Ear, Nose & Throat Journal
1–3

DOI: 10.1177/01455613251335385
journals.sagepub.com/home/ear
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Artificial Intelligence-Assisted Diagnosis of an Unusual Cause of Periodic Epistaxis: A Case Report

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Abstract

Epistaxis is a common cause of consultation in otolaryngology, manifesting either as isolated episodes or recurrent events. In this paper, we report the history of an 8-year-old African male who presented with periodic epistaxis temporally associated with joint inflammation. The diagnostic work-up, supported by artificial intelligence-based clinical decision support, led to the diagnosis of rheumatic fever. This case describes a very rare etiology of periodic epistaxis and demonstrates the potential usefulness of artificial intelligence as an adjunctive clinical tool. To our knowledge, this is the first reported case of rheumatic fever presenting with periodic epistaxis in the pediatric population.

Keywords

rheumatic, fever, otolaryngology, epistaxis, nasal, bleeding, hemorrhage, periodic

Introduction

Epistaxis is one of the most frequent emergencies in otolaryngology-head and neck surgery department with a prevalence of *10%-12%* in the general population, and an annual incidence of 30/100.000 in adults and 2.4/1000 in children.[1,2] Epistaxis may occur in up to 9% of children and it is commonly associated with local inflammation and trauma, including nose picking.[3] In some cases, epistaxis can be a manifestation of an inflammatory systemic disease in children, including hereditary hemorrhagic telangiectasia,[4] tuberculosis,[5] Cushing syndrome,[6] or vascular malformation disease.[7] Pediatric epistaxis typically presents as either isolated events or recurrent episodes with irregular intervals, rarely demonstrating a predictable temporal pattern.[3,8]

In this paper, we report the history of an African child male who presented with periodic epistaxis temporally associated with joint inflammation.

Case Report

An 8-year-old African male presented to the otolaryngology consultation with periodic epistaxis temporally associated with joint inflammation. The articular manifestations were characterized by polyarthralgia and pyrexia (>38.5°C), with a distinctive periodicity of 3-week intervals between episodes. The patient resided with his family in a rural region in the Kenyan mountains (Rift Valley). Past medical history revealed recurrent episodes of rhinitis and pharyngitis over the past few years, without

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Received: March 10, 2025; revised: March 24, 2025; accepted: March 31, 2025

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microbiological documentation. The surgical history was unremarkable. The child did not receive a vaccine in childhood. The clinical examination was unremarkable. The most recent epistaxis and articular inflammation occurred 2 weeks prior to presentation. Therefore, at the consultation time, the clinical examination was unremarkable. Regarding the uncommon clinical presentation, the patient history and findings were entered into Q-Exp-Claude-3.5-2kk large language model through Poe® platform (Anthropic Inc., San Francisco, USA). The tuned large language model suggested a potential atypical presentation of a rheumatic fever according to the presence of one clinical major criterion (arthritis) and two minor criteria (fever ≥38.5°C, and arthralgia). The Anti-Streptolysin O Titer (ASOT) confirmed the diagnosis. A penicillin G injection was performed in the child and the symptoms did not recur. Written informed consent was obtained from the patient's legally authorized representative.

Discussion

In Western developed countries, the prevalence of acute rheumatic fever (ARF) has significantly decreased in the past few decades due to improvements in healthcare access, the use of antibiotics, and public screening programs.[9] To date, the incidence of ARF is estimated between 1 and 4/100,000 children in Europe[9] and the United States.[10] The clinical presentation of ARF consists of major (eg, periodic fever with articulation inflammation (arthritis), chorea, carditis, erythema marginatum, Subcutaneous nodules) and minor (fever (≥38.5°C) and arthralgia) findings.[11] Although this patient presented with one major criterion and two minor criteria, the clinical presentation was atypical regarding the periodic epistaxis leading to a consultation at the ear, nose, and throat office. To the best of our knowledge, there is no mention in the literature of periodic epistaxis due to ARF. The periodic epistaxis was related to the pathophysiological cycle of the disease. From a pathophysiological standpoint, post-streptococcal molecular mimicry initiates cross-reactive autoantibodies against cardiac and synovial tissues, precipitating a T-cell mediated inflammatory cascade with elevated pro-inflammatory cytokines (eg, IL-6, TNF-α, and IL-1β).[12] The resultant immune-complex deposition and complement activation generate a cyclic pattern of tissue inflammation manifesting as periodic synovitis and pyrexia, with the latter precipitating episodes of epistaxis due to vasomotor alterations.[12] This autoimmune phenomenon persists through episodic activation of memory T cells and sustained production of anti-streptococcal lysoganglioside antibodies, maintaining the inflammation-remission cycle until immunomodulatory intervention disrupts the pathological sequence.[12]

The increase in medical knowledge and the rarity of ARF make the diagnosis difficult. In the present paper, we report the interest of high-powered large language models as an adjunctive tool in clinical practice. While using artificial intelligence software for diagnosing rare or complicated diseases has not reached a consensus in clinical practice, we believe that its use was crucial in reaching the correct diagnosis of this rare disease. Poe® platform allows access to a large number of tuned large language models, which were evaluated in numerous studies for diagnosis accuracy.[13] A recent state-of-the-art review reported that regardless of the subspecialty, ChatGPT-4 reported the highest accuracy in providing a plausible primary diagnosis for common otolaryngological cases (47%-79%).[13] Concerning rare diseases, Claude-3.5-Sonnet demonstrated superior performance in diagnosis (54.3%), significantly outperforming Gemini-1.5-Pro (28.6%) and surpassing ChatGPT-4o (45.7%).[14] The authors selected Claude-3.5-Sonnet due to its superior performance compared to other chatbots. A substantial number of other chatbots are still available, including Perplexity and Fusion.ai, but studies of the literature mainly assessed ChatGPT, Claude, and Gemini in providing information for real clinical cases.[13] Although AI should never replace clinical judgment, our experience suggests that these tools could serve as adjunctive tools for diagnosing rare conditions in the future, particularly when pattern recognition across large medical knowledge bases is required. Nevertheless, this approach requires careful validation and should be incorporated within proper clinical reasoning and expert medical supervision.

Conclusion

To our knowledge, this is the first reported case of rheumatic fever presenting with periodic epistaxis in the pediatric population. The artificial intelligence was used as adjunctive clinical tool to make the diagnosis.

Acknowledgments

Dr. Johnson for proofreading the paper.

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Ethical Considerations

The IRB was not required for the publication of this case report. Written informed consent was obtained from the patient's legally authorized representative.

Author Contributions

AM, LAV, SS, and SH: conception and design, analysis and interpretation of data, and drafting of the manuscript; JRL and AM: revising the manuscript for important intellectual content, and final approval of the version to be published.

Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

Declaration of Conflicting Interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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