Case Report

Clinical and Radiological Identification and Management of SAPHO Syndrome

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Abstract

Introduction
Synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome is an autoinflammatory disease whose acronymic name stands for the symptoms commonly seen in the disease. These symptoms typically occur simultaneously, in different combinations, in patients during exacerbations of the disease. SAPHO syndrome is a rare disease, most frequently seen in patients aged 30 to 50. It is estimated to be found in 1 in 10 000 persons in White populations, with an even lower incidence seen in non-White populations.

Case Presentation
A post-menopausal woman with a medical history of SAPHO presented to the emergency department with left foot pain and chest pain with palpitation. She had chronic pustular rashes, located on the bilateral soles of the feet, bilateral palms, neck, and abdomen, which she stated appeared hours after the initial presentation of her pain. Chest X-ray readings showed osteitis and sclerosis of the sternocostoclavicular joint and first rib, a radiological finding of SAPHO syndrome. The patient was successfully treated with nonsteroidal anti-inflammatory drugs for pain relief and IV corticosteroids for the rheumatoid-like inflammatory aspect of SAPHO.

Conclusion
While SAPHO syndrome is a rare disease, it is important to be aware of its manifestations and symptoms, such as the patient's rash, foot and chest pain, in addition to the dermatological symptoms appearing simultaneously. An early diagnosis can provide patients with accurate and appropriate treatment.

Keywords
SAPHO syndrome; acquired hyperostosis syndrome; synovitis; acne; pustulosis; hyperostosis; osteitis; case reports

Introduction
Synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome is a rare autoinflammatory disease with 5 primary symptoms that can be present at the time of diagnosis. During exacerbations of the disease, patients commonly experience the dermatological (acne and pustulosis) and osteoarticular (synovitis, hyperostosis, and osteitis) symptoms that it is named after, making it unique and which typically occur simultaneously and in different combinations. SAPHO syndrome is a rare disease, most frequently seen in adolescence to late adulthood, with the median age between 30 to 40 years old.[1] It is estimated to be found in 1 in 10 000 of White populations, with an even lower incidence seen in non-White populations.[2] In the case of our patient who has a known case of SAPHO syndrome and experienced a flare-up of its typical symptoms, the presentation of a rare disease can raise awareness and educate physicians on the treatment

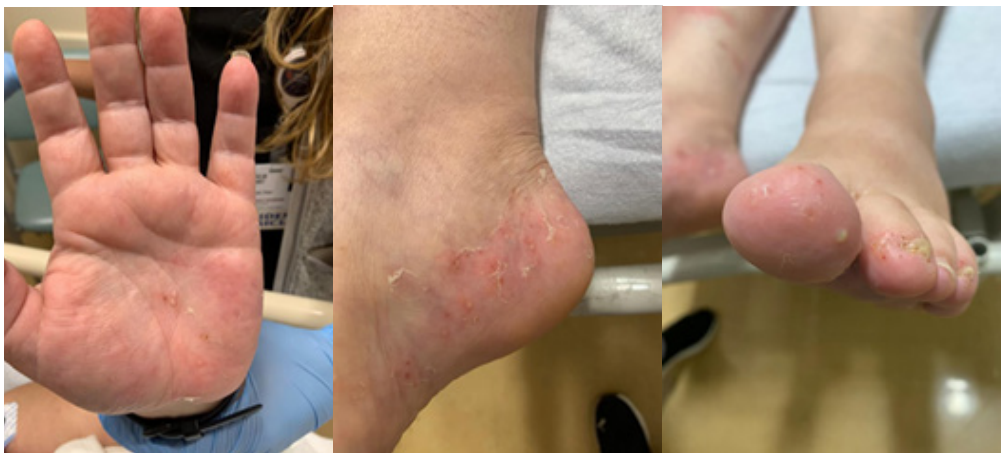

Figure 1. A physical exam revealed dermatological pustulosis on multiple areas of the body, a sign of synovitis, acne, pustulosis, hyperostosis, and osteitis syndrome.

while helping differentiate the disease from other common rheumatic diseases. Primarily with bone manifestations, SAPHO is typically a diagnosis of exclusion that is frequently misdiagnosed because of its variable presentations.[3] It is important for clinicians to be aware of the manifestations so that patients with SAPHO syndrome can be appropriately diagnosed and treated. This process includes ruling out inflammatory disorders, such as infectious arthritis and Langerhans cell histiocytosis,[1] as SAPHO syndrome is commonly confused with them and also ruling out other etiologies associated with osteitis, such as Ewing sarcoma and osteoblastoma.[4] Patients who both complain of musculoskeletal pain and have a presenting rash should be considered as having SAPHO syndrome. Typically, diagnosis is confirmed via bone scan with the pathognomonic "bull's head" sign, which shows an increased bilateral sternoclavicular bone uptake and represents ossification.[5]

Case Presentation

A post-menopausal woman with a known medical history of SAPHO syndrome presented to the emergency department (ED) with concerns of left foot and chest pain that began 3 days prior to admission. The patient reported her foot pain was similar to pain she had in the past and that it became unbearable the day before she presented to the ED. She also reported that her chest pain was aggravated by palpation. Physical exam revealed chronic pustular rashes, located on the bilateral soles of the feet, bilateral palms, neck, and abdomen, which she stated appeared hours after the initial presentation of her pain (**Figure 1**). The patient stated the rashes had worsened over the past 3 days. A physical exam also revealed a healing, eczematous rash on her right shin, which she said manifested after her second dose of monoclonal antibody medication for management of arthritis, with no prior history of eczema before this episode. Initial vitals and cardiac tests taken in the ED were unremarkable. An X-ray of the left foot and left ankle showed no bony abnormalities. Chest X-ray readings showed osteitis and sclerosis of the sternocostoclavicular joint and first rib, a radiological finding of SAPHO syndrome[5] (**Figure 2**). The patient was given nonsteroidal anti-inflammatory drugs (NSAIDs) for pain relief and IV corticosteroids for the rheumatoid-like inflammatory aspect of SAPHO, as is standard with SAPHO flare-ups. There was immediate improvement in her pain symptoms, and the skin symptoms gradually improved throughout her hospital stay. The patient was discharged home 2 days later with a referral to her rheumatologist.

Discussion

This patient's symptoms were most indicative of a SAPHO syndrome exacerbation. As the patient had already been diagnosed with it in the past, management and treatment of her symptoms were straightforward. One must look at clinically pathognomonic signs, such as the patient's rash, and foot and chest pain, in addition to the dermatological symptoms appearing simultaneously. It is also important to rule out other differential diagnoses when diagnosing SAPHO for the first time. For example, since the patient complained of chest pain, cardiac

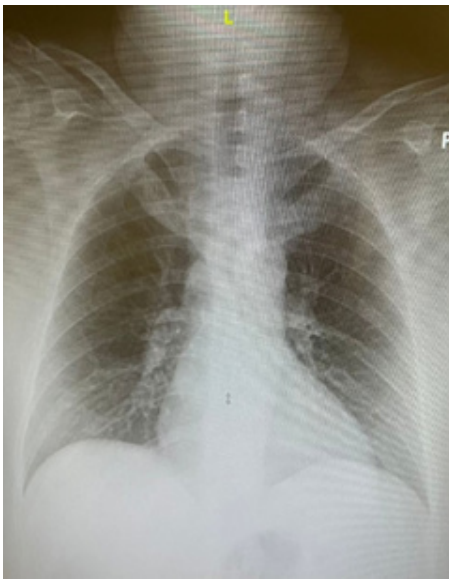

Figure 2. A chest X-ray shows a "bull's head," which is pathognomic radiologically on bone scintigraphy. However, there is noticeable thickening of the sternoclavicular joint and the first rib, representing osteitis, a radiological finding of synovitis, acne, pustulosis, hyperostosis, and osteitis syndrome.

etiologies had to be ruled out. The patient's troponin levels were less than 0.012 ng/mL, and all cardiac tests done were unremarkable and showed no evidence of myocardial injury. While not performed during her hospital stay, a bone scan can help rule in SAPHO syndrome as patients will frequently have increased sternoclavicular uptake. The results should show a "bull's head" sign on imaging, which is pathognomonic for the disease.[5]

The patient's foot pain was also considered a symptom of SAPHO syndrome exacerbation, as bone pain can result from the osteitis and synovitis seen in the disease.[6] This diagnosis was primarily supported by the fact the patient stated this pain was similar to the pain she had in prior SAPHO syndrome exacerbations, and she denied recent trauma to the area. However, other etiologies of her bone pain, such as osteomyelitis and bone malignancy, had to be considered and ruled out. The patient's white blood cell count was found to be normal, and the patient was afebrile throughout her entire stay. Therefore, a diagnosis of osteomyelitis became unlikely, and no blood cultures were taken during her hospital visit. The patient had no history of cancer and had negative findings of bony abnormalities on X-rays of her left foot and left ankle, making metastatic bone disease an unlikely diagnosis. Deep vein thrombosis (DVT) was also considered but ruled out due to the lack of DVT findings on the lower extremity bilateral duplex ultrasound.

The patient's diffuse pustular rashes of the extremities, neck, and abdomen were also considered to be due to a SAPHO syndrome exacerbation. The appearance of the patient's rashes was most consistent with that of palmoplantar pustulosis, which is the most common dermatological presentation of SAPHO syndrome. Palmoplantar pustulosis is a pustular rash in which the patient develops sterile pustules, most commonly of the palms and soles.[7]

Conclusion

While SAPHO syndrome is a rare disease, it is important to be aware of its manifestations and symptoms, as an early diagnosis can provide patients with accurate and appropriate treatment. A diagnosis can typically be made via bone scan, which demonstrates increased uptake at sites of involvement.[6] A chest X-ray, while not as specific, can also be used to support a diagnosis of SAPHO syndrome with corresponding clinical symptoms by looking for the sclerosis of the medial clavicles and first ribs.[8] Blood tests, such as a positive HLA-B27, can be seen in 30% of patients.[6] Generally speaking, it is the combination and manifestation of the majority of the 5 core symptoms of SAPHO syndrome that lead to a diagnosis.

Treatment of the disease is primarily focused on providing symptomatic relief and modifying the inflammatory process through the use of NSAIDs, corticosteroids, antimetabolites, and other anti-inflammatory medications. Antimetabolites have been used with success in previous treatments of SAPHO syndrome, with most of the treatment effects revolving around joint pain relief with mild improvement of skin conditions.[9] Dermatological manifestations are treated with acne medications, from topical antibiotics to vitamin A derivatives.[6]

Conflicts of Interest

The authors declare they have no conflicts of interest.

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