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Case Report

Agenesis of the posterior arch of the atlas and complex alterations of the craniovertebral junction: A case report

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ABSTRACT

The craniovertebral junction is a unique part of the somite-derived axial skeleton. The absence or hypoplasia of the posterior arch of C1 is frequently associated with compensatory hypertrophy of the anterior arch of C1 and of the spinous process of C2. Here, we report a patient with agenesis of the posterior arch of C1 without neurologic deficits. Our patient presented with complex alterations of the craniovertebral junction that involved interactions between the condyles, clivus, atlas, and epistropheus. To our knowledge, dislocation of the odontoid process above the Chamberlain line, including cranial migration of the anterior arch of C1, has not been reported in the literature.

Introduction

The craniovertebral junction (CVJ) comprises the occiput, atlas (C1), axis (C2), and supporting ligaments, as well as soft tissue structures such as the medulla, spinal cord, and lower cranial nerves. Development of the CVJ is complex and developmental anomalies may occur due to abnormal resegmentation of sclerotomes. Congenital defects of the posterior atlas arch are rare, are generally asymptomatic, and are usually detected incidentally [1–3]. Congenital segmentation and developmental anomalies of the atlas may be associated with multiple alterations of the joint between the clivus, atlas, and dens. The absence or hypoplasia of the posterior arch of C1 is frequently associated with compensatory hypertrophy of the anterior arch of C1 and the spinous process of C2. However, the stability of the CVJ is dependent on the integrity of the articular surface and on the arrangements of muscles and ligaments. In early studies, radiography was the only imaging method used to evaluate the CVJ morphology (on lateral cervical spine and skull plain radiographs). However, the modern computed tomography (CT) and magnetic resonance imaging (MRI) have improved their technological capability, offering a high quality 3-dimensional visualization of the CVJ [4–7]. CT evaluates bones very well, but MRI is absolute necessary in joint, ligament, vascular, and soft tissue evaluation.

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Here, we report a patient with agenesis of the posterior arch of the C1 that was associated with complex alterations of the CVJ.

Case report

A 20-year-old male underwent diagnostic examinations for the onset of blurry vision. He developed his symptom suddenly. He denied symptoms suggestive of demyelinating disease and he had no prior history of headache. Imaging procedures were ordered by the neurologist. MRI and CT did not reveal any parenchymal alterations (Fig. 1e). However, we observed a complex malformation of the CVJ that was primarily characterized by agenesis of the posterior arch of C1 (Fig. 1a-d). Therefore, the following craniometric parameters were assessed.

1. Chamberlain line (a line joining the basion to the opisthion): the anterior arch of the atlas and the odontoid process lie below this line. In general population, the odontoid process is 1-6.6 mm from the Chamberlain line. The length of the Chamberlain line was 98.8 mm in this patient (Fig. 2a). The odontoid process and the anterior arch of the atlas both were 21.6 mm above this line.
2. Wackenheim clivus baseline (a line drawn from the clivus and extending into the upper cervical canal): the clivus-canal angle, the angle between the main axis and the Wackenheim clivus baseline, was 143.5° (normal range, 150° to 180° in extension; Fig. 2b).
3. McRae line (a line joining the anterior and posterior margins of the foramen magnum; the odontoid process normally lies below this line): this line was not overlapped by the odontoid process of C2 in this patient (Fig. 2c).
4. Height index of Klaus (the distance between the tip of the dens and the tuberculum cruciate line): this was 29 mm (normally 40-41 mm) and formed a partial basilar invagination (Fig. 2d).
5. Welcher basal angle (the intersection of the nasion-tuberculum line and the tuberculum-basion line): this was 141°, but is normally <140° (Fig. 2e).
6. Atlantooccipital joint axis angle: this angle was inverted with a value of 201° (normal range, 124°-127°) due to aplasia of the condyles (Fig. 2f).

After 1 week, the patient had a spontaneous resolution of the symptom. Because the patient was asymptomatic, no intervention was suggested, but only a specialist follow-up was advised to him.

Discussion

The CVJ of this patient had the following features: agenesis of the occipital condyles; agenesis of the caudal tubercle of the clivus; recognizable spheno-occipital synchondrosis and a basiocciput derived from the first 2 sclerotomes; agenesis of the posterior arch of the atlas; cranial migration of the anterior arch of the atlas, interposed between the odontoid process of C2 and the residual portion of the basiocciput; hypertrophy of the odontoid process of C2; and hypertrophy of the spinous process of C2.

These findings highlight the complex alterations of the CVJ in this patient that involved interactions between the condyles, clivus, atlas, and epistropheus (Fig. 1a and b). These alterations, which reflect the anatomy of the vertebrobasilar-vascular system and the elongated medulla, caused marked lateral dislocations of these structures (Fig. 1c and d).

Currarino et al [1] described 5 types of hypoplasia of the posterior arch that varied by the level of hypoplasia and the presence/absence of the posterior tubercle. Our case belongs to type E, namely total agenesis of the entire posterior arch, including the tubercle. Hypoplasia of the basiocciput and agenesis of the occipital condyles resulted in basilar invagination in our patient. Furthermore, although the Welcher basal angle was slightly greater than normal in this patient, we could not recognize the anthropometric elements of the platybasia, suggesting that basilar invagination and platybasia may be independent [8,9]. The use of these measurements (lines, distances, and angles) helps the diagnosis of alterations of CVJ. However, there has been considerable variation in craniometric parameters across studies. This variance could be related to a lack of validation and standardization of the old craniometric techniques adapted to CT and MRI [10,11].

The abnormalities described in this case report were detected incidentally by CT and MRI and were not apparently related to the patient's symptom. The odontoid process of the C2 joined the anterior arch of the atlas and did not extend beyond the plane of the foramen magnum, without apparent impingement on the nervous system. For the first time in the literature, we describe this configuration as being compatible with life in an asymptomatic patient.

In our opinion, the presence of multiple alterations did not elicit secondary compensatory changes, but instead might be the result of a single phenomenon of abnormal embryogenesis of the CVJ caused by defective development of occipital sclerotome IV, which is the origin of all of the altered structures in our patient.

Normal development of the first cervical sclerotome generally results in normal (hypertrophic) development of the odontoid process of C2 and the anterior arch of the atlas, but not the lower portion of the posterior arch. This event is the only secondary alteration in our patient, and probably resulted from the absence of the molecular and/or cellular environment provided by occipital sclerotome IV.

These features support the hypothesis that disturbances in gene expression involved in the transcription of *PAX* genes, a gene family that plays critical roles in the formation of tissues and organs during embryonic development, can affect the control of sclerotome resegmentation [12].

The complexity of the alterations described here demonstrates the unique embryogenic and morphostructural characteristics of this region. This case confirms that the anatomy of the CVJ should be evaluated using topographic and morphogenetic approaches. To our knowledge, no similar cases have been reported in the medical literature. This case report underscores the importance of careful diagnosis of CVJ anomalies to support their eventual treatment. The early diagnosis of these abnormalities may help to reduce the severity of their

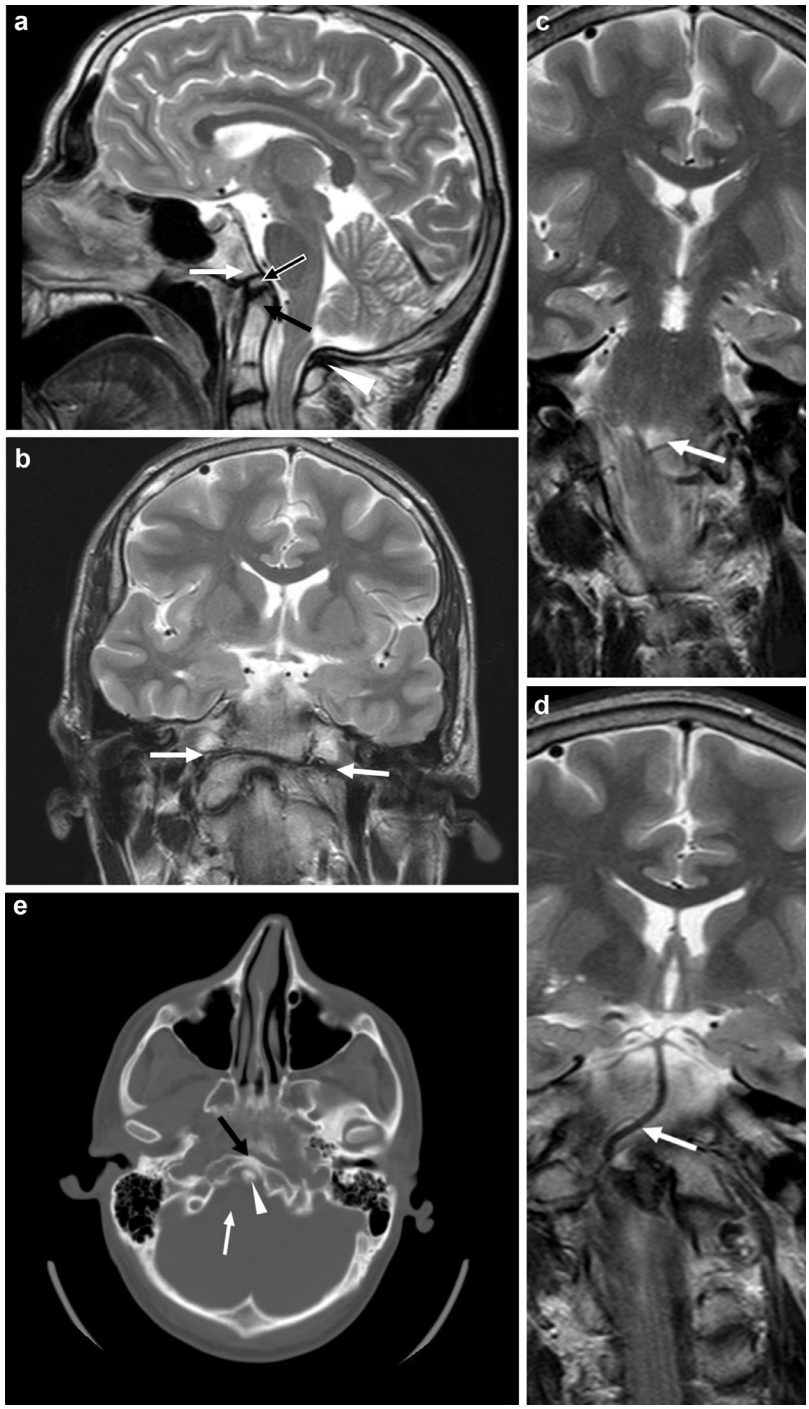

Fig. 1 – (a) MR sagittal section shows clivus agenesis (white arrow); hypertrophy of the C2 dens (black arrow); and anterior arch of atlas (bicolor arrow); agenesis of the posterior atlas arch (arrow head). (b) MR frontal section shows agenesis of occipital condyles (white arrows) and inversion of atlantooccipital joint axis angle. (c) MR frontal section: medium-lateral dislocation of the medulla oblongata and pons (white arrow). (d) MR frontal section: medium-lateral dislocation of the basilar artery (white arrow). (e) CT transverse section shows C2 dens (arrow head), anterior arch of atlas (black arrow), and agenesis of the posterior atlas arch (white arrow).

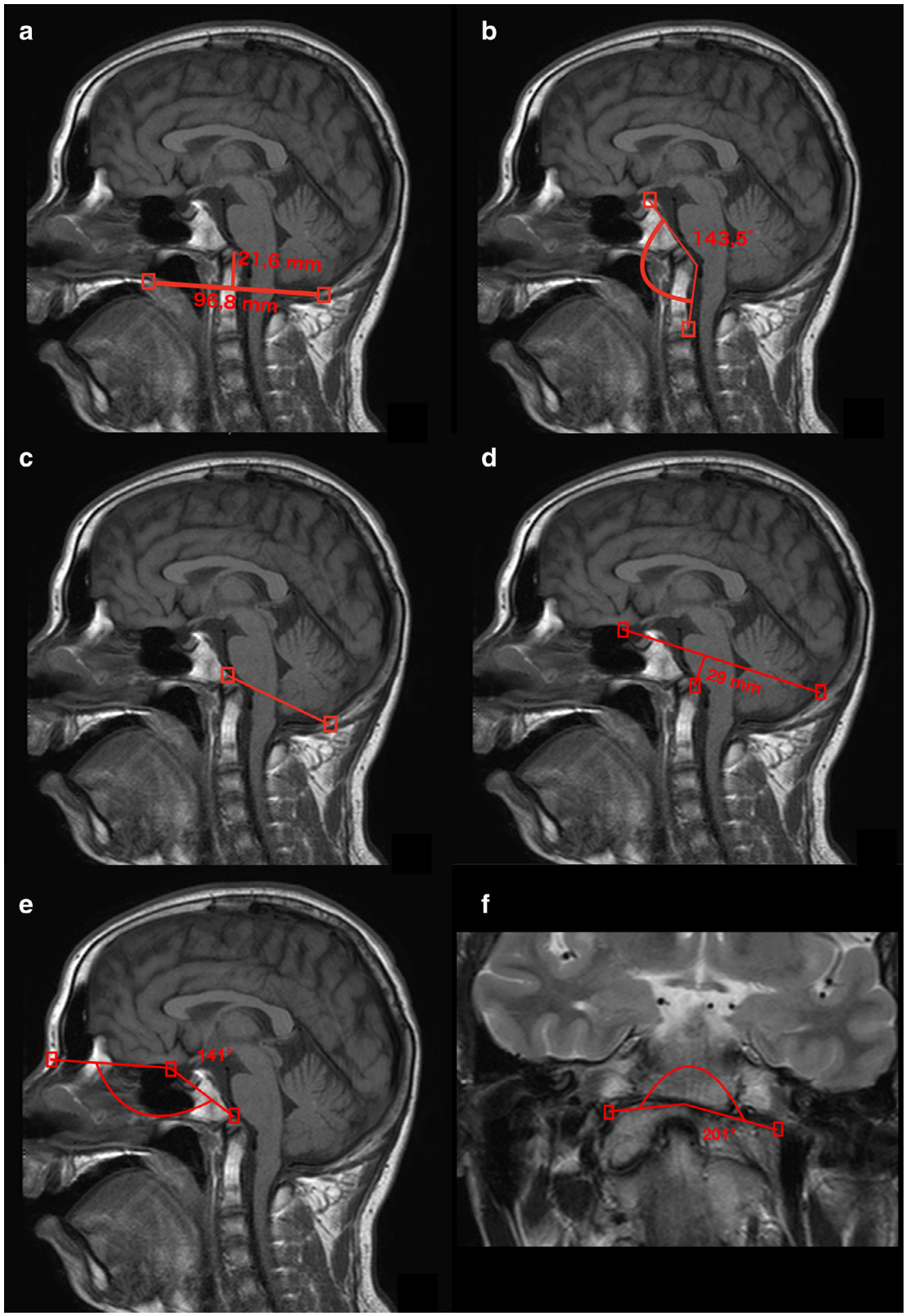

Fig. 2 – (a) Odontoid process overhangs of 21.6 mm the Chamberlain line (96.8 mm). (b) Wackenheim clivus canal angle (143.5°). (c) McRae line: the odontoid process lies below this line. (d) Height index of Klaus: 29 mm (partial basilar invagination). (e) Welcher basal angle: 141°. (f) Inverted atlantooccipital joint axis angle: 201°.

potential consequences, preventing or delaying aggravation of the pathology.

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