Case report

Ankylosing Spondylitis in a 33- years old man with Hypermobility Joints

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Abstract

Background: Ankylosing spondylitis, is a chronic inflammatory autoimmune disease that mainly affects spine joints. It can cause severe, chronic pain, spine fusion and chronic disability. Ehlers-Danlos syndrome is a rare heterogenous group of hereditary connective tissue disorders which are characterized by skin hyperextensibility, joint hypermobility and tissue fragility.

Case report: A 33-year old man was admitted to the Rheumatology Department of 251 Airforce General Hospital in order to decide on his capability to serve his duty as an aircraftsman at Hellenic Airforce. He was diagnosed with ankylosing spondylitis twelve years ago. On physical examination joint hypermobility and skin hyperextensibility were observed and after further investigation, he was diagnosed with hypermobility type of Ehlers Danlos Syndrome. Laboratory investigation was normal. MRI radiologic investigation revealed bony sclerosis of the left sacroiliac joint and bone marrow oedema on right sacroiliac joint. No extraskeletal manifestations were detected. The patient was informed about both conditions and he was given medical advice about their proper management.

Conclusion: Rare diseases sometimes manifest as atypical inflammatory arthritis syndromes. Careful medical history, thorough physical examination and use of diagnostic criteria are useful in such patients for disease identification and treatment.

Key words: Ankylosing Spondylitis, Ehlers Danlos Syndrome, Joint Hypermobility

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Abbreviations

SpA: Spondyloarthrtitis

AS: ankylosing spondylitis

PsA: psoriatic arthritis

ReA: reactive arthritis

NSAIDs: non-steroidal anti-inflammatory drugs

bDMARDs: biological disease-modifying antirheumatic drugs

EDS: Ehlers-Danlos syndrome

cEDS: classic EDS

cvEDS: cardiac – valvular EDS

hEDS: hypermobile EDS

vEDS: vascular EDS

kEDS: kyphoscoliotic EDS

aEDS: arthrochalasia EDS

dEDS: dermatosparaxis EDS

BCS: brittle cornea syndrome

spEDS: spondylodysplastic EDs

mcEDS: musculocontractural EDS

mEDS: myopathic EDS

pEDS: periodontal EDS

JHS: joint hypermobility syndrome

Introduction

Spondyloarthrtitis (SpA) is a heterogenous group of chronic inflammatory arthropathies. They affect not only the spine but also certain extra articular sites (Zhu et al, 2019) . The SpA group includes axial SpA - including ankylosing spondylitis (AS), peripheral SpA, psoriatic arthritis (PsA), enteropathic arthritis, reactive arthritis (ReA) and juvenile SpA (Carli et al, 2019). AS usually begins in late adolescence or early adulthood. Onset after the age of 45 is uncommon. It affects mostly males (F:M ratio 2-3:1). The typical AS patient presents with low back pain with prolonged morning and sometimes nocturnal stiffness which improves only with exercise. Another feature of AS is persistent buttock pain which may alternate from side to side at the early stages of the disease (Watad et al, 2018). Physical examination reveals decreased spinal mobility, sacroiliac joint tenderness, and sometimes enthesitis and reduced chest expansion. Radiographic sacroilitis is the hallmark of AS and there is a strong association with HLA-B27. Exercise and NSAIDs can control the inflammatory symptoms. Patients with high disease activity and extra articular manifestations may benefit from bDMARDs such as anti-TNF and anti- IL17 agents (Taurog et al, 2016).

Ehlers-Danlos syndrome (EDS) is a rare heterogenous group of hereditary connective tissue disorders which are characterized by skin hyperextensibility, joint hypermobility and tissue fragility. Diagnosis is based upon a series of major and minor criteria according to EDS type and family history. There are thirteen types of EDS (Jesudas et al, 2019):

1. Classic (cEDS)
2. Classical-like EDS
3. Cardiac-Valvular (cvEDS)
4. Hypermobile (hEDS)
5. Vascular (vEDS)
6. Kyphoscoliotic (kEDS)
7. Arthrochalasia (aEDS)
8. Dermatosparaxis (dEDS)
9. Brittle cornea syndrome (BCS)
10. Spondylodysplastic (spEDS)
11. Musculocontractural (mcEDS)
12. Myopathic (mEDS)
13. Periodontal (pEDS)

Hypermobility EDS (Tinkle et al, 2019) includes large and small joint as well as spine hypermobility, frequent joint dislocations, scoliosis and premature osteoarthritis. Skin's hyperextensibility is not as severe as in classic EDS. Wound healing is normal and mitral valve prolapse is uncommon. It is characterized by genetic heterogeneity. In most patients inheritance appears to be autosomal dominant but there is a rare subtype of this EDS type due to tenascin X deficiency. As a result, genetic testing for most patients is not available.

Differential diagnosis includes other types of EDS and other connective tissue disorders, including Marfan syndrome.

Case report

A 33-year old man was admitted to the Rheumatology Department of 251 Airforce General Hospital in order to decide on his capability to serve his duty as an aircraftsman at Hellenic Airforce. His medical history was remarkable for inflammatory back pain with prolonged morning stiffness and radiographic sacroiliitis starting 12 years ago. He was also positive for HLA-B27. As a result, AS was diagnosed. He had not received any medication after he had been diagnosed with AS, except for low doses of aceclofenac on demand.

The patient was clinically reexamined and had a complete laboratory and radiologic examination to confirm the diagnosis of AS, define the disease

activity and detect any extraskeletal manifestations of AS.

The clinical examination revealed limited loss of cervical range of motion, normal chest expansion, Scober test +4cm, unilateral sacroiliac joint pain at Gaenslen's test and Achilles enthesitis. No peripheral joint involvement was detected.

Laboratory investigation was normal. Radiologic investigation revealed bony sclerosis of the left sacroiliac joint in plain radiographies and bone marrow oedema of the right sacroiliac joint in the MRI (Figure 1). No extraskeletal manifestations were detected.

Clinical examination also revealed joint hypermobility (Figure 2), which became subject of further clinical investigation. A Beighton score was performed (7/10).

After the performance of an extent differential diagnosis (including other types of EDS and other connective tissue diseases such as Marfan syndrome), the most probable diagnoses were joint hypermobility syndrome (JHS) and Hypermobility EDS. The connection between them remains uncertain but the majority of experts considers them equal. The patient fulfills both Brighton criteria for JHS (1 major, 2 minor criteria) and 2017 international diagnostic criteria for hEDS. Due to hyperextensibility (>5cm) of the skin the patient probably should be diagnosed as hypermobility EDS (Malfait et al, 2017).

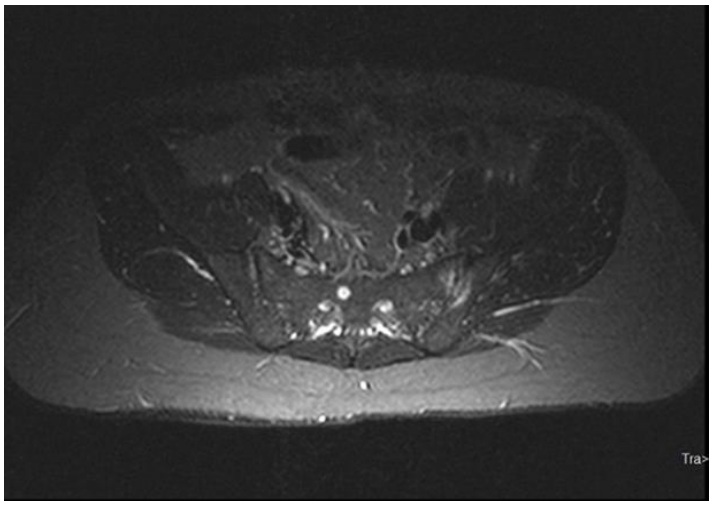

Figure 1: MRI of the sacroiliac joints. Bone marrow oedema of the right sacroiliac joint is illustrated.

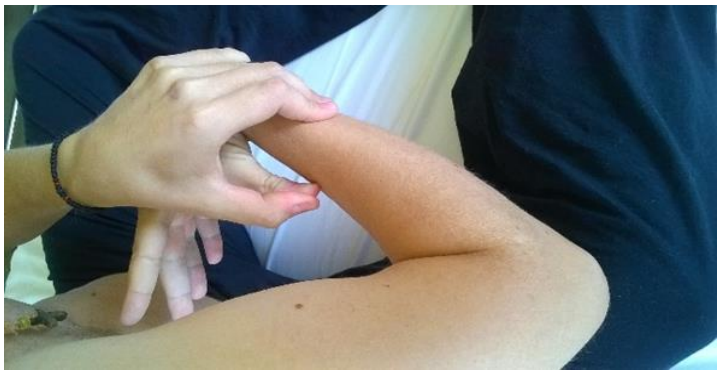

Figure 2: Passive apposition of the thumb to the flexor aspect of the forearm

Discussion

The management strategy for this patient was divided into four steps. The first was to establish the diagnosis of AS, define the disease activity and the existence of extraskeletal manifestations such as anterior uveitis and IgA nephropathy. It was important that the diagnosis of AS was questioned because several manifestations of the disease resemble articular involvement of a joint hypermobility syndrome. There are clinical cases in medical literature which highlight that a syndrome like this can be misdiagnosed as arthritis (Rodgers et al, 2017).

The second phase was to establish the diagnosis of Hypermobility EDS using clinical criteria. The complex differential diagnosis was made between the different types of EDS and other heritable connective tissue diseases such as Marfan syndrome. Careful clinical evaluation, clinical criteria for the diagnosis of these syndromes and a diagnostic flow chart from American Journal of Medical Genetics were the tools that established the diagnosis of Hypermobility EDS (Tinkle et al, 2017).

The third and fourth steps consisted of providing the appropriate treatment for both conditions although they are clinically very different (Shur et al, 2020). Regarding the low disease activity and the fact that the patient did not present extraskeletal manifastetions, treatment with NSAIDS on demand with proper exercise and physical therapy was continued. The patient was advised to follow an exercise and physical therapy program with an expert in order to avoid injury of hypermobile joints and recurrent episodes of joint dislocation. Finally, he was informed about possible medical emergencies, chronic pain, neurophychiatric and other manifestations of hypermobility EDS in order to look for proper medical care (Joseph et al, 2018).

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