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Case Report

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Whose team do you ski for? The simultaneous presence of Klippel-Feil syndrome and diffuse idiopathic hyperostosis mimics clinical presentation of advanced ankylosing spondylitis.

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Abstract

Introduction: *Klippel-Feil syndrome (KFS) is a congenital fusion of two or more cervical vertebrae. Diffuse idiopathic hyperostosis (DISH) is a disorder in which there is calcification of ligaments and tendons, primarily of the spinal column. The simultaneous occurrence of DISH and ankylosing spondylitis (AS) has seldom been described in the literature. Coexistence of KFS and DISH clinically resembling "skier's posture" of the advanced AS, to the best of our knowledge has never been described before in literature.*

Case Report: *59-years old male was examined by rheumatologist due to inflammatory back pain and prolonged morning stiffness in spinal joints. He had "skier's posture" which could be clinical sign of advanced axial disease in AS. Functional measurements had also spoken in favor of spondyloarthropathy. Spinal joints radiography described 2nd and 3rd cervical vertebral fusion along with prominent osteophytes and dorsal spine hyperostosis. Sacroiliac joints had no inflammatory or degenerative radiographic changes. Hip joints ultrasound excluded synovitis and effusion.*

Conclusion: *Patient has two joint diseases, resulting in severe spinal degenerative changes, together resembling clinical presentation of advanced AS.*

Keywords: Ankylosing Spondylitis, Diffuse Idiopathic Hyperostosis, Forestier's Disease, Klippel-Feil Syndrome

Introduction

Ankylosing spondylitis (AS) is a seronegative arthritis affecting sacroiliac joints, spine and peripheral joints. Inflammation may also involve eyes, guts, skin, etc [1]. Despite diagnostic criteria development, diseases of the spondyloarthritis spectrum are still not enough recognizable and there is significant treatment delay [2]. Inflammatory back pain may be treated as pain due to mechanical causes, without thorough diagnostic approach, for many years. Severe spinal involvement, namely "bamboo spine" and "skier's posture" may be first clinical findings in such patients.

Klippel-Feil syndrome (KFS) is a congenital fusion of cervical vertebrae causing neck's limited range of motion and secondary degenerative changes. Since it is genetic disorder it may be associated with different skeletal or extra skeletal anomalies.

Diffuse idiopathic hyperostosis (DISH) is a condition with pathologic bone formation in soft tissues surrounding the spine, but may be extended to the rest of axial and peripheral joints.

Case Report

59-years old male was examined as an outpatient at the rheumatology clinic for his long term night, early morning and at rest back pain associated with prolonged spinal joints stiffness. He had very limited range of motions in his neck and could not tie shoelaces.

He had no comorbidities, surgeries, fractures, accidents involving musculoskeletal system and no drug allergies. He was non-smoker and without of a family history significant for rheumatologic conditions. His inflammatory markers complete blood count, biochemistry and urine sample analysis was normal.

He had "skier's posture", possibly hallmark of advanced axial AS. Functional measurements were as follows: BASDAI 7.8, ASDAS-CRP 4.8, BASFI 9.5, Schober test was less then 2cm. He had limited external and internal rotations and flexion of both hips. Other physical findings were normal.

His cervical spine radiography showed 2nd and 3rd cervical vertebrae in block which is a diagnostic sign of KFS (Figures 1

and 2). He had skolyosis, prominent kyphosis of thoracic spine and lumbar spine lordosis. He had osteophytes and hyperostosis of thoracic spine which is diagnostic for DISH (Figures 3 and 4). His pelvic radiography, including sacroiliac joints study, showed no inflammatory or degenerative changes (Figures 5 and 6). His lung radiography was normal. Hips ultrasound revealed no inflammatory or significant degenerative changes.

Sacroiliac joints MRI and HLA-B27 test were not done due to obvious radiographic changes speaking in favour of two coexisting conditions: KFS of cervical spine and DISH of thoracic spine, different from AS.

Figure 1: CXR neck frontal view.

Figure 2: CXR neck sagittal view.

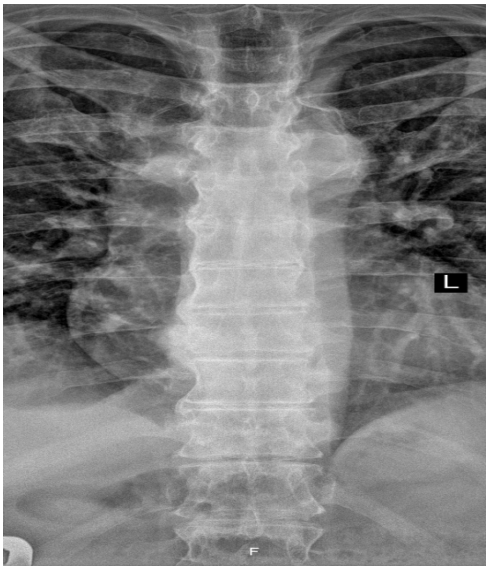

Figure 3: CXR thoracic spine frontal view.

Figure 4: CXR thoracic spine sagittal view.

Figure 5: CXR lumbosacral spine frontal view and sacroiliac joints frontal view.

Figure 6: CXR lumbosacral spine lateral view.

Conclusion

Patient has two musculoskeletal diseases which affect spine: KFS of neck and DISH of thoracal spine. Together they mimic specific patients' posture characteristic for severe axial spondyloarthritis.

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