Case Report

Multiple developmental craniovertebral junction anomalies associated with fusion of the third and fourth cervical vertebrae: A case report and review of the literature

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ABSTRACT

Craniovertebral junction (CVJ) is formed by the occipital condyles of the skull, atlas (C1), and axis (C2). CVJ anomalies are a complex group of disorders with a variable course and complicated line of management. A cross-sectional imaging is indispensable for the evaluation of CVJ as it may unmask pathologies unsuspected by clinical evaluation and conventional radiography. We present a case report in which we describe multiple developmental CVJ anomalies in a 25-year-old male patient. These include fusion defects of anterior and posterior arches of C1, os odontoideum, atlantoaxial subluxation (with compression of the cervical cord at the cervicomedullary junction), and fusion of C3 and C4 vertebral bodies as well as posterior elements.

Key words: Craniovertebral junction, CVJ anomalies, Vertebral fusion defects, Os odontoideum, Atlantoaxial subluxation

Craniovertebral junction (CVJ) is the transition zone between the base of the skull and spine. It is formed by two joints: Atlanto-occipital joint and atlantoaxial joint. Congenital or acquired disease processes of the occipital condyles, atlas (C1), and axis (C2) vertebrae or their articulations may result in CVJ anomalies (CVJAs) [1]. Developmental CVJAs tend to present insidiously and lead to a range of clinical manifestations due to sensorimotor abnormalities, brainstem dysfunction, cranial neuropathy, and vascular compromise [2]. CVJAs have been traditionally classified as reducible and irreducible, depending on the feasibility of surgical correction. Radiology, especially cross-sectional imaging, plays a key role in the assessment of patients as it can evaluate the alignment, direction of the mechanical compression, and reducibility of CVJAs. Imaging is doubly important as it can detect coexistent pathologies in the CVJ which can complicate the management [3,4]. Herein, we report a rare case of a young adult male patient suffering from multiple CVJ anomalies. As per our knowledge, no similar case has been reported in the past.

CASE REPORT

A 25-year-old male patient presented with complaints of low-grade neck pain, weakness of both the lower limbs, and fecal incontinence. According to the patient, the symptoms began at the age of 13 years and had progressed gradually, so that, now, he was bedridden for 6 months. There was no history of trauma. On examination, the vitals were within normal limits. Power was reduced in all four limbs. Saddle anesthesia was present. Anal sphincter tone was reduced. There was no cranial nerve dysfunction.

A non-contrast computed tomography (CT) of the CVJ was performed which showed a fusion defect of the anterior as well as the posterior arch of the C1 vertebra (Fig. 1). Os odontoideum was present, the odontoid process of the C2 vertebra was expanded with a globular contour, and the atlanto-dens interval was increased (measuring approximately 8 mm) causing severe narrowing of the spinal canal with compression of the cord at cervicomedullary junction. Vertebral bodies as well as the posterior elements of C3 and C4 vertebrae were fused (Fig. 2). Clinical and imaging findings were diagnostic of multiple CVJAs. The patient was offered surgical decompression for the correction of atlantoaxial instability but he declined and was subsequently lost to follow-up.

DISCUSSION

Depending on the anatomical structure involved, developmental CVJAs can be classified into those involving the occiput (condylus

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tertius, condylar hypoplasia, basiocciput hypoplasia, and atlanto-occipital assimilation), those involving the first cervical vertebra, or atlas (anterior arch anomalies, posterior arch anomalies, and split atlas), those involving the second cervical vertebra, or axis (persistent ossiculum terminale, os odontoideum, and odontoid aplasia), and others (fusion of two or more vertebrae, atlantoaxial subluxation) [2].

Fusion anomalies involving the posterior arch of the atlas are relatively common, with the defect ranging from failure of midline fusion to complete absence of the posterior arch. Currarino *et al.* have classified the posterior arch defects from types A to E (Table 1). Our case had a type E posterior arch defect [5].

Fusion anomalies of the anterior arch are much rare. In their 2006 retrospective study, Senoglu *et al*. found that the incidence of anterior arch defect was only 0.09% [6]. Similarly, a 2009 study by Kwon *et al*. reported the frequency of the anterior arch defects to be 0.087% [7]. It is important to distinguish fusion defects from fractures [8].

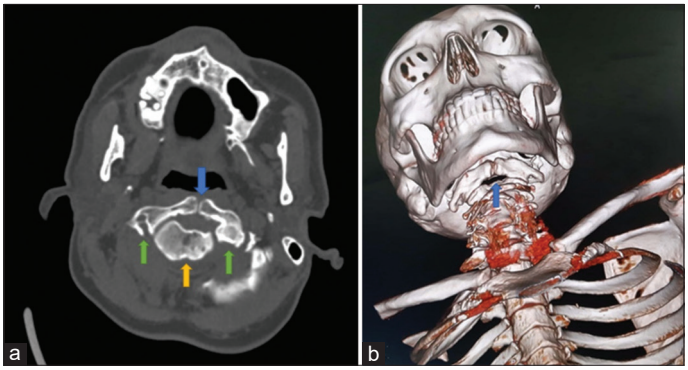

Figure 1: (a) Axial bone window CT at the level of C1 vertebra shows a fusion defect of its anterior arch (blue arrow) as well as posterior arch (green arrows). Odontoid process of C2 (yellow arrow) is expanded and globular; (b) 3D reconstruction shows the fusion defect of anterior C1 vertebral arch (blue arrow)

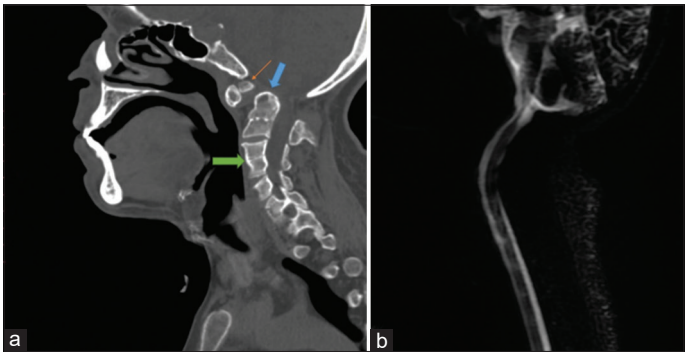

Figure 2: (a) Sagittal bone window CT depicts an os odontoideum (thin orange arrow), expanded and globular shape of the odontoid process of C2 vertebra (block blue arrow), and fused C3 and C4 vertebral bodies (block green arrow). Posterior elements of C3 and C4 vertebrae were also fused (not shown). Increased atlanto-dens interval can be appreciated; (b) sagittal MR myelogram image of the same patient shows severely thinned spinal cord at the cervicomedullary junction

Table 1: Currarino classification of posterior arch defects

Types	Defects
A	Posterior midline fusion defect of two hemiarches
B	Unilateral arch defect
C	Bilateral arch defect
D	Posterior arch is absent, persistent posterior tubercle
E	Entire posterior arch, including posterior tubercle, is absent

Os odontoideum is a smooth bony ossicle that has separated from a shortened odontoid process and demonstrates continuity with the C2. It may be normal in position, moving normally with the anterior arch ("orthotopic") or displaced ("dystopic"). Although asymptomatic in many cases, it is a risk factor for atlantoaxial instability and may lead to compression of the cervical cord [9]. In our case, os odontoideum was associated with atlantoaxial subluxation and a malformed, globular odontoid process.

Block vertebra results from the fusion of adjacent vertebral bodies. Fusion of the posterior elements is a frequent association in such cases. Both these features were present in our case. Block vertebrae are often asymptomatic but may predispose to biomechanical instability [10].

CONCLUSION

CVJAs are not a single entity but comprise a plethora of malformations and pathologies. Moreover, multiple anomalies can coexist in a single patient. A cross-sectional imaging is a must to define the extent of the disease and plan the management which is often multimodal and multispecialty.

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