become apparent.

Ethical concerns surrounding patient data, algorithmic bias, and the responsible use of AI must be addressed to harness the full potential of these technologies in improving rare disease diagnosis and treatment while upholding patient rights and wellbeing (Dave et al., 2023; Liebrenz et al., 2023). This emphasis on ethical considerations ensures that the integration of AI in healthcare aligns with ethical standards and benefits patients without compromising their rights.

Looking ahead to the future, ChatGPT's role is poised to transcend language comprehension and expand into a comprehensive medical model. This evolution entails a multifaceted approach, encompassing text-based interactions, image recognition for assessing physical symptoms, and the capability to analyze uploaded medical images such as rare skin conditions or radiological scans for a holistic diagnosis. In this advanced medical application, ChatGPT would serve as a versatile healthcare assistant, capable of processing both textual and visual data. Patients and healthcare professionals would be able to interact with the model through text, voice, or image inputs, facilitating a more comprehensive understanding of patients' health concerns. For instance, when presented with textual descriptions of symptoms, ChatGPT can offer diagnostic suggestions and potential treatment options. Simultaneously, it can analyze uploaded images to identify visible physical symptoms, such as rare skin diseases or other external manifestations, further enhancing the diagnostic process. Additionally, ChatGPT can process complex medical imaging data, helping healthcare providers in identifying and evaluating intricate conditions, from rare diseases to anomalies detected in radiological scans (Bhayana et al., 2023). This expanded role of ChatGPT in the medical domain holds the promise of improving the accuracy and efficiency of diagnosis and treatment, particularly in the case of rare and complex diseases. However, it is crucial to address ethical and privacy considerations rigorously, ensuring the responsible use of patient data and maintaining a strong collaboration between AI and healthcare professionals. As AI technologies continue to advance, their integration into comprehensive medical models like ChatGPT represents a significant step forward in enhancing healthcare delivery and patient outcomes.

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I am experiencing extreme fatigue, joint swelling, and frequent mouth ulcers. ChatGPT, can you help me identify which rare disease these symptoms might be related to and provide treatment information?

 $\mbox{l'm}$ not a doctor, but \mbox{l} can suggest some potential conditions that could be associated with the symptoms you've described, including extreme fatigue, joint swelling, and frequent mouth ulcers. These symptoms can be indicative of several different medical conditions, including some rare diseases. However, it's crucial to consult with a healthcare professional for a proper diagnosis and appropriate treatment. Here are a few possibilities to consider:

- Behçet's Disease: Behçet's disease is a rare autoimmune disorder that can cause a variety of symptoms, including mouth ulcers, joint swelling, and extreme fatigue. Treatment typically involves medications to control inflammation, manage symptoms, and suppress
- 2. Systemic Lupus Erythematosus (SLE): Lupus is another autoimmune disease that can cause fatigue, joint swelling, and mouth ulcers. Treatment may involve a combination of medications, including anti-inflammatory drugs and immunosuppressants, to manage the condition and alleviate symptoms.
- 3. Sjögren's Syndrome: Sjögren's syndrome is an autoimmune disorder primarily affecting the salivary and tear glands, leading to dry mouth and dry eyes. Joint pain and fatigue can also be present. Treatment focuses on relieving symptoms and may include artificial tears and medications to manage joint pain.
- Vasculitis: Various forms of vasculitis, including ANCA-associated vasculitis, can present with joint swelling, fatigue, and mouth ulcers. Treatment depends on the type and severity of vasculitis but often involves immunosuppressive medications to control inflammation.
- 5. Other Rare Autoimmune Diseases: There are many rare autoimmune diseases that can manifest with these symptoms. Consulting with a rheumatologist or autoimmune specialist is crucial for a thorough evaluation and diagnosis.
- 6. Other Rare Conditions: While less common, there are also non-autoimmune rare diseases that can cause similar symptoms. These may include certain genetic or metabolic disorders. A medical specialist would be best equipped to explore these possibilities.

It's essential to seek medical attention to determine the exact cause of your symptoms. A healthcare provider will conduct a comprehensive evaluation, including a physical examination and possibly blood tests or other diagnostic tests, to reach a precise diagnosis Once a diagnosis is established, your healthcare team can recommend an appropriate treatment plan tailored to your condition.

Remember that early diagnosis and treatment are crucial for managing these conditions effectively, so don't hesitate to reach out to a healthcare professional for help.

I frequently experience difficulty breathing, especially at night. ChatGPT, what rare disease do you think this might be, and can you provide treatment information?

I'm not a doctor, but I can suggest some potential causes of nighttime difficulty breathing. If you're experiencing this symptom, it's essential to consult a healthcare professional for a proper evaluation and diagnosis. Nighttime breathing difficulties can be related to various medical conditions, including rare diseases, and they require individualized assessment and treatment. Here are a few possibilities to consider:

- Obstructive Sleep Apnea (OSA): OSA is a common sleep disorder characterized by repeated interruptions in breathing during sleep. It can lead to nighttime difficulty breathing, snoring, and excessive daytime sleepiness. Treatment options include lifestyle changes, continuous positive airway pressure (CPAP) therapy, and surgery in some cases.
- 2. Congenital Central Hypoventilation Syndrome (CCHS): This rare genetic disorder affects the autonomic nervous system's control of breathing. It can result in hypoventilation and difficulty breathing, especially during sleep. Management often involves lifelong ventilatory
- Idiopathic Pulmonary Fibrosis (IPF): IPF is a rare lung disease that causes scarring of lung tissue. It can lead to progressive breathing difficulties, particularly during exertion and sleep. Treatment may include medications to slow the disease's progression, supplemental oxygen, and lung transplant evaluation in severe cases.
- Congenital Disorders of the Central Nervous System: Some rare congenital neurological disorders can affect breathing control and may lead to nighttime breathing difficulties. Treatment depends on the specific disorder and may involve specialized care and interventions.
- Other Rare Respiratory Conditions: There are many rare respiratory diseases and syndromes, such as various interstitial lung diseases, that can cause nighttime breathing problems. Treatment options vary depending on the specific condition and its severity.

It's crucial to seek medical attention if you're experiencing nighttime difficulty breathing, as it can be a sign of a serious underlying condition. A healthcare provider will conduct a thorough evaluation, which may include a sleep study, pulmonary function tests, and imaging studies, to determine the cause of your symptoms. Once a diagnosis is made, they can recommend an appropriate treatment plan tailored to your specific condition and needs.

Please consult a healthcare professional for an accurate diagnosis and guidance on managing your symptoms.

Example of ChatGPT in helping diagnosing and treating rare and complex diseases.

In conclusion, ChatGPT and AI/NLP technologies offer innovative solutions for addressing the diagnostic and therapeutic challenges presented by rare and complex diseases. By leveraging these advancements, healthcare professionals can enhance their ability to diagnose rare diseases promptly and accurately, customize treatments, and accelerate drug discovery. However, it is imperative to address limitations and ethical considerations associated with AI in healthcare, ensuring both patient safety and data privacy. As AI continues to evolve, its role in managing rare diseases is likely to expand, ultimately benefiting patients and advancing our understanding of these complex conditions.

Data availability statement

The original contributions presented in the study are included in the article/supplementary material, further inquiries can be directed to the corresponding authors.

Author contributions

YZ: Conceptualization, Investigation, Writing—original draft. XS: Investigation, Writing-original draft. BF: Investigation, Software, Writing-original draft. KK: Investigation, Software, Writing—original draft. YY: Data curation, Validation, Writing review & editing. AZ: Conceptualization, Funding acquisition, Project administration, Resources, Supervision, Validation, Writing-review & editing. YW: Conceptualization, Funding acquisition, Project administration, Resources, Supervision, Validation, Writing—review & editing.

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Conflict of interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships

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References

Alves, V. M., Korn, D., Pervitsky, V., Thieme, A., Capuzzi, S. J., Baker, N., et al. (2022). Knowledge-based approaches to drug discovery for rare diseases. *Drug Discov. Today* 27, 490–502. doi: 10.1016/j.drudis.2021.10.014

Aronson, J. K. (2023). When I use a word . . . ChatGPT: a differential diagnosis. *Bmj* 382, 1862. doi: 10.1136/bmj.p1862

Au, K., and Yang, W. (2023). Auxiliary use of ChatGPT in surgical diagnosis and treatment. *Int. J. Surg.* 109, 3940–3943. doi: 10.1097/JS9.0000000000000686

Bhayana, R., Krishna, S., and Bleakney, R. R. (2023). Performance of ChatGPT on a radiology board-style examination: insights into current strengths and limitations. *Radiology* 307, e230582. doi: 10.1148/radiol.230582

Dave, T., Athaluri, S. A., and Singh, S. (2023). ChatGPT in medicine: an overview of its applications, advantages, limitations, future prospects, and ethical considerations. *Front. Artif. Intell.* 6, 1169595. doi: 10.3389/frai.2023.1169595

Derevianko, A., Pizzoli, S. F. M., Pesapane, F., Rotili, A., Monzani, D., Grasso, R., et al. (2023). The use of artificial intelligence (AI) in the radiology field: what is the state of doctor-patient communication in cancer diagnosis? *Cancers* 15, 470. doi: 10.3390/cancers15020470

Haver, H. L., Ambinder, E. B., Bahl, M., Oluyemi, E. T., Jeudy, J., and Yi, P. H. (2023). Appropriateness of breast cancer prevention and screening recommendations provided by ChatGPT. *Radiology* 307, e230424. doi: 10.1148/radiol.230424

Kiely, D. G., Doyle, O., Drage, E., Jenner, H., Salvatelli, V., Daniels, F. A., et al. (2019). Utilising artificial intelligence to determine patients at risk of a rare disease: idiopathic pulmonary arterial hypertension. *Pulm. Circ.* 9, 2045894019890549. doi: 10.1177/2045894019890549

Krishnan, G., Singh, S., Pathania, M., Gosavi, S., Abhishek, S., Parchani, A., et al. (2023). Artificial intelligence in clinical medicine: catalyzing a sustainable global healthcare paradigm. *Front. Artif. Intell.* 6, 1227091. doi: 10.3389/frai.2023.1227091

Lapidus, D. (2023). Strengths and limitations of new artificial intelligence tool for rare disease epidemiology. *J. Transl. Med.* 21, 292. doi: 10.1186/s12967-023-04152-0

Liebrenz, M., Schleifer, R., Buadze, A., Bhugra, D., and Smith, A. (2023). Generating scholarly content with ChatGPT: ethical challenges for medical publishing. *Lancet Digit. Health* 5, e105–e06. doi: 10.1016/S2589-7500(23)00019-5

Liu, J., Wang, C., and Liu, S. (2023). Utility of ChatGPT in clinical practice. *J. Med. Int. Res.* 25, e48568. doi: 10.2196/48568

Tustumi, F., Andreollo, N., and Aguilar-Nascimento, J. (2023). Future of the language models in healthcare: the role of chatGPT. *Arq. Bras. Cir. Dig.* 36, e1727. doi: 10.1590/0102-672020230002e1727

Vaishya, R., Misra, A., and Vaish, A. (2023). ChatGPT: is this version good for healthcare and research? *Diabetes Metab. Syndr.* 17, 102744. doi: 10.1016/j.dsx.2023.102744