Case Report: SAPHO syndrome previously mistaken for Paget’s disease

Abstract

SAPHO syndrome is a rare clinical entity, an acronym named for its frequent coincident clinical manifestations, synovitis, acne, pustulosis, hyperostosis, and osteitis. Its clinical phenotype is widely heterogeneous, which coupled with its rarity causes many misdiagnoses. Herein, we present a case of a patient that was previously diagnosed with Paget’s disease. After an MVA, the full extent of his disease was detected via incidental imaging, which demonstrated sclerosis and ankylosis of the lumbar spine, sclerosis of the clavicles, and the pathognomonic bull’s head configuration on bone scan. Upon further questioning, the patient also harbored intermittent palmar pustulosis, psoriasis, sternoclavicular pain and tenderness, and widespread morning joint stiffness that ameliorates with activity. A high index of suspicion and knowledge of its variable manifestations are paramount to arrive at a SAPHO diagnosis and thereby provide appropriate therapy to the patient.

Keywords

sapho, spondyloarthritis, bone scan, paget's disease

Introduction

SAPHO syndrome is an acronym for the frequently coincident skin and osteoarticular manifestations that occur in this malady, namely synovitis, acne, pustulosis, hyperostosis, and osteitis (1, 2). SAPHO is a rare disease, with an estimated prevalence of roughly 1/10,000 (1, 3). Patients between 30-50 years old are most frequently afflicted, but it may occur at any age and is characterized by repeated remission and recurrence (1, 2, 4). The clinical presentation is heterogeneous, with differing degrees of osteoarticular and cutaneous involvement; its most common osteoarticular manifestation is anterior chest wall involvement, and the most common dermatological is palmoplantar pustulosis; however, a large variety of osteoarticular and cutaneous lesions may be seen (1, 2, 4, 5). Given its heterogeneity and rarity, it is often misdiagnosed. Herein, we present a patient that was previously diagnosed with Paget’s disease, who upon incidental imaging following a motor vehicle accident (MVA) was worked up and diagnosed with SAPHO syndrome.

Case Report

The patient is a 37-year-old male who was transferred to the emergency department following an MVA in which he lost consciousness. At the outside facility, CT scans demonstrated a C5 transverse process fracture and multiple lytic/blastic lesions in his lumbar spine concerning for infectious vs metastatic etiology. On admission, the patient complained of neck and diffuse right-sided pain, with superficial abrasions present across his extremities. His PMH was significant for Paget’s disease of the bone diagnosed 3 years prior following a right hip fracture and the presence of early-onset, advanced osteoarthritis. His lab values exhibited considerably elevated
ESR and CRP and a mildly decreased hemoglobin. CXR displayed bilateral clavicular enlargement and sclerosis. Neurosurgery was consulted for the spinal lesions, and MRI scans were ordered.

Cervical MRI revealed small disc osteophyte complexes at C5-C6. Thoracic MRI demonstrated hyperintense signal changes within multiple vertebral bodies (T1-T2 and T4-T6). No bony metastatic disease was noted. Lumbar MRI showed unusual appearing sclerosis and ankylosis of L4-L5 and L5-S1, plus unilateral left-sided sacroiliitis, suggesting a spondyloarthropathy with the involvement of the hips, spine, SI joints, and sternoclavicular joints, with special suspicion for SAPHO syndrome. Subsequently, a bone scan was ordered to evaluate for extent of his disease process and confirm the diagnosis of SAPHO syndrome. The bone scan revealed unusual uptake centered on the medial clavicles, sternal manubrium, and proximal sternal body in a bull’s head configuration, which is pathognomonic for SAPHO syndrome (Fig. 1). There was aberrant uptake noted in several vertebral bodies (from mid-thoracic to lumbar), along with bilateral sacroiliitis (left greater than right), bilateral hips (right greater than left), and medial compartments of the knee and mid feet.

Upon further questioning, the patient states he has intermittent pain, tenderness, and swelling across his sternoclavicular joints and hip and lower-back morning stiffness that improves with activity; this has occurred throughout his lifetime. He also had intermittent palmar pustulosis and ongoing palmar psoriasis. Previous x-rays showed severe bilateral hip arthritic changes.
Fig 1: Zoomed view from frontal projection Te99m HDP whole-body bone scan concentrating on the upper chest showing the “bull’s head” configuration of hyperostosis and osteitis in clavicles and sternum; the clavicles (long arrow) represent the horns of a longhorn steer, while the sternum (short arrow) represents the face and snout.
Fig 2: Frontal radiograph of the pelvis shows several imaging manifestations from which the syndrome gets its name. Abnormally increased density of the L4 vertebral body (long arrow) represents osteitis. Abnormal density and erosions surrounding the left sacroiliac joint (short arrow) represent the sequela of long-standing synovitis. Notice also the advanced arthritis of both hips, irregularity of the pubic symphysis, and sclerosis of the pubic bones. Incidental findings include hardware from prior operative fixation for a right femoral neck fracture and excreted contrast in the urinary bladder from recent IV administration for a CT scan.

Discussion

SAPHO syndrome is a rare entity with variable clinical presentations, making a diagnosis challenging. The most frequent osteoarticular manifestations are anterior chest wall involvement in 65%-90% of patients, with a pathognomonic bull’s head change noted on the bone scan. The SI joint and spine are affected in 32%-52% of patients, and unilateral SI joint involvement
predominates. Peripheral joints are involved in ≤ 30% of patients, with hips, knees, and ankles commonly affected (1, 2, 4, 6). Sternoclavicular involvement is the first symptom in upwards of 70% of patients (7, 8). Arthritis has been reported in 92.5% of cases, affecting the axial skeleton in 91% of those cases, and peripheral joints in 36% (1). Cutaneous manifestations include palmpoplantar pustulosis in up to 60% of patients and nodulocystic acne in 25%, along with psoriasis, hidradenitis suppurativa, and less commonly any variety of neutrophilic dermatoses (9). These patients may develop uveitis or scleritis, harbor low-grade anemia, or have concurrent inflammatory bowel disease in upwards of 10% (1, 4, 9). Importantly, skeletal symptoms and skin disease do not always co-occur, but the typical time interval between the skin and osteoarticular involvement is < 2 years (10).

Radiological features are key for diagnosis, with various findings indicative of the ongoing hyperostosis and osteitis. This includes sclerosis of bone in both processes, generalized cortical, periosteal, and endosteal thickening, narrowing of the medullary canal in hyperostosis, and osteosclerosis involving trabecular infrastructure in osteitis (11). Axial lesions include vertebral body corner lesions, spondylodiscitis, osteodestructive or osteosclerotic lesions, paravertebral ossification, and sacroiliitis (2, 11). Most patients do not have intervertebral disc enhancement on MRI (5). MRI scans are useful for a detailed evaluation of individual lesions, and bone scans can reveal the extent of disease, plus identify asymptomatic sites of active disease (11).

The patient’s characteristics are congruent with prior reports. He had essentially life-long morning joint stiffness that improved with activity, pain and tenderness across his sternoclavicular joints, bilateral hip arthritis, psoriasis, and palmar pustulosis. Radiologic findings corroborated a SAPHO diagnosis, with sclerosis and ankylosis demonstrated in his lumbar spine, sclerosis of his clavicle, and the pathognomonic bull’s head configuration on the bone scan.

Herein, we present a case of a patient with SAPHO syndrome that was previously misdiagnosed with Paget’s disease. A high index of suspicion, along with knowledge of the disease and its heterogeneous manifestations, is key for an accurate diagnosis. Crucially, SAPHO may be a disease subset within the family of spondyloarthropathies, with its own designated subtypes (6). The genetics of SAPHO closely align with other spondyloarthropathies, and the genetics and pathogenesis are entirely distinct from Paget’s disease (12-15). Importantly, a correct diagnosis can allow for appropriate therapy, as Paget’s disease is treated almost solely with bisphosphonates. However, a wide variety of agents are utilized for SAPHO, including NSAIDs, antibiotics, TNF-α inhibitors, methotrexate, sulfasalazine colchicine, glucocorticoids, and bisphosphonates (1). With treatment, SAPHO follows a chronic, relapsing-remitting course with a typically good prognosis, although rarely a disabling course can occur, alongside the potential for late-stage amyloidosis (16, 17). Differing disease manifestations respond differently to distinct treatments, making the role of greater treatment options and idiosyncratic patient drug tailoring a necessity.
References