SAPHO SYNDROME: MISDIAGNOSED AND OPERATED

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Abstract

SAPHO is a rare disorder that results in synovitis, acne, pustulosis, hyperostosis and osteitis. Patients with this syndrome typically present with musculoskeletal complaints, frequently localized to the anterior chest wall. However, diagnosis can be difficult in case of involvement of only one symptomatic bone without skin lesions. Awareness of SAPHO syndrome is necessary for accurate diagnosis and to prevent inappropriate and unnecessary treatment.

Keywords: SAPHO Syndrome; Mediastinal Fibrosis; Sweet’s Syndrome.

Case report

A 53 year old female patient presented to our outpatient clinic with complaints of pain and stiffness of the neck and shoulders and low back pain. On physical examination the patient’s Schober’s test was 2.5 cm (normal ≥ 5.0 cm), chest expansion was 1.5 cm (normal >2.5 cm) and the range of motion of the neck was severely limited. Bordered erythematous desquamated plaques psoriatic-like skin lesions were seen in the extensor regions of the elbows and on the anterior chest wall (Figure 1). The histopathological examination revealed perivascular and interstitial dermatitis, edema and marked leukocytoclasis, compatible with Sweet’s syndrome (Figure 2). Laboratory examination revealed the following: white blood cell count 11.600/mm², C-reactive protein 37mg/L (normal: <5mg/L) and...
Erythrocyte sedimentation rate (ESR) 80 mm/1 hr. She also had thrombocytosis (562,000/mm$^3$). Rheumatoid factor and HLA B27 were negative. Cervical spine radiograph showed a bridging from C4-C7 (Figure 3). Magnetic resonance imaging studies of the cervical region revealed ankylosis of the facet joints.

In her medical history she reported an admission to the Cardiology Center with a complaint of chest pain in 1993. Echocardiography revealed cardiomegaly and pericardial effusion which was consistent with the diagnosis of pericarditis. She received antibiotic treatment for a month. However, her chest pain continued despite resolution of echocardiography findings. Six months later, she was referred to the Chest Diseases and Surgery Hospital due to high fever, elevated ESR and persistent anterior chest pain. No skin lesion was present at that time. Bone scintigraphy showed increased osteoblastic activity in sternoclavicular and costosternal joints (Figure 4). Sternoclavicular hyperostosis in the presence of high fever and elevated sedimentation rate rendered a diagnosis of malignancy and the lesion in question was surgically excised. Nevertheless, pathological examination of the biopsy material revealed mediastinal fibrosis (Figure 5). In 1995, neck and low back pain were added to the clinical picture. During the following four years, the pain gradually increased, becoming a real problem for her.

Based on the patient’s medical history and the result of the physical and radiographic examinations, diagnosis of SAPHO was made. The patient received physical therapy for cervical and low back pain, NSAID (indometacin followed by naproxen) and Sulfasalazine. Patient’s sternal and low back pain resolved but she continued to experience mild neck pain and stiffness.

**Discussion**

SAPHO is a seronegative syndrome of bone and joints which is associated with variable dermatolo-
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It is observed mainly in young and middle-aged adults. However, sternoclavicular hyperostosis typically affects patients in the fourth to sixth decades of life. Anterior chest wall involvement is the most characteristic feature of the disease, but other sites in the axial and appendicular skeleton may also be involved. In most cases, spine lesions involve several adjacent vertebrae, evolving with the years towards a vertebral fusion. Sacroiliitis can also occur and unilateral involvement is frequently seen. Radiological findings include hyperostosis and osteitis as manifestations of a chronic inflammatory reaction, resulting in bone sclerosis or sclerotic lesions with areas of osteolysis within it. Bone scintigraphy demonstrates greatly increased focal activity in affected areas, including clinically apparent regions. The diagnosis of SAPHO syndrome is based on the presence of one of the criteria suggested by Benhamou et al, which include: (a) multifocal osteitis without skin manifestations, (b) sterile joint inflammation associated with skin disease, (c) sterile osteitis in the presence of skin manifestations and (d) a disease resembling chronic recurrent multifocal osteomyelitic and exclusion of infectious osteomyelitic or arthritis, primary or metastatic tumors and Paget’s disease.

Skin involvement is variable and includes pustulosis, acne conglobata or fulminans, hidradenitis suppurativa, pustular psoriasis and Sweet’s syndrome, all of these being pseudo abscesses with neutrophilic involvement on the skin. Pustulosis is a chronic eruption of yellowish, intradermal, sterile pustule involving the palms and soles with pustular eruptions. This lesion may be misdiagnosed as an infection, so the patient may receive unnecessary antibiotic therapy. Although skin lesions have been reported in 55% of patients with SAPHO syndrome, osteoarticular abnormalities can occur without skin lesions. Davies et al reported a 20-year interval between the cutaneous and bone involvement. In spite of laboratory findings being usually within normal ranges, erythrocyte sedimentation rate can be high and the patient may experience increased body temperature.

Our patient presented with a primary complaint of pain in the anterior chest wall. Although this is a typical symptom in adult patients with SAPHO syndrome, the absence of accompanying skin lesions or other symptoms pertaining to musculoskeletal system led to a misdiagnosis and consequent mistreatment, including invasive procedures with potential complications. High fever and elevated erythrocyte sedimentation rate complicated the clinical picture. The absence of manifest skin lesions and the lack of information about SAPHO syndrome in those years limited diagnostic elaboration.

Figure 4. Bone scan showing increased uptake in the sternocostoclavicular regions

Figure 5. Dense fibrosis and chronic inflammation in adipose tissue (Stain, hematoxylin and eosin; original magnification, 40 x)
and the patient underwent an unnecessary surgical procedure. The diagnosis of SAPHO syndrome could be established 10 years after the surgery, when her skin lesions and osteitis were evaluated according to diagnostic criteria of Benhamou et al.

Patients presenting with only one specific symptom, like chest pain, should be thoroughly evaluated and SAPHO syndrome should be included in the differential diagnosis, taking into account the natural course of the disease and gradual appearance of symptoms of other systems in the time course, in this case skin lesions. Thus, careful evaluation and considering SAPHO syndrome in the differential diagnosis may prevent many unnecessary examinations and invasive procedures.

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