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Tofacitinib for Atypical SAPHO Syndrome in an Adolescent: A Case Report

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Abstract

Background: SAPHO (synovitis, acne, pustulosis, hyperostosis, and osteitis) syndrome is a rare autoinflammatory disorder that most typically involves the anterior chest wall. In adolescents, however, osteoarticular lesions may predominantly affect the long bones, with normal inflammatory markers, constituting an atypical presentation that is easily overlooked. Optimal treatment for such presentations remains to be defined. Case presentation: We report a 14-year-old female adolescent who initially presented with refractory desquamation of both feet, which progressed to miliary sterile pustules consistent with palmoplantar pustulosis (PPP), accompanied by lower-extremity bone pain. Inflammatory markers were within normal limits, whereas magnetic resonance imaging (MRI) demonstrated osteitis of the bilateral femoral metadiaphyses, Computed tomography (CT) demonstrated sparing of the sternoclavicular joints. According to the Kahn (1994) diagnostic criteria, a diagnosis of SAPHO syndrome with an atypical osteoarticular distribution was established. Treatment and outcome: The skin lesions had relapsed upon discontinuation of prior topical agents and traditional Chinese medicine. The patient was therefore treated with oral tofacitinib (5 mg once daily). After five months, the plantar lesions had improved markedly, with reduced scaling, and the lower-extremity bone pain had subsided. No adverse effects were observed during treatment. Conclusion: This case illustrates an atypical adolescent presentation of SAPHO syndrome, characterized by palmoplantar pustulosis with bilateral femoral osteitis but without anterior chest wall involvement or elevated inflammatory markers, and documents a favorable response of both

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cutaneous and skeletal manifestations to tofacitinib. These findings support the potential of JAK inhibitors as a therapeutic option for SAPHO syndrome, although further studies are warranted.

Keywords

SAPHO Syndrome, Tofacitinib, Palmoplantar Pustulosis, JAK-STAT Pathway, Adolescenty

1. Introduction

SAPHO syndrome is a rare autoinflammatory disorder characterized by synovitis, acne or pustulosis, hyperostosis, and osteitis, arising from aberrant cytokine production by innate immune cells [1] [2]. The anterior chest wall is the most frequently affected osteoarticular site, and sternoclavicular hyperostosis is regarded as a typical radiological feature [1] [3]. In children and adolescents, however, the disease often follows an atypical course: lesions may be confined to the long bones of the limbs in a pattern resembling chronic recurrent multifocal osteomyelitis (CRMO), the sternoclavicular joints may be entirely spared, and inflammatory markers may remain within normal limits, all of which contribute to delayed or missed diagnosis [3]. In addition, no standardized treatment has been established for SAPHO syndrome; recent reports describing the successful use of the Janus kinase (JAK) inhibitor tofacitinib in refractory cases, including a young adult, suggest a promising therapeutic avenue [4]-[6]. Here, we report an adolescent with atypical SAPHO syndrome, manifesting as palmoplantar pustulosis associated with bilateral femoral osteitis but without sternal involvement, who was successfully treated with tofacitinib, and we discuss the diagnostic and therapeutic implications of this presentation.

2. Presenting History

A 14-year-old female adolescent initially presented with peeling of the skin of both feet in December 2022. The lesions progressively worsened, with the development of scattered pustules. She had been treated with topical agents and traditional Chinese medicine soaks, neither of which provided significant relief. In 2024, oral traditional Chinese herbal decoctions were initiated, and transient improvement was observed within a few days. However, upon discontinuation of this treatment in July 2024, the cutaneous symptoms rapidly recurred and worsened, accompanied by new-onset bone pain in the lower extremities that were aggravated by activity, without local redness, warmth, or fever.

3. Physical Examination and Investigations

Physical examination revealed scaling and scattered miliary sterile pustules on the soles, with hyperkeratosis and desquamation in some areas, consistent with the

typical presentation of palmoplantar pustulosis (PPP). Laboratory tests showed inflammatory markers within normal limits: C-reactive protein (CRP) < 5.00 mg/L (reference range 0 - 8 mg/L) and erythrocyte sedimentation rate (ESR) 6.00 mm/h (reference range 0 - 20 mm/h). Magnetic resonance imaging (MRI) demonstrated osteitis involving the bilateral femoral metadiaphyses, with lesions showing low signal on T1-weighted imaging (T1WI) and patchy high signal on short-tau inversion recovery (STIR) sequences, suggestive of bone marrow edema, without evident sequestrum or abscess formation. CT showed that the sternoclavicular joints were spared. According to the Kahn (1994) diagnostic criteria, the patient fulfilled criterion 3 (any sterile osteitis associated with palmoplantar pustulosis), thereby confirming the diagnosis of SAPHO syndrome [7].

4. Treatment and Outcome

Given the relapse of symptoms upon discontinuation of prior therapies and the confirmed diagnosis of SAPHO syndrome, the patient was started on oral tofacitinib 5 mg once daily after informed consent was obtained. After five months of treatment, the plantar lesions had improved significantly, with reduced scaling, and pustule formation, and the lower-extremity bone pain had subsided (**Figure 1**). No adverse effects were observed during treatment.

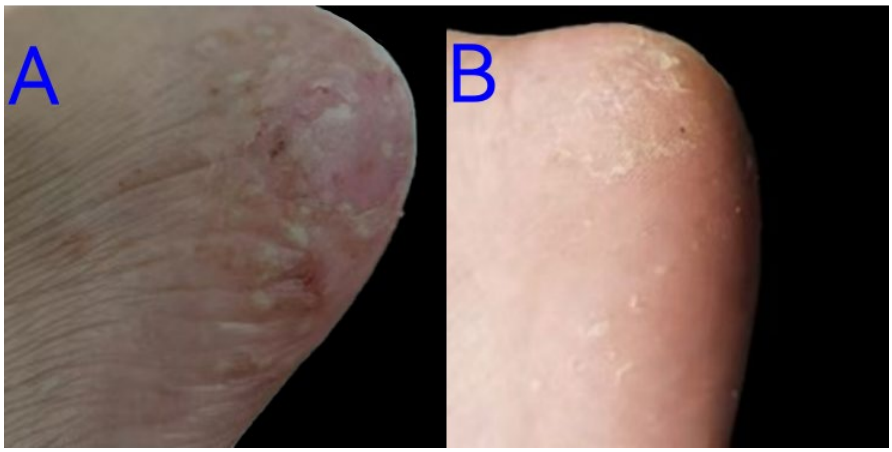

Figure 1. Clinical improvement of plantar lesions in palmoplantar pustulosis after tofacitinib treatment. (A) Before treatment, extensive desquamation and pustules are visible on the heel and sole. (B) After 5 months of treatment, the lesions show prominent re-epithelialization with markedly reduced scaling.

5. Discussion

The present case is noteworthy for two reasons: the atypical osteoarticular distribution of SAPHO syndrome in this adolescent, and the favorable response of both the cutaneous and skeletal manifestations to tofacitinib.

Osteoarticular involvement in SAPHO syndrome most commonly affects the anterior chest wall, with sternoclavicular hyperostosis as a typical manifestation; however, not all patients have sternoclavicular involvement [1] [3]. In children

and adolescents, lesions may be confined predominantly to the metaphyses of the long bones of the limbs, presenting as a chronic recurrent multifocal osteomyelitis (CRMO)-like pattern, in which the sternoclavicular joints may be entirely normal [3]. Consistent with this adolescent-type presentation, our patient had no sternal pain, the sternoclavicular joints were spared, and the lesions were located in the bilateral femoral metadiaphyses. In addition, her inflammatory markers remained within normal limits despite active osteitis, which further departed from the classic picture and increased the risk of diagnostic delay. Recognition of this atypical presentation is therefore essential to avoid misdiagnosis as infectious osteomyelitis or other dermatoses.

SAPHO syndrome is characterized by elevated levels of multiple proinflammatory cytokines. Among these, cytokines such as IL-6 and IL-23 signal directly through the JAK-STAT pathway, whereas TNF-alpha signals mainly through the NF-kappaB pathway and IL-17 mainly through the ACT1/TRAF6-NF-kappaB and MAPK pathways [2]. Tofacitinib, a JAK1/3 inhibitor, competitively binds ATP and blocks JAK kinase activity, thereby inhibiting downstream STAT phosphorylation and attenuating Th17 cell differentiation and the production of multiple inflammatory cytokines [8]-[10]. This mechanism may account for its therapeutic effect on both the cutaneous and skeletal manifestations of SAPHO syndrome. Indeed, favorable outcomes with tofacitinib have previously been reported in adults with refractory SAPHO syndrome [4] [5] and, more recently, in an 18-year-old patient [6]. The present observations extend this evidence to a younger adolescent with an atypical long-bone distribution of osteitis.

Several limitations of this report should be acknowledged. As a single-case observation with a relatively short follow-up period, the efficacy and safety of tofacitinib cannot be generalized, and the possibility of spontaneous fluctuation in disease activity cannot be excluded. Larger studies with longer follow-up are required to establish the role of JAK inhibitors in the management of SAPHO syndrome, particularly in pediatric patients with atypical presentations.

6. Conclusion

This case describes an adolescent with atypical SAPHO syndrome characterized by palmoplantar pustulosis and bilateral femoral osteitis without sternal involvement or elevation of inflammatory markers, in whom both the cutaneous and skeletal manifestations improved substantially after tofacitinib treatment. These findings highlight the importance of recognizing atypical osteoarticular presentations of SAPHO syndrome in adolescents and suggest that JAK inhibitors may represent a valuable therapeutic option, although their exact mechanism and long-term efficacy require further investigation.

Data Availability

The data supporting the findings of this study are available from the corresponding author upon reasonable request.

Consent to Participate

Written informed consent was obtained from the patient and her legal guardian.

Consent to Publication

Written informed consent for publication of this case report and any accompanying images was obtained from the patient and her legal guardian.

Ethics Statement

In compliance with Chinese laws and relevant institutional regulations, this retrospective case analysis is exempt from ethical review. The study was conducted in accordance with local legislative and institutional requirements. Written informed consent was obtained from the patient and her legal guardian for participation in this study and for the publication of any potentially identifiable images or data included in this article.

Author Contributions

Yini Li (YL): Data curation, Writing original draft, Conceptualization, Formal Analysis, Methodology, Writing review & editing. Junye Liu (JL): Investigation, Writing original draft, Conceptualization, Resources, Visualization, Writing review & editing. Yan Ma (YM): Data curation, Visualization, Investigation, Resources, Formal Analysis, Writing original draft, Writing review & editing.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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