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J Rheumatol 2015;42;2514
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To the Editor:

I have read with great interest the article by Aljuhani, et al1, regarding a large monocentric case series of patients with synovitis, acne, pustulosis, hyperostosis, and osteitis (SAPHO) syndrome. I fully agree with Aljuhani, et al on the frequent involvement of soft tissues around inflamed bone, with the consequent diagnostic difficulties. SAPHO syndrome, in fact, could be a real diagnostic challenge, and Aljuhani, et al described a significant diagnostic delay; this is important because the diagnostic delay leads to inappropriate investigations and prescriptions. With regard to diagnostic delay, it is worth recalling that skin lesions may not be accounted for when they are episodic or appear after bone lesions2. In 1987, Chamot, et al3 suggested the acronym SAPHO to unify various conditions characterized mainly by osteitis/hyperostotic lesions of the anterior chest wall (ACW).

Therefore, SAPHO syndrome could be considered a “useful concept” that unifies several disorders sharing some clinical, radiological, and pathologic characteristics, especially osteitic/hyperostotic lesions of the ACW. This means that the understanding of SAPHO syndrome is also very important in musculoskeletal radiology, because such awareness could facilitate the differentiation from other diseases that produce similar radiographic findings but have different prognoses and treatments, such as osteomyelitis, Ewing sarcoma, metastasis, and Paget disease of bone4.

In a case series described in Arthritis and Rheumatism5, my colleagues and I found that osteitis has sometimes been associated with soft tissue involvement. I also agree with the authors that when inflammation of the surrounding soft tissue of osteitic bone is detected, a biopsy should be mandatory to exclude malignancy or infections. It is also worth recalling that SAPHO syndrome is probably a primitive reactive osteitis in genetically predisposed subjects and that Propionibacterium acnes, Staphylococcus aureus, and other germs have been isolated from osteoarticular lesions in the ACW, spine, pustules, and synovial fluid tissue6. In 2007, Trotta and I7 that SAPHO syndrome is lost after its discontinuation8. As far as the elusive nosological framing of SAPHO syndrome, my colleagues and I first described a dysregulation of extracellular ATP-dependent P2X7-IL-1β axis in a case of SAPHO syndrome effectively treated with the interleukin 1 receptor antagonist anakinra, suggesting that SAPHO syndrome could be considered a polygenic autoinflammatory disease9.

In another study, we described 5 first-degree relatives of patients with SAPHO syndrome2, and then we looked for single-nucleotide polymorphisms (SNP) in 3 genes (PSTPIP2, LPIN2, and NOD2), previously analyzed in a French cohort10, as well as in 2 genes (PSTPIP1 and PTPN22), which we looked at for the first time. As in the French cohort, no SNP were found in association with SAPHO syndrome in our cohort of 53 Italian patients11.

According to Aljuhani, et al, diagnosis of SAPHO syndrome could be challenging and the diagnostic delays are relevant. Moreover, it is very important to be cautious in those cases with involvement of soft tissues because in such cases it is necessary to exclude malignancy. Besides, in those cases the biopsy may also be useful for directing the treatment in case of isolation of pathogens.

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