

A Concomitant Occurrence of the Atlantoaxial Subluxation with Rare Vertebral Formation and Segmentation Defects

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An atlantoaxial subluxation from the unstable Os odontoideum by the failure of proper integrations between the embryological somites might be a commonly reported pathology. However, its suspicious origin or paralleled occurrence with other congenital anomalies of vertebral body might be a relatively rare phenomenon. The authors present two cases, who simply presented with clinical signs of prolonged, intractable cervicgia without any neurological deficits, revealed this rare feature of C1–2 subluxation from the unstable, orthotropic type of Os odontoideum that coincide with congenitally fused cervical vertebral bodies between C2–3. Surprisingly, in one case, when traced from the lower cervical down to the thoracic-lumbar levels during the preoperative work-up process, was also compromised with multi-level butterfly vertebrae formations. Presented cases highlight the association of various congenital vertebrae anomalies and the rationale to fuse only affected joints.

Key Words : Atlanto-axial joint · Congenital anomaly · Os odontoideum · Joint instability.

INTRODUCTION

The odontoid process is a part of the C2 or axis vertebra and might be the central pillar that forms the crucial pivot among the consisting structures inside the craniovertebral junction⁵⁾. The development of odontoid is a complex process that is already involved with four ossification centers from the birth⁹⁾. Due to these embryological complexity, it might be vulnerable to a variety of congenital and acquired conditions such as the occurrence of an os odontoideum with its subsequent instability by atlantoaxial dislocation (AAD).

Apart from rarity, its combination with multiple congenital

formation and segmentation defects inside the vertebral column might put a demanding task over a radiologist or surgeon in the selection of a description or terminology for a proper diagnosis as well as its interpretation, and would subsequently be in dilemma in its management choice.

The authors present two case of orthotropic type Os odontoideum induced AAD combined with the congenitally fused cervical vertebrae. One of two cases was treated by C1–C2 fusion and the feasible rationale for such a combination of rare congenital formation and segmentation defects is discussed.

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CASE REPORT

Case 1

This 65-year-old female presented with the clinical signs of neck pain and subjective systemic spasticity spanning over recent few years. However, the objective finding acquired during the detailed clinical examinations was limited to an intractable cervicgia provocation only especially during the neck extension without any neurological deficits.

The initial dynamic radiographies of the cervical spine revealed the AAD instability phenomenon on neck extension, which was completely reducible during the patient's self-flexional maneuver, suggesting a flexibility for this condition (Fig. 1). The main background causing this AAD instability was found out to be the inherent existence of an orthotropic type of Os odontoideum, as revealed through the 3-dimensional reconstructed images by the computed tomography (CT). Surprisingly, as concomitantly featured inside the same frame through these CT scans, there were coexistent anomalous features of the inherently fused cervical spine between C2–3 level and inside C3–4 vertebral bodies, respectively. This congenitally fused, bony bridging was most notable along the both anterior and posterior vertebral body margins as well as between the lateral masses and spinous processes across the C2–3 (Fig. 2).

In a whole spinal evaluation in order to detect the other fea-

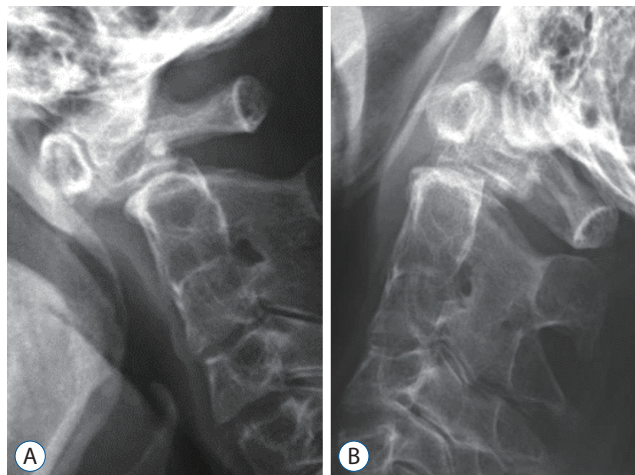


Fig. 1. The dynamic radiographies for the patient during the flexion (A) and extension (B) feature an unstable atlantoaxial dislocated condition. Note the complete reducibility for this condition during the patient's self-flexional maneuver. The congenitally fused vertebrae between C2 and C3 with its prominent bony bridging formation between the spinous processes is also noted.

sibly affected levels, she was also compromised with the multilevel butterfly vertebrae formations when traced from the lower cervical down to the thoracic-lumbar junctional levels by the CT work-up process (Fig. 3). Fortunately, none of these butterfly vertebrae infiltrated levels was affected with other pathologic or degenerative spinal conditions mandating another treatment. Also, there was no other congenital syndromes or anomalies involving the systemic organs associated.

A reduction by posterior screw fixation augmented with allografts on lay over the C1–2 level was schemed and performed to completely resolve the initial intractable discomfort as well as to deter further neurological deterioration for the patient by providing a solid stability at the corresponding unstable level. After this reductive operation, full radiological restoration of the AAD instability as well as the complete resolution of the original cervicgia was achieved for the patient (Fig. 4).

Case 2

A 26-year-old female patient was referred to the outpatient department seeking for an accurate diagnosis on her cervical spinal condition after having suffered from the intractable cervicgia as well as ensuing cervicogenic headache for over the past 3 years. Despite the clinical process had become particularly worse during the past 2 months prior to this visit, she

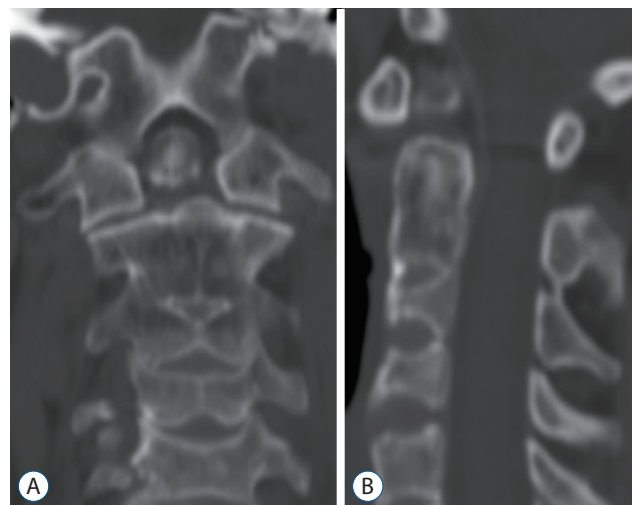


Fig. 2. The 3-dimensional reconstructed coronal (A) and sagittal (B) scans by the cervical spine computed tomography reveal an orthotropic type of Os odontoideum, margined and separated from the caudal C2 body by clean cortical bone. The fused, bony bridging phenomena are prominent along the both anterior and posterior vertebral body margins as well as between the lateral masses of the C2, C3, and C4.

was not aware or informed of her feasible pathologic condition due to the seldom visits to the non-eligible pain clinics for the

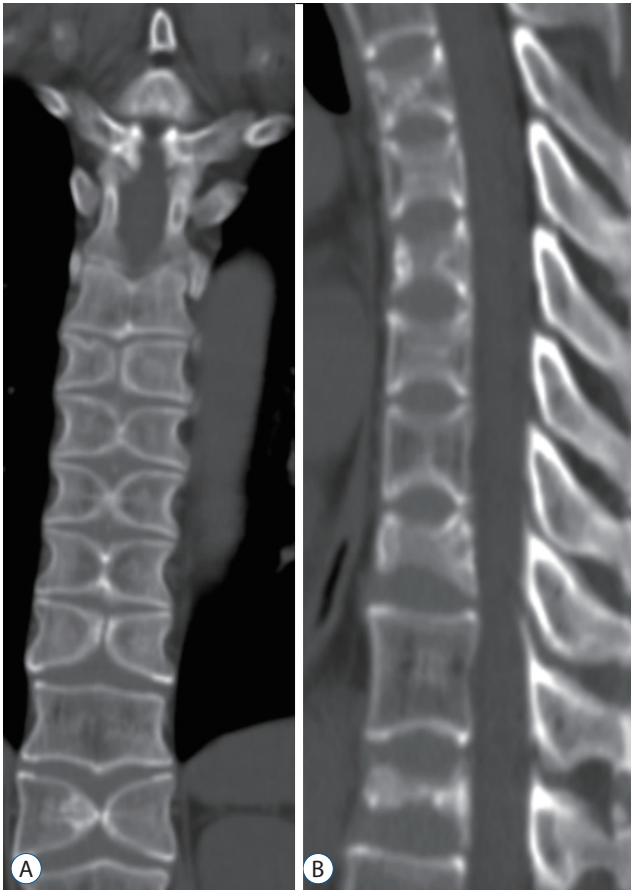


Fig. 3. The 3-dimensional reconstructed coronal (A) and sagittal (B) computed tomography scans with the inclusion of the whole vertebral levels clearly depict the extension of the multilevel butterfly vertebrae formations from the lower cervical to thoracic-lumbar junctional levels with the few intervening normal shaped vertebrae.

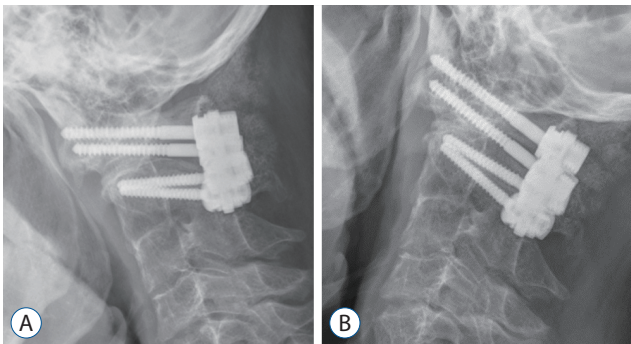


Fig. 4. The dynamic radiographs during the neck flexion (A) and extension (B) taken 9 months' post-operative period after the reduction with allograft on-layed feature a full radiological restoration of the inherent atlantoaxial dislocated instability.

sake of mere, temporary symptom relief during her daily career.

The lateral view among the plain radiographs of her cervical spine showed the AAD feature with slightly retropulsed Os odontoideum over C2 body proper, which was untowardly stabilized by cortical rim connection with ventral portion of the C3 vertebra (Fig. 5). She was not managed to take additional dynamic radiographs for the further verification of AAD phenomenon due to the exaggerated symptomatic provocation during the enforced neck extension maneuver. As differentiated from the lower subaxial cervical vertebrae, the decreased anterior-posterior (AP) diameter of the vertebral bodies are also noted at the fused C2, C3, as well as C4 levels.

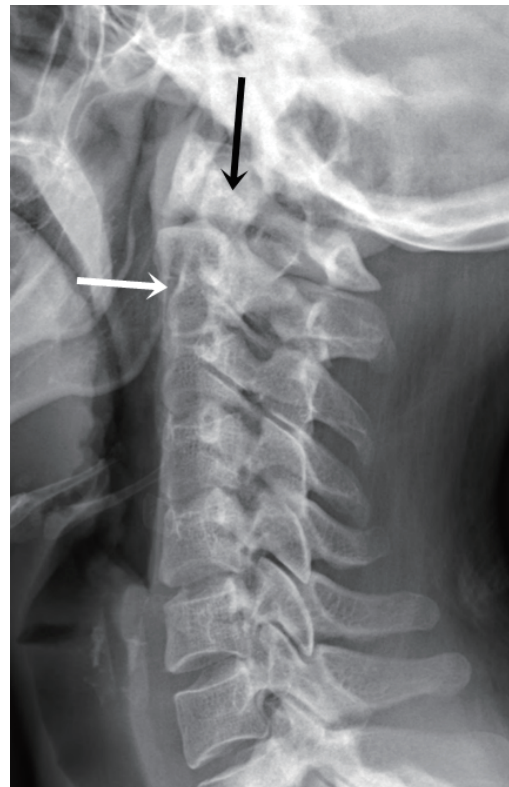


Fig. 5. The plain radiographs, lateral view of the case 2 showed the AAD feature with slightly retropulsed os odontoideum over C2 body proper (black arrow), which was untowardly stabilized by cortical rim connection with ventral portion of the C3 vertebra (white arrow). Note the decreased anterior-posterior diameter of the vertebral bodies at the fused C2, C3, as well as C4 levels as compared with the lower subaxial cervical bodies.

DISCUSSION

Congenital fusion of cervical vertebrae (CFCV) from the segmentation defects of cervical spine is a rare anomaly but do not have low incidence^{4,7)}. Although this fused condition might be either congenital or acquired, the most important differentiated radiographic aspect for CFCV would be the decreased AP diameter of the vertebra, as seen in this case. Also, the individual measurements of the two vertebrae bodies' height is equal to the two fused vertebrae's height including the intervertebral disc¹⁸⁾. One of the proposed reason for the development of a CFCV would be a disturbance in normal spinal segmentation during embryological development, followed by a decrease in local blood supply between 3rd to 8th week of embryonic period¹⁴⁾. In this condition, two fused vertebrae appear structurally and functionally as one unit¹⁸⁾ and subsequently might result in a biochemical stress overload to the adjoining intervertebral functional unit, which could lead to a premature degenerative spondylotic change or even a graver consequence like instability problems at the adjacent segments^{13,14)}. This condition has been sporadically reported to predispose a congenital AAD development, with its preferential level would be between the facet joints of the C2 and C3, as depicted in this current case^{10,16,17)}. Fused cervical vertebrae with congenital AAD is rare, but some cases have been reported (Table 1). Unlike the previous reports, the presented case is very rare in that it was occurred with thoracic butterfly vertebrae.

The presence of adjacent multilevel butterfly vertebrae complicates this combination of anomalies further. These anomalies might originate from the defects pertaining to formation (either partial or complete) and the intercalation of central ossification center or a sagittal cleft caused by the persistence of the notochord might give rise to a butterfly vertebra⁸⁾. The

two parts of bodies are connected by cartilage that prevents splaying¹²⁾ and possibly transmits the body weight efficiently to the normal vertebral bodies below. Due to this efficient load transmission, they are usually asymptomatic and do not require treatment¹¹⁾. Nonetheless, multiple previous reports have linked butterfly vertebrae with low back pain which might be the most common presenting complaint occurring in 23% of all cases and 30% of non-syndromic cases⁶⁾. However, still, the association between butterfly vertebrae and low back pain remains unproven and might be unrelated to the structural abnormality; thus the etiology of this phenomena has not been rigorously explained.

It is important to understand the radiographic features of butterfly vertebrae since they have been frequently misdiagnosed as one of the compression fracture previously. Close examination of the adjacent vertebrae will show the features of long-standing deformity such as exaggerated elongation anterior margins to occupy the defect, and a normal intervertebral disc²⁾. In addition, butterfly vertebrae typically cause less kyphosis than would be expected from the significant anterior wedging¹⁾.

According to a systematic review from Katsuura and Kim⁶⁾, there was a high association between the presence of multiple butterfly vertebrae and a syndromic presentation (40/45 cases, 89%). The associated organ system that is most frequently vulnerable would be any structural abnormality inside the vertebral column that is not affected with the butterfly vertebrae, such as scoliosis (most common), failure of formation and segmentation, block vertebrae, kyphosis, and spina bifida. However, despite pre-existing several descriptions on these findings with concomitant neurological deficits, these reports were mostly based on the clinical symptoms as opposed to structural problems, and perhaps were unrelated directly to the butterfly vertebrae³⁾. Therefore, the feasible hypothesis on

Table 1. List of congenital atlantoaxial instability previously reported in the literatures

Study	Age (years)	Sex	Fused segment	Butterfly vertebrae	Others
Deepak et al. ³⁾ (2017)	14	F	C2-3	C3	Absent posterior C2
Shah et al. ¹⁵⁾ (2018)	4	M	C2-7	-	1 patient; assimilation of atlas
	5	F	C2-6 or 7	-	3 patients; bifid anterior and posterior arches of the atlas
	14	F	C2-7	-	
	27	F	C2-6 or 7	-	

F : female, M : male

how these vertebrae formation and segmentation defects have been elucidated simultaneously as in this case might not be available even up to recent evidences. A consideration in terms of sclerotomes developmental anomaly that might interpret these phenomena as a whole should be pondered on by the author in the future.

CONCLUSION

The combinations of vertebrae formation and segmentation defects might coexist. However, the combination of multilevel butterfly bodies along with congenitally fused cervical vertebrae as well as AAD is a rare phenomenon. Their presences should raise suspicion for a coexisting broader congenital syndrome and should prompt the systemic diagnostic workup. However, the presence of such defects would not preclude or alter the management of AAD. Fusing the C1–C2 joints despite associated other complex anomalies appears to be a balanced approach as in this case.

CONFLICTS OF INTEREST

No potential conflict of interest relevant to this article was reported.

INFORMED CONSENT

Informed consent was obtained from all individual participants included in this study.

AUTHOR CONTRIBUTIONS

Conceptualization : JHL

Data curation : JHL

Formal analysis : MKC

Methodology : MKC

Project administration : SBK

Visualization : MKC

Writing - original draft : JHL

Writing - review & editing : MKC

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