Case Report

Report of two cases with omovertebral bone and Klippel–Feil syndrome with craniovertebral junction instability

ABSTRACT

We present a report of two patients having the association of omovertebra, Sprengel's deformity of the shoulder and Klippel–Feil abnormality with craniovertebral junctional instability. Our literature survey did not locate any report of such association. Significance of bone alterations is analyzed. Two young patients presented with neck pain, torticollis, webbed neck, and spastic quadriparesis. In both patients, the investigations revealed basilar invagination, Klippel–Feil abnormality and Sprengel's deformity of the shoulder. Apart from these relatively common associations, both the patients had omovertebral bone that extended from the transverse process of C5 vertebra to scapula. Following atlantoaxial stabilization surgery, the patients rapidly recovered from all symptoms. Musculoskeletal abnormalities at the craniovertebral junction that include Klippel–Feil abnormality, Sprengel's shoulder, and omovertebra are secondary alterations to primary atlantoaxial instability.

Keywords: Atlantoaxial instability, basilar invagination, Klippel–Feil abnormality, omovertebral bone, Sprengel's shoulder

INTRODUCTION

Approximately 20%–25% of patients with Sprengel's shoulder deformity have an omovertebral connection that is either fibrous or osseous between the superomedial border of the scapula and the spinous process, transverse process, or the lamina of the C4–7 cervical vertebrae.[1-3] We present a hitherto unreported association of Klippel–Feil abnormality, Sprengel's deformity, and omovertebral bone connection with basilar invagination and atlantoaxial instability. The significance of musculoskeletal alterations in cases with chronic or long-standing atlantoaxial instability is discussed on the basis of our earlier published work.

CASE REPORTS

Case illustration 1

A 23-year-old male patient complained of pain in the nape of the neck, restricted neck movements and stiffness and weakness of all four limbs for approximately 3 years. The symptoms were progressive. He had a short neck and torticollis since early childhood. When admitted, he had spastic quadriparesis-left side being worse, power in the limbs being graded as 4 and he could walk and perform only with support. There was no sensory deficit. His Goel clinical grade was Grade 3. He had short and webbed neck, low hairline and torticollis to the left features typically associated with Klippel-–Feil abnormality. He had a Sprengel's shoulder deformity on the left side and a firm bony swelling could be palpated over the shoulder that extended toward the cervical spine. Investigations included dynamic computed tomography (CT) scan with the head in the flexed and extended position and magnetic resonance imaging (MRI). Investigations revealed basilar

Abhidha Shah[1], Akshay Hawaldar[1], Aditya Lunawat[1], Saswat Dandpat[1], Atul Goel[1,2]

[1]Department of Neurosurgery, K.E.M. Hospital and Seth G.S. Medical College, [2]Lilavati Hospital and Research Centre, Mumbai, Maharashtra, India

Address for correspondence: Prof. Atul Goel, Department of Neurosurgery, K.E.M. Hospital and Seth G.S. Medical College, Parel, Mumbai - 400 012, Maharashtra, India.
E-mail: atulgoel62@hotmail.com

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invagination Group A that was associated with assimilation of the atlas, C2–3 fusion and fusion of C6 to T3 vertebral bodies [Figure 1]. There was vertically mobile and Type 1 atlantoaxial facetal instability.[4,5] Thick omovertebral bone connecting the posterior elements of C5 vertebra to the left scapula was identified. MRI showed odontoid process-related compression at the cervicomedullary junction and the presence of external syringomyelia in the cervical spinal region.[6] There was no neural compression related to omovertebral bone. The surgery involved atlantoaxial fixation with the techniques described by us earlier and are summarized here.[7] The patient was placed in the prone position with the head placed under Gardner-Wells traction and the head end of the operation table elevated by about 30°. The surgical procedure involved the opening of the atlantoaxial facetal articulation, denuding of the articular cartilage, stuffing of bone graft pieces within the facet joint and lateral mass plate and screw fixation. The presence of short neck, assimilation of atlas and omovertebral bone hump that interfered in obtaining suitable angle made the surgical procedure technically complex. Muscles attached to the C2-spinous process were sharply sectioned. No manipulation of the omovertebral bone was done. Posterior elements of the C2 vertebra and suboccipital bone were drilled to make the region suitable as host for bone graft harvested from the iliac crest and placed in the region. The patient was then placed in firm cervical collar for 2 months. The patient rapidly improved in all his symptoms as early as in immediate postoperative period. Postoperative imaging showed reduction in the basilar invagination and atlantoaxial dislocation [Figure 1]. Although not adequately measured, it seemed that the neck size increased and torticollis improved following surgery. At a follow-up of 27 months, he was essentially asymptomatic. The bone hump related to omo-vertebra persisted, but it appeared to be less prominent.

Case illustration 2

A 15-year-old female patient presented to another institute with the complaints of neck pain, headache, visual bobbing, difficulty in walking, disequilibrium, and frequent falls. On examination, she had a short stature, a short and webbed neck, low hairline and torticollis to the left, features typical of a Klippel–Feil syndrome. She also had Sprengel's shoulder deformity of the left side and a bony hump in the base of the neck related to underlying omovertebra. Neurological examination revealed spastic Grade 4/5 quadriparesis. She also had severe gaze-evoked nystagmus in all directions and a depressed gag reflex. She was unable to walk independently and needed support to walk. Her Goel clinical grade was Grade 4. CT scan of the cervical spine showed Group A basilar invagination, assimilation of the atlas, vertical mobile and Goel Type 1 atlantoaxial instability. There was the fusion of C6, C7 and T1 vertebrae. The spinous processes of all the cervical vertebrae were bifid. There was the presence of omovertebral bone that extended from the spinous process of the C5 vertebra to the medial border of the

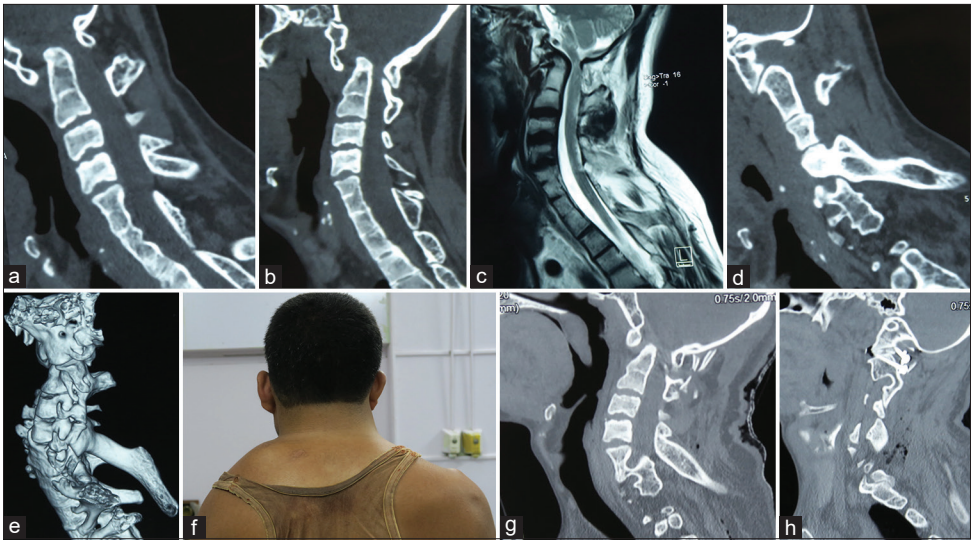

Figure 1: Images of a 23-year-old male. (a) Computed tomography scan with the head in flexion shows assimilation of atlas, C2–3 fusion, increased atlantodental interval and fusion of vertebral bodies C6–T3. (b) Computed tomography scan with the head in extension shows vertical atlantoaxial instability and mild reduction of dislocation. (c) T2-weighted magnetic resonance imaging shows compression of the neural structures opposite the odontoid process. (d) Computed tomography scan with the cut passing through the facets showing atlantoaxial facetal instability. Omovertebra can be seen arising from C5 vertebral body in the region of the transverse process. (e) Three-dimensional computed tomography scan showing the large omovertebra arising from the C5 body and travelling toward the scapula. (f) Photograph of the back of the patient showing the hump over the left shoulder made by the underlying omovertebra. (g) Postoperative computed tomography scan showing reduction of the instability and fixation. (h) Postoperative computed tomography scan image through the facets showing the implant

scapula. CT angiography revealed an abnormal course of the left vertebral artery, which coursed over the lamina of C2 vertebra. MRI showed basilar invagination, Chiari formation and syringomyelia. The patient was operated at another institute and a craniovertebral stabilization surgery was planned. However, due to the complex craniovertebral junction alterations, the surgeons performed only a foramen magnum decompression with duroplasty. The patient had marginal improvement in her symptoms following surgery but after about 5 months of surgery, she again started worsening. When she presented to us she had severe nystagmus in primary position, which disturbed her vision, frail voice and breathing difficulty. She needed heavy support to stand or walk as she had severe imbalance on walking. The patient was re-operated and an atlantoaxial fixation was performed. The key surgical steps have been highlighted above. A C1 lateral mass and C2 pedicle screw fixation were performed on the right side after exposure of the joint. In view of the complex craniovertebral junction architecture and the presence of anomalous vertebral artery loop on the left side a C1–C2 transarticular screw fixation was performed. No manipulation of the omovertebral bone was done. The patient improved in all her symptoms as early as in immediate postoperative period. Her neck size increased, and torticollis improved. Her voice became louder and her breathing was better. Postoperative imaging showed reduction in the basilar invagination and atlantoaxial dislocation [Figure 2]. At a follow-up of 14 months, she was relieved of all her major symptoms and could walk unaided and perform household activities independently.

DISCUSSION

Basilar invagination is frequently associated with Klippel–Feil abnormality and Sprengel's shoulder.[8,9] Assimilation of atlas and C2–3 fusion are the more common bone fusions seen in association with basilar invagination. Klippel–Feil abnormality consists of a triad of short neck, low hairline, and restricted neck motion.[9] It is characterized by the fusion of two or more cervical vertebrae and has been known to be associated with atlantoaxial instability. Sprengel's deformity is categorized as an abnormal dysplastic and superiorly elevated scapula resulting in the neck and shoulder pain, restricted movements and a cosmetic deformity.[10] It is associated with Klippel–Feil abnormality in 16%–27% of cases.[3]

The significance of omovertebral bone, cartilage or fibrous tissue connection between lower cervical vertebrae with medial edge of the scapula has not been appropriately defined in the literature. It was first described by Willet and Walsham in 1880 and is associated with Sprengel's deformity in 30%–60% of patients.[3,11] The indication of surgery in patients with Sprengel's deformity and omovertebral bone is usually for cosmesis and improvement of range of motion.[2,3] Rarely, the omovertebral bone has been implicated to cause cervical myelopathy and has been treated with laminectomy and excision of the omovertebral bone.[1,3]

In 2009, we reported that musculoskeletal alterations that include short neck and torticollis and Klippel–Feil bone fusions are not primary pathological issues or related to embryological dysgenesis but are secondary and probably protective natural responses to chronic or long-standing atlantoaxial dislocations.[8] It was speculated that long-standing atlantoaxial instability initiates protective neck movement restriction; muscle spasm and muscle contractures lead to reduction in the disc space height, osteophyte formation and ultimately bony fusion.[8,12,13] We also identified that Chiari formation and syringomyelia are neural manifestations of chronic atlantoaxial instability.[14] The fact that several musculoskeletal and neural alterations are reversible following surgery for atlantoaxial stabilization provides credence to the hypothesis. No direct manipulation of bones or soft tissues is necessary in such clinical situation. Although it was not identified, we reported that there is a potential for bone fusions at subaxial spinal levels to unfuse following atlantoaxial fixation.[8]

Klippel–Feil abnormality and Sprengel's shoulder deformity are common associations of basilar invagination and craniovertebral junction instability. Although the association of omovertebral bone with Klippel–Feil abnormality and with Sprengel's shoulder has been described, its clinical correlation with craniovertebral junctional instability has never been reported.

Our observation that is based on experience with a large number of cases with basilar invagination in general and following treatment of the cases in discussion, in particular, is that omovertebral bone formation could be a part of secondary alterations related to craniovertebral junction instability. Further reasoning on this hypothesis leads one to wonder whether the omovertebral bone is a natural formation that provides stability to the neck in the form of internal arthrodesis. The location and physical form of omoverterba simulate a bicycle stand as if it is introduced by Nature to support the unstable neck. The long-term consequences and effects of atlantoaxial stabilization on omovertebral bone remain to be seen. However, it does appear that when omovertebral bone is identified, it is mandatory that the craniovertebral junction is appropriately assessed and when necessary atlantoaxial instability is treated.

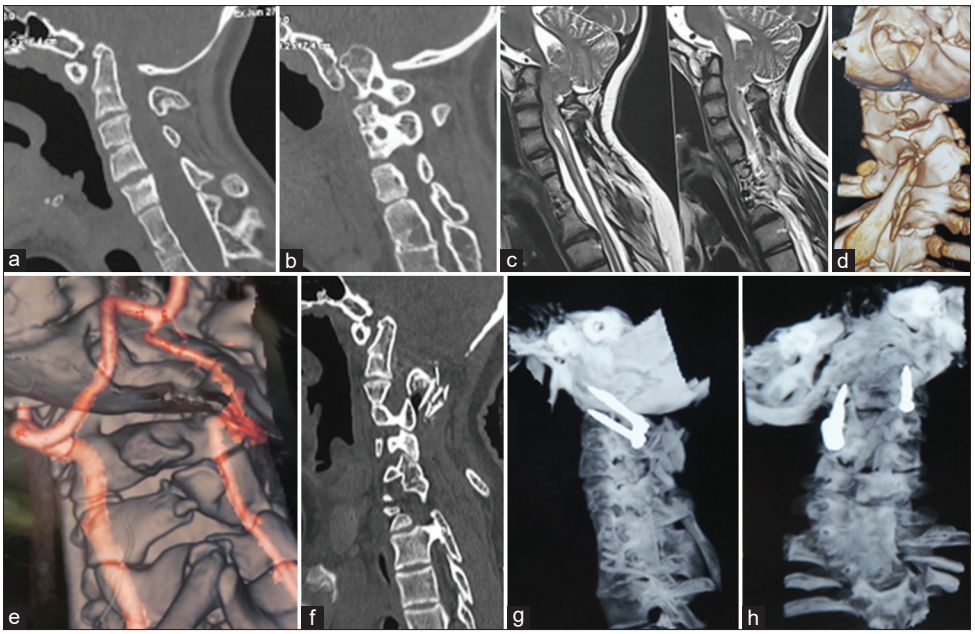

Figure 2: Images of a 15-year-old female patient. (a) Computed tomography scan shows assimilation of atlas, Group A basilar invagination, increased atlantodental interval and fusion of vertebral bodies C6–T1. (b) Computed tomography scan with the cut passing through the facets showing atlantoaxial facetal instability. (c) T2 weighted magnetic resonance imaging showing basilar invagination, Chiari formation and syringomyelia. (d) Three-dimensional computed tomography scan showing the omovertebra arising from the C5 body and travelling toward the scapula. (e) Computed tomography angiography showing the abnormal course of the vertebral artery on the left side. (f) Postoperative computed tomography scan showing reduction of the instability and fixation. (g) Postoperative computed tomography scan image showing the implant. (h) Antero-posterior view of postoperative computed tomography scan showing C1 lateral mass and C2 pedicle screw fixation on one side and C1–C2 transarticular fixation on the other side

CONCLUSION

The presence of omovertebral bone formation could indicate craniovertebral junction instability.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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