A Case of Klippel-Feil Syndrome Causing a Mass Appearance in the Nape

Ensede Kitle Görünümü Oluşturan Bir Klippel Feil Sendromu Olgusu

Klippel-Feil Sendromu / Klippel-Feil Syndrome

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Özet

Klippel-Feil Sendromu fetal gelişimin erken haftalarında servikal vertebranın normal segmentasyon ve bölünmesindeki yetersizlikten kaynaklanan, multipl sistem anomalilerinin eşlik ettiği bir sendromdur. Bu yazıda 14 yaşında boyunda şişlik ve kitlenin malign karakterde olma endişesi ile ortopedi kliniğine başvuran olguyu sunduk.

Anahtar Kelimeler

Klippel-Feil Sendromu; Servikal Vertebra; Anomali

Abstract

Klippel-Feil Syndrome is a syndrome arising from insufficiency of the normal segmentation and cleavage of the cervical vertebrae in the early weeks of fetal development, accompanied by multisystem abnormalities. In this paper, we present a 14-year-old subject who referred to the orthopedics clinic concerned about the swelling and the mass in the cervical region possibly being malignant.

Keywords

Klippel-Feil Syndrome; Cervical Vertebrae; Abnormalities

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Introduction

Klippel-Feil Syndrome (KFS) is a rare congenital malformation characterized with the triad of short neck, low hair line and restricted neck movements due to insufficiency of segmentation of two or more cervical vertebrae.[1] In this report, we have presented the clinical and the radiological findings of the patient with KFS, whose leading complaint was a mass in the back of the neck. Although KFS has been well defined in literature, any case has not been observed as we report.

Case Report

A 14-year-old female patient presented to the orthopedics clinic with the complaints of a mass in the nape and restriction in neck movements. The patient stated that she had the complaints for 3 years, and that the mass displayed enlargement during the preceding 6 months and she was concerned that the mass may have had a malignant character. On her physical examination, a firm and fixed mass of approximately 3 cm in diameter was detected, causing no pain, showing protrusion on the left of the midline in the posterior cervical region (Figure 1). The patient

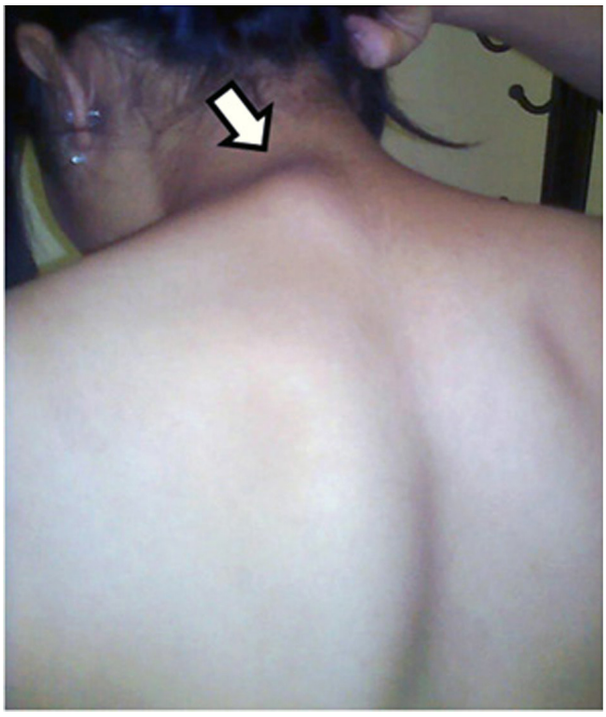


Figure 1. View of protrusion on the left of the midline in the posterior cervical region.

also had a short neck and restricted head and neck movements. No neurological deficits were detected. The AP/lateral X-Rays of the cervical region revealed scoliosis with left convexity, a fusion defect in the spinous process of the C6 vertebrae, bilateral cervical ribs, fusion in the C6-C7 corpus vertebrae, hypertrophy towards the posterior in C5-C6 and C7 vertebrae's spinous processes and synostosis between them. (Figure 2). It was understood that the palpated mass was the synostosed hypertrophic spinous processes of the C5-C6-C7 vertebrae. The anomalies were not detected on the thoracic and lumbar vertabral X-Rays. On computed tomography, a fusion defect (spina bifida occulta) was observed in the C1 and C6 vertebrae. No organ pathologies were found on cerebral tomography and panabdominal ultrasonography. The patient was diagnosed as having KFS as she had scoliosis in the cervical region, fusion in the corpuses of the



Figure 2. Cervical AP/Lateral X-ray images of the case show scoliosis with left convexity, a fusion defect in the spinous process of the C6 vertebrae, bilateral cervical ribs, fusion in the C6-C7 corpus vertebrae, hypertrophy towards the posterior in C5-C6 and C7 vertebrae's spinous processes and synostosis between them.

vertebrae, synostosis in the spinous processes and a short neck. The patient was told that the cause of the swelling in the nape was a mass of benign structure and that a surgical intervention would not be planned unless neurological symptoms develop. Follow-up of the mass was recommended.

Discussion

KFS was first described by Klippel and Feil in 1912. It is seen at a rate of approximately 1 in 42,000. It is more prevalent among girls. KFS arises from insufficiency of the normal segmentation and cleavage of the cervical vertebrae in the early weeks of fetal development.[2] It is a congenital malformation characterized with the triad of short neck, low hair line and restricted neck movements.[1,3] Cases in which the classical triad is observed constitute approximately 50% of KFS cases.[4] In the cases presented here, the finding of low hair line was not observed. Skeletal system abnormalities, urinary system abnormalities, synkinesis, hearing loss, congenital heart disease and brainstem abnormalities may accompany the classical triad.[5]

It is observed that there were also deformities related with skeleton system like scoliosis, spina bifida, costa anomaly, cervical costa, sprengel deformation and fasial asymmetry in patients who have KFS. Thomsen et. al. defined that 70% scoliosis, 26% sprengel deformation and 9% extremity anomaly was seen in the first examination of 57 patients. In our patient we observed that there were cervical scoliosis, spina bifida, bilateral cervical costa and fusion of C5-C6-C7 cervical vertebra spinoz processes.[6]

Konstantinou et. al. determined fusion between C1 and T3 and bilateral cervical costa in the cervical x-ray imaging of a patient who has pain in upper extremity, paresthesia, weakness and color change in the skin. In our case, bilateral cervical costa that constitute costa syndrome was seen.[7] In the research of Mahiroğulları et. al. which was conducted in Turkey, only in one patient, fusion was seen in lumbar spine; whereas it was observed that there were scoliosis, cervical fusion, low hair line and short neck in all of the 23 patients. It was observed that one patient had renal agenesia, nine patients had hearing loss at various levels and five patients had cardiac problem.[4]

Three types of the disease have been described. In type 1, there are synostosis and cervical fusion of the upper thoracic vertebra (40%); in type 2, there is isolated cervical vertebral fusion (47%), and in type 3, there is cervical fusion with lower thoracic or upper lumbar vertebra (13%).[4,5] Our case was consistent with type 2 KFS.

Consequently, KFS has to be taken into consideration for the cases that has system anomaly. Especially it has to be kept in mind that, the patients who have cervical hypermobility can need surgical treatment and the patient should be informed about the syndrome.[6] The fact making the case interesting was the presence of a mass developing as the result of excessive hypertrophy and fusion of the vertebral processes, causing concern by the patient. No cases appearing like a mass as in our case have been encountered in the English literature. The need of treatment depends on the pathologies that may be caused, mostly by deformity and the other accompanying system abnormalities.[8] However, in this case, determination of no malignancy of the mass is more important than its treatment.

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