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Case report: A rare case of SAPHO syndrome mimicking chronic osteomyelitis

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Abstract
SAPHO syndrome is a rare inflammatory disorder characterized by a constellation of osteoarticular and dermatological manifestations, including Synovitis, Acne, Pustulosis, Hyperostosis, and Osteitis. It often poses a diagnostic challenge due to its overlapping features with other musculoskeletal conditions.
Chronic Non-Bacterial Osteomyelitis (CNO) is a rare autoinflammatory bone disorder, with a reported prevalence of approximately 1 in 1,000,000 individuals. It typically presents in children and adolescents and is characterized by sterile bone inflammation, often mimicking infectious osteomyelitis.
We present a rare case of a 16-year-old boy who initially presented with clinical and radiological features suggestive of chronic non-bacterial osteomyelitis. However, further evaluation, including the presence of cutaneous manifestations and imaging findings, led to a diagnosis of SAPHO syndrome, mimicking chronic osteomyelitis. This case highlights the importance of considering SAPHO syndrome in the differential diagnosis of chronic osteomyelitis, especially in paediatric populations with atypical clinical courses.

Keywords: SAPHO syndrome, Chronic Nonbacterial Osteomyelitis, synovitis, hyperostosis

Introduction
Chronic nonbacterial osteomyelitis (CNO) is a bone disorder characterized by inflammatory changes commonly seen in adolescent age group and children, characterized by a waxing and waning course [1]. When CNO is associated with cutaneous features such as acne and pustulosis, it is referred to as SAPHO syndrome (Synovitis, Acne, Pustulosis, Hyperostosis, Osteomyelitis) [2, 3].
One of the main challenges in diagnosing CNO is that its clinical features may occur many years apart, and patients may not exhibit all manifestations. There is no globally accepted diagnostic criteria or blood markers for CNO, hence it remains a diagnosis of exclusion. Though it is a benign disease, mild to debilitating complications including growth arrest is seen.
We should consider other differential diagnosis like malignancies, chronic osteomyelitis, lymphomas in these cases [4, 5]. Delayed diagnosis, often due to lack of leading investigations, can lead to negative outcomes including chronic pain, disfigurement and joint destruction. Herein, we present a case of CNO in an adolescent boy that closely mimics chronic osteomyelitis in all aspects without any microbiological positivity.

Case presentation
16 year old boy presented to our hospital with the complaints of pain, swelling, redness over the right foot for the past 3 years. The patient gives a past history of twisting injury to the right foot while he was playing and no history of penetrating trauma to the foot. For the past 3 years, patient had waxing and waning course of the symptoms associated with fever sometimes. Whenever patient developed the symptoms he was managed by conservative therapy and the symptom settled for a temporary phase and recurred at some interval. Patient did not give any history of chronic drug intake, chronic diseases or any other immuno-compromised state. The boy had normal growth milestones, immunized up to date and no significant history of prior hospitalization.
On examination of the right foot, we found that the third and fourth toe was deformed and hyperpigmented.

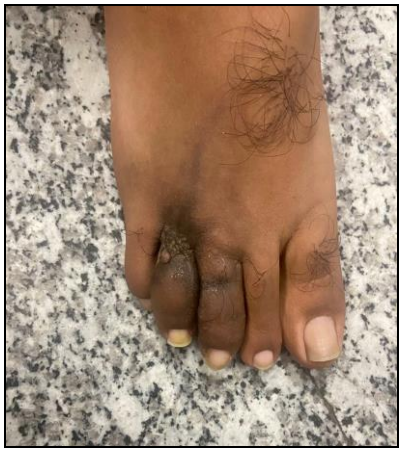

Fig 1: clinical photograph of right foot showing swelling, hyperpigmentation, hyperhidrosis,pustules over the 3rd and 4th toe

Blood investigations showed raised ESR (erythrocyte sedimentation rate) at the rate of 52 m per hour and a normal CRP (C reactive protein) 2 mm/dl. Other rheumatological investigations like RA factor, ANA profile, serum uric acid and immunological work up were normal. Patient also evaluated for metabolic bone diseases and all parameters were normal including bone scan. Patient initially evaluated for tubercular infection, atypical bacterial infections, fungal disease and oncology opinion was sought but all came out to normal.

Initial radiological investigations at the time of onset of symptoms were normal. There was cortical erosion of the proximal phalanges of the 3rd and 4th toe on follow up X rays (Fig 2). On repeat MRI diffuse bone marrow edema of the third metatarsal shaft, mild over the fourth metatarsal, mild effusion involving third metatarsophalangeal joint and moderate surrounding soft tissue edema was noted. Recent repeat MRI scan revealed osteitis of the proximal phalanx of the 3 rd toe with myositis surrounding it and subtle cortical erosions.

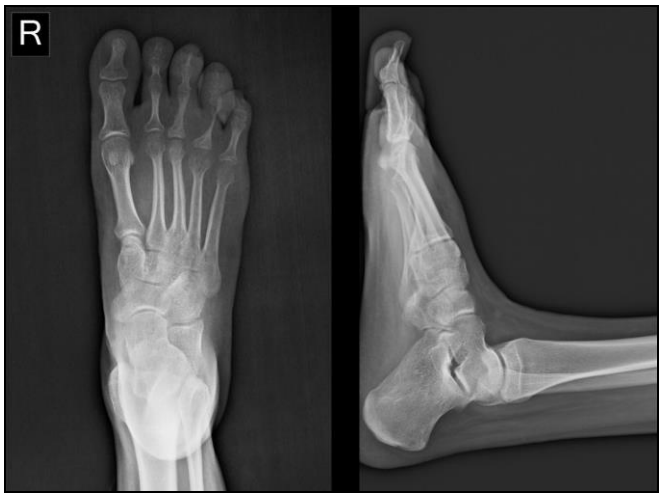

Fig 2: pre op x ray showing cortical erosion and thinning of 3rd and 4th proximal phalanx and distal phalanx

3rd toe proximal phalanx biopsy sections showed sparse intervening hemorrhage and no signs of granuloma or atypical cells. Skin biopsy revealed squamous papillomatous lesion showing mild hyperkeratosis and epithelial hyperplasia, the submucosal tissue with collection of acute and chronic inflammatory cells, no signs of dysplasia or malignancy. Patient drastically improved with surgical debridement of the proximal phalanges and surrounding soft tissues and supportive therapy (Fig 3). Patient is on a remission now.

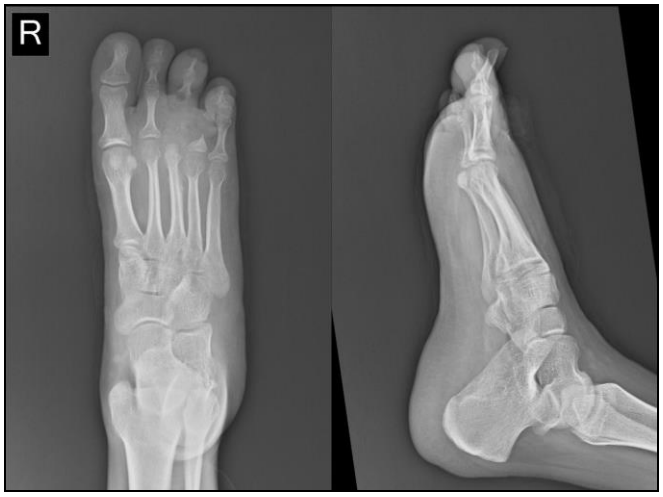

Fig 3: post op x ray after debridement

Discussion

This case underscores the importance of considering CNO as a key differential diagnosis for bacterial osteomyelitis, highlighting its variable clinical manifestations. The symptoms of warmth, redness, swelling, and pain can complicate differentiation from bacterial osteomyelitis. Although swelling is generally less pronounced in bacterial osteomyelitis, cases involving septic arthritis in affected joints are not uncommon. While fever absence is often seen as a distinguishing factor, some studies have noted mild to moderate fever in children with SAPHO syndrome. We also observed extra-articular features like acne and hyperpigmented pustular lesions, which are commonly associated with CNO and can aid in diagnosis. SAPHO tends to affect various bones but shows a preference for the lower extremities, particularly the metaphysis of long bones. However, these characteristics alone are insufficient to rule out bacterial osteomyelitis.

A variety of laboratory tests are used to aid in diagnosing bacterial osteomyelitis, though they often lack specificity. Elevated inflammatory markers like C-reactive protein and erythrocyte sedimentation rate are typically used as supplementary tools for diagnosis and treatment. However, in this case, increased ESR and CRP levels were also observed in SAPHO, diminishing their diagnostic utility. On the other hand, persistently normal CRP and ESR levels generally suggest the absence of osteomyelitis.[4]

Initial treatment for SAPHO often includes antibiotics before confirmation of the diagnosis, potentially delaying proper treatment and increasing the risk of complications. Late diagnosis of nonbacterial osteomyelitis can lead to significant morbidity. Although CNO is more often diagnosed in teenagers, children outside this age range should be assessed for possible underlying malignancies, such as lymphoma in adults or leukemia in younger children.[5]

The diagnosis of CNO becomes more definitive when characteristic skin lesions are present alongside typical bone lesions on MRI. Nonetheless, a conclusive diagnosis of bacterial osteomyelitis or bone malignancy still depends on positive bone culture results and histopathological examination. For children with persistent or atypical symptoms, and when diagnostic resources are available, whole-body MRI can be a useful tool to detect clinically silent CRMO lesions in other areas.

Conclusion

SAPHO syndrome is a diagnosis of exclusion and need a multidisciplinary approach to diagnose it involving many specialties. A delay in diagnosis results in heavy financial burden and poor quality of life in patients suffering from it. Although the diagnosing it at earlier stage is challenging, early treatment results in favorable outcome.

Conflict of Interest

Not available

Financial Support

Not available

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