An isolated case report of Klippel-Feil syndrome: an acute pathology?

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Summary

The aim of this paper was to review clinical heterogeneity; radiographic abnormalities in klippel Feil syndrome which simulate acute pathology. We insist in comprehensive evaluation and delineation of diagnostic and prognostic classes.

Introduction

Klippel-Feil syndrome (KFS) is defined as congenital fusion of two or more cervical vertebrae and is believed to result from faulty segmentation along the embryo's developing axis during weeks 3-8 of gestation. It manifests as a short neck with reduced mobility and a low posterior hairline, occurring only in 40-50% of patients. Decreased range of motion is the most frequent clinical finding. Patients with upper cervical spine involvement tend to present at an earlier age than those whose involvement is lower in the cervical Spine. In addition, a wide spectrum of associated anomalies may be present. This heterogeneity has complicated elucidation of the diagnosis and management of the syndrome.

Case report

A healthy 32-year-old women complained of torticollis. Her medical and family histories were unremarkable. Physical examination reveals Pain on palpation of vertebral spine apophyses and neck muscles. There is limited neck extension and rotation. Neurological examination revealed normal cranial nerve function. The muscle strength was grade 4 over limbs without muscle atrophy. Never associated anomalies were noted. A radiography show a C5-C6 fusion on the cervical spine and the isolated klippel Feil syndrome was diagnosed.

Discussion

In 1919 Feil defined three morphological subtypes of this anomaly (Table 1). Different classifications have been proposed and 4 classes (KF 1, 2, 3 and 4) was identified according to position of cervical vertebra fusion, status of familial trait and its characteristics (Clarke et al, 1999).

<table>
<thead>
<tr>
<th>Class of Klippel-Feil syndrome</th>
<th>Inheritance</th>
<th>Vertebral fusion</th>
<th>Overlap with Klippel and Feil's original classification</th>
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</thead>
<tbody>
<tr>
<td>KF1</td>
<td>autosomal recessive</td>
<td>Rostral fusion at C1 and severe associated anomalies (short neck, cardiac defects, and craniofacial anomalies)</td>
<td>Types I, II and III</td>
</tr>
<tr>
<td>KF2</td>
<td>autosomal dominant</td>
<td>C2-3 fusion and possible craniofacial anomalies</td>
<td>Types I, II and III</td>
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<tr>
<td>KF3</td>
<td>reduced penetrance</td>
<td>singular isolated fusion, most rostral at C3</td>
<td>Type II</td>
</tr>
<tr>
<td>KFS4</td>
<td>X-linked inheritance</td>
<td>vertebral fusion and ocular anomalies</td>
<td>Commonly referred to as Wildervanch syndrome</td>
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</table>

Here We report a case of an isolated klippel Feil syndrome with C5-C6 fusion on the cervical spine. It's the rarest form of congenital fused cervical vertebrae which is predisposed to the risk of spinal cord injury and neurologic problems. Although patients with Klippel-Feil syndrome are often asymptomatic, they may develop a number of spontaneous neurologic sequelae as result of their bony anomalies. Axial neck symptoms were highly associated with Type I patients, whereas predominant radicular and myelopathic symptoms occurred in Type II and Type III patients. All these complications simulate an acute pathology This classification system has promise for early detection for cervical spine-related symptoms (CSS).

Klippel-Feil syndrome is usually diagnosed in the patients during childhood. However; Clinical presentation varies because different associated syndromes and anomalies may occur in these patients lately (Naikmasur et al, 2011). Thus; the challenge of the specialist is to recognize the associated anomalies that can occur with Klippel–Feil syndrome and to perform the appropriate workup for diagnosis.

The prognosis for most individuals with Klippel-Feil Syndrome is good if the disorder is treated early and appropriately. Activities that can injure the neck should be avoided. Key considerations in the management of KFS include radiographic evaluation for hypermobile cervical segments, recognition of high-risk patterns of skeletal anomalies and proper referral because of the associated anomalies. Awareness of these congenital anomalies is important for several reasons. First, recognizing young patients should lead to more careful investigation for spinal cord syndromes (eg, central cord or anterior cord) that are otherwise more common in older individuals. Second; this awareness should also prompt patient education concerning their risks for spinal injury even after mild traumatic events. Lastly, the potential for other abnormalities should be considered and evaluated through appropriate referral.

Conclusion

We insist that clinical heterogeneity and radiographic abnormalities found in Klippel-Feil syndrome may simulate acute pathology and thus require comprehensive evaluation and delineation of diagnostic and prognostic classes.

References