Case Report

A giant hypertrophy of C5 spinous process in Klippel-Feil syndrome

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Summary

Congenital cervical spinal anomalies are relatively common and can be seen in upper and lower cervical regions. However, hypertrophy of the lamina and spinous process of cervical vertebra is extremely rare. Here we reported an 11-year-old girl with unilateral hypertrophy of the lamina and spinous process of C5 vertebra coexistence with Klippel-Feil syndrome, occipitalization of atlas and atlantoaxial congenital fusion. Because of cosmetic anomaly patient underwent surgery and spinous process excised. To the best of our knowledge, this coexistence of congenital cervical bony anomalies in a child has not been reported previously in English literature.

Key words: Congenital abnormalities, Unilateral hypertrophy of spinous process, Klippel-Feil Syndrome, Occipitalization of atlas, Atlantoaxial congenital fusion

INTRODUCTION

Congenital cervical spinal anomalies are relatively common and can be seen in upper and lower cervical regions. Various congenital bony anomalies of the upper and lower cervical spine have been reported such as; basilar invagination, atlantoaxial rotatory subluxation, occipitalization of atlas, Klippel-Feil syndrome, congenital absence or hypoplasia of a cervical pedicle, persistent apophyses of the transverse processes, vertebral hypoplasia and dysplasia of the vertebral arc \(^{(2,4,5,6,8)}\). In contrast, unilateral hyperplasia of the lamina and spinous process of cervical vertebra is extremely rare \(^{(2,4,5,6)}\) and its coexistence with Klippel-Feil syndrome, occipitalization of atlas and atlantoaxial congenital fusion in a child is never seen in English literature.
CASE PRESENTATION
An 11-year-old girl was admitted to neurosurgery department with complaints of swelling in posterior cervical region. On physical examination, we determined hard immobile mass with palpation in posterior cervical region. The neurological examination was normal. Plain radiographs of the cervical spine demonstrated hypertrophy of the C5 spinous process. Cervical computed tomography (CT) scan revealed left unilateral hypertrophy of the lamina and spinous process on axial plane (Fig-1A). Sagittal reconstruction of the CT identified Klippel-Feil syndrome (KFS) with fusion of C2-3 and C4-5-6 vertebrae (Fig-1B), occipitalization of the atlas and atlanto-axial congenital fusion. Cervical, thoracic and lumbar magnetic resonance imaging (MRI) did not demonstrate pathological findings such as intraspinal anomalies, Chiari malformation, diastematomyelia, or other spinal cord malformations. Also, we investigated whole body for several organ anomalies. However we could not detect additional abnormality such as congenital heart defects and renal anomalies. Because of suspicion for C1-2 instability, lateral, neutral and functional cervical x-rays were taken. Atlanto-dental interspace was not widened (less than 5 mm) and we did not determine excessive movement between C1 and C2 as the evidence for stability of upper cervical spine, which means atlantoaxial congenital fusion.

Surgical resection of C5 spinous process was planned because of cosmetic anomaly. C5 spinous process was resected with posterior midline approach and histological examination of the specimen revealed normal bone tissue. Postoperative cervical MRI demonstrated excision of C5 spinous process (Fig-1C). Postoperative period was uneventful and she was discharged on postoperative 3rd day.

DISCUSSION
Developmental abnormalities of cervical spine are common and could be diagnosed as a sole developmental defect or may seen as a part of congenital syndromes such as Klippel-Feil syndrome (KFS), Fibrodisplasia ossificans progressiva, Down’s syndrome, Rubinstein-Taybi syndrome, Morquio’s syndrome and Osteogenesis imperfecta(4,5,6,9). These abnormalities can cause clinical symptoms.

Figure-1: A-) Axial plane of cervical CT demonstrated right spinous process (black arrow head) and hypertrophy of the left lamina and spinous process (black arrow) B-) Sagittal reconstruction of cervical CT is showing Klippel-Feil syndrome with fusion of C2-3 and C4-5-6 (white arrow heads) C-) Postoperative cervical MRI demonstrated (yellow dotted line) wasp-waist sign and resection of C5 spinous process.
such as neck pain, neurological deficits, limitations of the cervical movements, cervical mass and aesthetic anomalies\(^{(2,4,5,6,9)}\).

The pathogenesis of unilateral hypertrophy of spinous process and lamina is not known. The development of vertebral column begins in 3-6\(^{th}\) weeks of gestation\(^{(4,5,6)}\). In 2\(^{nd}\) embryonic month ossification centers are appear however spinous process does not have ossification centre. Enchondral growing osseous extensions from both vertebral arches are fused and formed the spinous process in the first year of life. As the time passes tip of the spinous process develops in pubertal period from secondary ossification centre\(^{(5,6)}\). There are very few published cases with unilateral hyperplasia of spinous process and lamina in literature\(^{(2,4,5,6)}\). Summary of the case reports for unilateral hyperplasia of spinous process and lamina that we can found in literature are shown in Table-1. Our patient presented with posterior cervical mass. Radiological investigations revealed C5 unilateral hypertrophy of spinous process and lamina coexistence with Klippel-Feil syndrome, occipitalization of the atlas and atlantoaxial congenital fusion. Because of cosmetic anomaly in posterior cervical region, surgery was applied and she was discharged after an uneventful postoperative course.

Table-1: Summary of the case reports for unilateral hyperplasia of spinous process and lamina that we can found in English literature

<table>
<thead>
<tr>
<th>References</th>
<th>Age/Sex</th>
<th>Presentation</th>
<th>Pathology</th>
<th>Additional anomaly</th>
</tr>
</thead>
<tbody>
<tr>
<td>Heyer et al.(^{(5)})</td>
<td>24y/F</td>
<td>Neck pain</td>
<td>C7 unilateral hyperplasia SP+L</td>
<td>None</td>
</tr>
<tr>
<td>Esposito et al.(^{(2)})</td>
<td>5y/F</td>
<td>Cervical mass, neck pain</td>
<td>C6 unilateral hyperplasia SP+L</td>
<td>Schisis at the same level</td>
</tr>
<tr>
<td>Kazandi et.al (^{(6)})</td>
<td>20y/M</td>
<td>Traffic accident, neck pain</td>
<td>C6 unilateral hyperplasia SP+L</td>
<td>Schisis at the same level</td>
</tr>
<tr>
<td>Hedge et al. (^{(4)})</td>
<td>12y/M</td>
<td>Cervical mass, neck pain</td>
<td>C6 unilateral hyperplasia SP+L</td>
<td>Schisis at the same level</td>
</tr>
<tr>
<td>Present study</td>
<td>11y/F</td>
<td>Cervical mass</td>
<td>C5 unilateral hyperplasia SP+L</td>
<td>Klippel-Feil+ AACF+ OA</td>
</tr>
</tbody>
</table>

SP: Spinous process, L: Lamina, AACF: Atlantoaxial congenital fusion, OA: Occipitalization of atlas
Occipitalization of the atlas is one of the most common anomalies of craniovertebral junction\(^7,11\) and its incidence is between 0.08% to 2.8%\(^1\). Combination of C1 occipitalization and Klippel–Feil syndrome was reported by Gholve et al. In this study, KFS was present in 20% of patients who had C1 occipitalization\(^3\). KFS is a syndrome that is characterized by classical triad decreased cervical motion, short neck and low-set posterior hairline\(^1\). Its incidence is estimated approximately 1/2,100 births. KFS is divided in 3 types and; in type I, patients have a single congenitally fused cervical segment; in type II, patients have multiple non-contiguous, congenitally fused segments; and in type III, patients have multiple contiguous, congenitally fused cervical segments\(^9\). In type III, fusion of both cervical and lower thoracic or lumbar vertebrae occur. This syndrome may be associated with genito-urinary tract, cardiovascular and brain stem anomalies. Other skeleton system anomalies including Sprengel deformity, scoliosis and torticollis are also common findings. In addition, deafness and learning disabilities could be seen\(^1\). Our patient was success in school. She did not have any of the classical triad symptoms. Her CT determined multiple non-contiguous congenitally fused cervical segments, which indicates type II KFS. Also, we did not detect additional organ system anomaly in whole body investigation.

The radiological differential diagnosis includes variety of entities such as posttraumatic, postinfectious, rheumatoid arthritis and ossification of the posterior longitudinal ligament\(^10\). When the diagnosis of Klippel-Feil is suspected, one can perform conventional radiographs of the cervical spine for detection of fusion of the vertebral bodies. Other radiographic findings include fusion of the facet joints and spinous processes.

The wasp-waist sign, which is the anterior concave indentation at the site of the absent or reduced interspace between the fused vertebrae, with a smooth anteriorly concave surface forming a smooth arc along the anterior vertebral aspect is a valuable finding\(^10\). On our patient’s cervical MRI, yellow dotted line is showing wasp waist sign. Fused vertebrae form a continuous surface with concavity anteriorly (Fig 1C).

**CONCLUSION**

In the presence of subaxial spine abnormality such as unilateral hypertrophy of spinous process and lamina, remainder of the spine should be investigated for additional congenital abnormalities. If additional abnormality detected related radiological examinations should be done before surgery to prevent unexpected unpleasant outcomes.

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