RHEUMATOLOGY

Letter to the Editor (Case report)

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A 72-year-old man with life-threatening dyspnoea due to a neck mass of unexpected origin

Rheumatology key message

- In cases with rapid swelling of the sternoclavicular joints, SAPHO syndrome should be considered.

Dear Editor, SAPHO syndrome is a rare disorder, which consists of synovitis, acne, pustulosis, hyperostosis and osteitis [1]. Immunological, genetic and infectious factors underlie SAPHO aetiology, but the exact pathophysiological mechanisms remain elusive [1]. The innate immune system is likely to be involved in SAPHO syndrome via elevation of pro-inflammatory cytokines, including IL-8, IL-17 and TNF-α [1]. The bacterial species *Propionibacterium acnes* has been identified in previous studies on SAPHO syndrome [2]. Genetic factors are proposed to be involved in SAPHO syndrome, but no specific genes have been identified. SAPHO syndrome presentation can be heterogeneous, and diagnosis is based on the SAPHO diagnostic criteria [3]. Mainly the anterior chest wall joints are affected bilaterally, including sternoclavicular, costoclavicular and manubriosternal joints, though other sites throughout the body can be affected [1]. The bullhead sign has been reported in SAPHO bone scintigraphy studies, with little value for diagnosing this syndrome [4].

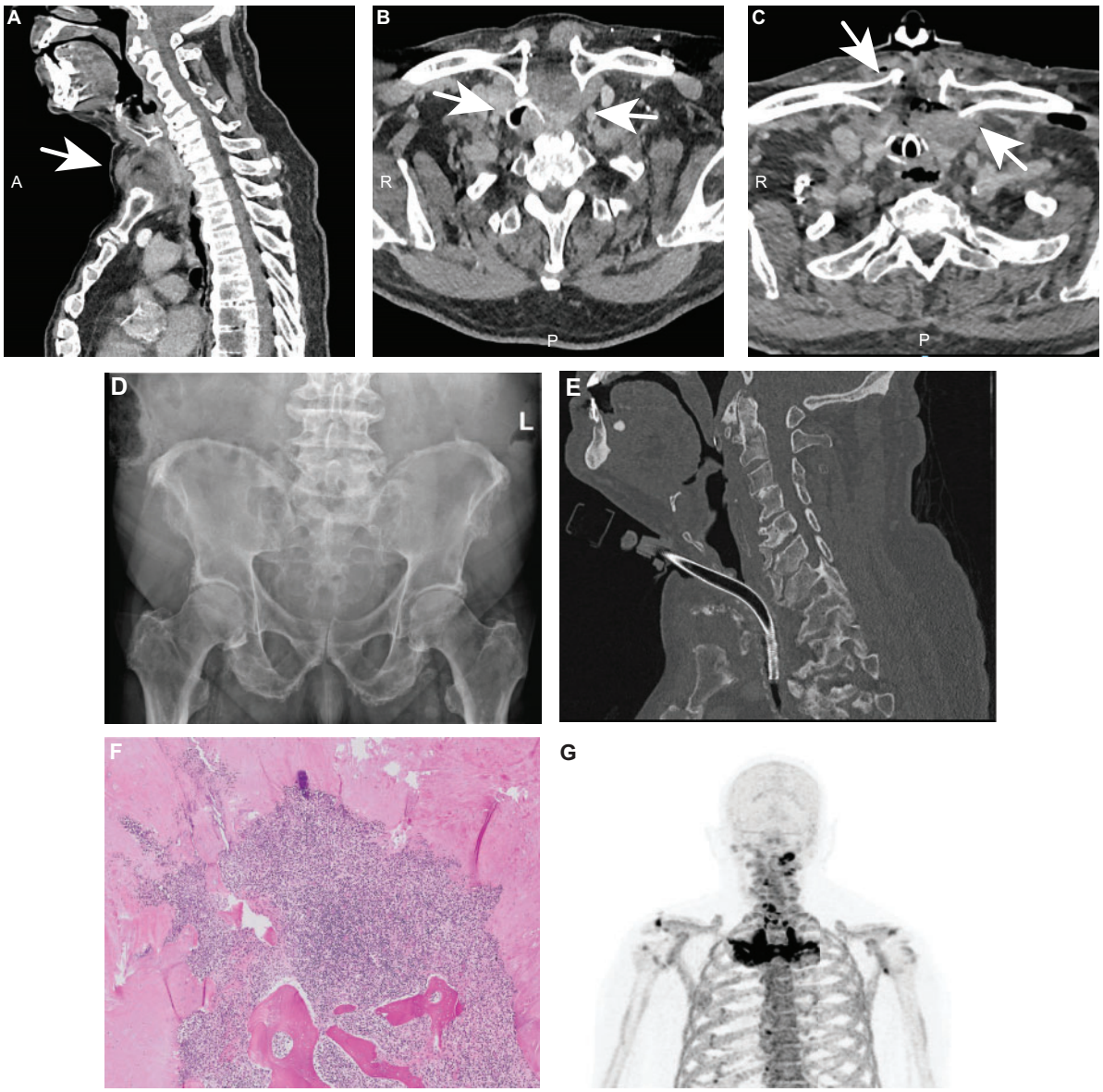
Treatment of pain using NSAIDs or IA or systemic administration of CS is the main focus in SAPHO syndrome management [1]. Furthermore, bisphosphonates can result in complete remission [1]. Several studies have shown that TNF-α blockade has a positive effect on osteoarticular symptoms in SAPHO syndrome; however, these drugs are less effective in patients with cutaneous symptoms [1].

We present a case of SAPHO syndrome with a complex diagnostic workup with acute life-threatening dyspnoea caused by a mass effect on the trachea. A 72-year-old man presented himself in the otorhinolaryngological outpatient clinic with progressive dysphagia and minimal dyspnoea, without weight loss or change of voice. No tumours or suspected lymph nodes were identified in the neck region. Flexible laryngoscopy showed a tissue mass located in the hypopharynx. A histological biopsy via laryngoscopy under general anaesthesia was necessary.

Two weeks later, on a national holiday and before the planned procedure, the patient presented himself at the emergency department because of severe dyspnoea and stridor. Swallowing had become more difficult. Laboratory testing showed leucocytosis and elevated CRP levels (53 mg/l). A contrast-enhanced CT scan of the neck and chest revealed a large mass in the neck with a luminal stenosis of the trachea starting 2.5 cm caudally from the larynx leaving a tracheal lumen of 3 mm× 14 mm (Fig. 1A and B). Based on the CT scan and progression of the tumour, the patient was informed about the highly suspected diagnosis of an anaplastic thyroid carcinoma, which has a 99% mortality rate [5]. The next day, the patient was discussed among the team of head and neck surgeons. A dedicated head and neck radiologist evaluated the original CT scan again, and found a collection of fluid leading to the sternoclavicular joints, which were swollen and showed local signs of arthritis and arthrosis. These signs made an inflammatory or infectious aetiology more likely. This caused narrowing of the space of the thoracic aperture, displacement of the thyroid gland in the direction of the hypopharynx and narrowing of the tracheal lumen. Multiple surgical procedures, including a tracheotomy, partial resection of the sternoclavicular joints and retrosternal osteophytes were necessary to obtain sufficient improvement of dyspnoea (Fig. 1C). Conventional X-rays showed degenerative changes, including osteophytes on different locations such as pelvis and cervical spine (Fig. 1D and E). Moreover, the pelvic X-ray showed osteitis around the pubic bone and the tendon insertions (Fig. 1D).

Histopathological analysis showed a partially active inflammation of costoclavicular bone and cartilage (Fig. 1F). Microbiological analysis of the biopsy found a variety of bacterial species, including *P. acnes*. Additional laboratory testing showed persistent elevated levels of leucocytes and CRP but negative HLA-B27. A sodium-fluoride PET scan showed the bullhead sign (Fig. 1G). Additionally, bisphosphonate therapy with 9 mg pamidronate was administered one time i.v. together with infliximab (i.v. 5 mg/kg) every 2 weeks for 8 weeks and prednisone 60 mg daily with a reduction of 10 mg/week. Based on the clinical symptoms, imaging, localization and results from the histopathological studies, it was concluded the patient was suffering from SAPHO syndrome. In addition to the major sternoclavicular findings, the very extensive enthesopathy, present in almost all joints, was the main symptom that led to the diagnosis of SAPHO syndrome in line with cases described earlier [6]. Other SAPHO criteria, namely extra-sternal articular manifestations, acne and pustulosis, were not present at time of diagnosis. After 4 months, the patient was discharged from

Fig. 1 CT imaging and histopathological studies



(**A**) Sagittal CT scan taken during presentation at the emergency department; arrow indicates the mass causing the tracheal deviation and stenosis. (**B**) Axial CT scan taken during presentation at the emergency department; arrows indicate the mass causing the tracheal deviation and stenosis. (**C**) Axial CT scan following second debulking and removal of osteophytes (accentuated via white arrows). (**D**) Pelvic X-ray reveals osteophytes on the pelvic bone and signs of osteitis along the pelvic contours at location of the tendon insertions. Moreover, there are signs of sclerosis and unclear margins of the pubic joint. (**E**) X-ray image of the cervical spine (2 weeks after tracheotomy) shows osteophytes at multiple locations. (**F**) Image shows a partially active inflammation of costoclavicular bone and cartilage. (**G**) Sodium-fluoride PET image reveals increased uptake around the sternoclavicular joints (bullhead sign). Written informed consent was obtained from the patient to publish these images. A: anterior; R: right, P: posterior; L: left.

the hospital with a closed tracheotomy and without dyspnoea.

This case of life-threatening dyspnoea showed that an adequate diagnosis can be challenging on holidays, and that during the multidisciplinary diagnostic work-up, malignancies, and infectious and inflammatory aetiologies should be included in the differential diagnosis. In cases with rapid swelling of the sternoclavicular joints, especially in cases with dermal symptoms, SAPHO syndrome should be considered.

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Data availability statement

Data are available upon reasonable request by any qualified researchers who engage in rigorous, independent scientific research, and will be provided following review and approval of a research proposal and Statistical Analysis Plan (SAP) and execution of a Data Sharing Agreement (DSA). All data relevant to the study are included in the article.

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