Case Report
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Atlantooccipital assimilation associated with combined atlas arch defect: a radiological case report

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Abstract: In this report, atlantooccipital assimilation (AS), anterior arch defect (AAD), and posterior arch defect (PAD) of the atlas, and several variations around the craniocervical junction were identified on computed tomography (CT) of a patient of unknown sex and age. Coronal and sagittal CT scans showed AS and bilateral fusion of the atlas and the base of occipital bone. Axial CT scan at the atlas revealed PAD type B on the left side and midline AAD. Morphometric measurements indicated a potential ventral spinal cord compression. In addition, mid-sagittal CT revealed the presence of fossa navicularis magna and incomplete formation of the transverse foramen on the right side. This study reports an extremely rare AS associated with AAD, PAD, and other variations of the clivus and the atlas. To our knowledge, no similar case has been reported in the literature.

Key words: Cervical vertebrae, Cervical atlas, Skull base, Klippel-Feil syndrome, Anatomic variation

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Introduction

Atlantooccipital assimilation (AS), also known as occipitalization of the atlas or atlas assimilation, is a rare anatomical variation characterized by partial or complete fusion of the atlas with the occipital bone. The prevalence of AS ranges from 0.12%–1.04% [1]. Partial assimilation refers to a condition where only a portion of the atlas has fused with the occipital bone, which can occur unilaterally or bilaterally. On the other hand, complete assimilation is when the entire atlas has become fused with the occipital bone, resulting in the absence of a distinct atlantooccipital joint. While both types of AS are generally asymptomatic, it may be associated with loss of mobility of the craniovertebral junction (CVJ) or neurological abnormalities in some cases [2]. Furthermore, AS associated with basilar invagination and spinal cord compression has been reported [3].

Arch defects of the atlas are well corticated gaps in one of the arches of the atlas. They are categorized into two types, posterior (PAD) and anterior (AAD) arch defects. In rare cases, the defect can be classified as a combined arch defect (CAD) or a bipartite or split atlas, when PAD and AAD are found together. Although most arch defects are asymptomatic and are found incidentally, they may be associated with atlantoaxial joint instability, and increased risk of craniovertebral or cervical injuries [4]. Other neurological condi-

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tions associated with PAD have been reported including myelopathy, Klippel-Feil syndrome, spinal stenosis, and Arnold–Chiari malformation [5]. These defects can also be confused with fractures, causing misdiagnosis [1]. The PAD is a well-known type of atlas arch defect with a prevalence from 0.2% to 5.2% across studies. The AAD and CAD, on the other hand, have received much less attention [1]. Descriptions are mostly restricted to case reports and there have been very few anatomical studies [1].

In this case report, we present a radiological case of AS associated with AAD and PAD. To our understanding, such configuration is extremely rare has never been reported. Embryological and clinical implications of the present case are discussed.

Case Report

This study was approved by Khon Kaen University Ethics Committees for Human Research (approval number: HE661212). Multiple defects and variations of the atlas and CVJ were discovered accidentally in a computed tomography (CT) scan of a patient with unknown sex and age during investigation of the anatomical variants of the clivus using an open-access CQ500 CT dataset. The dataset was downloaded from http://headctstudy.qure.ai/dataset, containing non-contrast CT scans of patients with calvarial fracture with or without intracranial hemorrhage taken at Advanced Research in Imaging, Neurosciences and Genomics, New Delhi, India [6].

Sagittal and coronal (Fig. 1A, B) scan and three-dimensional reconstruction (Fig. 2) showed complete and bilateral fusion of the atlas and the base of occipital bone. Axial CT scan at C1 vertebral level revealed midline AAD (Fig. 1C) and type B PAD on the left side (Fig. 1D). Anterior atlantodental interval (AADI), horizontal distance between the posterior cortex of the anterior arch of the atlas and the anterior border of the dens, was measured (Fig. 1E). The yielded AADI value was 3 cm. The odontoid process was normal. Axial CT scan of the atlas showed a midline AAD along with a PAD located posterior to the occipital condyle on the right

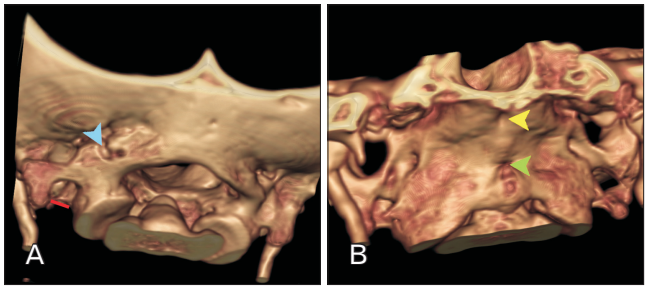

Fig. 2. Three-dimensional reconstruction from the posterior (A) and anterior (B) views of the craniocervical junction showing posterior arch defect (blue arrowhead), fossa navicularis magna (yellow arrowhead), incomplete transverse foramen (red line) and anterior arch defect (green arrow).

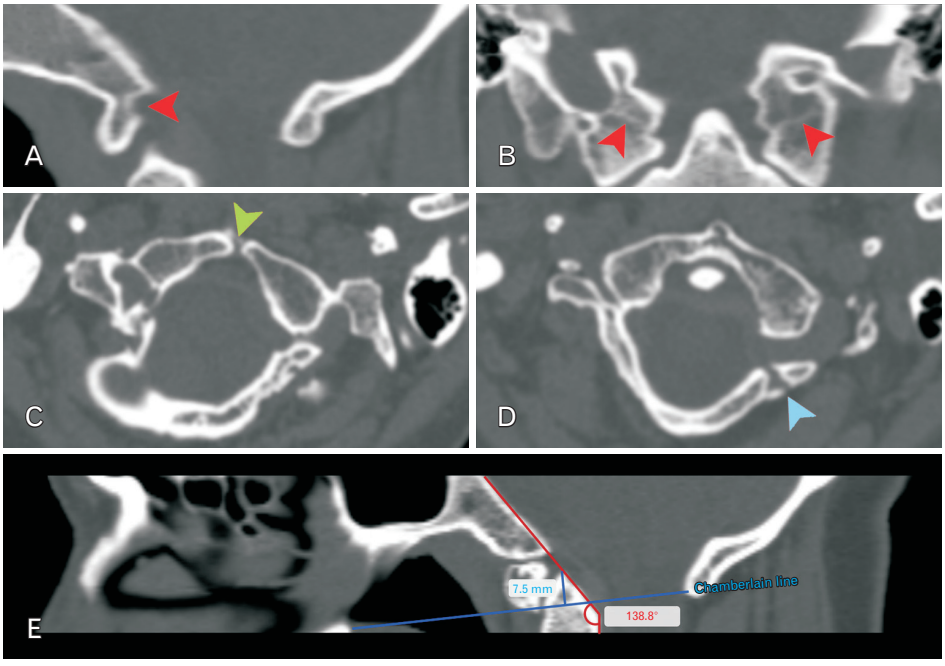

Fig. 1. Sagittal (A), coronal (B), axial (C, D) computed tomography scans showing the presence of atlas assimilation (red arrowheads), anterior atlas arch defect (green arrowhead) and posterior arch defect (blue arrowhead). Sagittal image (E) shows the distance between the odontoid apex and Chamberlain's line, lining between the hard palate and the opisthion (7.5 mm), suggesting basilar invagination into the posterior cranial cavity of skull. The angle between clivus and posterior aspect of odontoid process was 138.8°, indicating predisposition to anterior spinal cord compression.

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side. Sagittal CT also showed that the distance between the odontoid apex and Chamberlain's line (DOCL) was approximately 7.5 mm above Chamberlain's line, between the hard palate and the opisthion (Fig. 1E). Moreover, the clivus canal angle (CCA) or the angle between clivus and posterior aspect of odontoid process was 138.8° (Fig. 1E).

In addition to the arch defects found and measurements made, axial CT at C1 vertebral level also showed incomplete formation of the transverse foramen (ITF) on the right side (Fig. 2A). Fossa navicularis magna (FNM), a variant bony impression at the pharyngeal surface of the clivus, was also present (Fig. 2B).

Discussion

To our knowledge, this is the first study to report an AS associated with AAD and PAD of the atlas. AS is an extremely rare finding with a prevalence of 0.12%–1.04% across studies [7, 8]. While the PAD is a well-recognized and relatively common defect of the atlas, the CAD is extremely rare and only has an overall prevalence of 0.14% [1]. Classification of the AAD and CAD has recently been proposed by Suphamungmee et al. [1] based on 57 cases of AAD and CAD identified in the literature. The most prevalent type of a midline AAD is classified as type A. Type B, the second most common type, is used to describe any AAD that is linked to a PAD. Type C and type D are uncommon and are characterized as unilateral or bilateral AAD, respectively. Type E is used when there is a defect in the anterior arch in conjunction with the absence of the posterior arch. The CAD in the present study was classified as type B, which was found in 35 out of 57 cases of CAD reported in the literature [1].

In the case of AS, there is a failure of segmentation or separation between the atlas and occipital bone during development. As a result of the failed separation, the atlas fuses with the occipital bone, leading to the AS. The atlas, undergoes a series of ossification. In most individuals, there are three primary centers of ossification involved. The anterior center forms the anterior tubercle, while the two lateral centers give rise to the lateral masses and the posterior arch [9]. However, if the fusion between these two masses fails, a PAD can occur. In rare cases, a fourth ossification center may be present at the posterior midline [9], and the failure of fusion between this fourth center and the two lateral centers can give rise to type B and type C PAD, as classified by Currarino et al. [10]. The failure of fusion among these ossification centers can be linked to various forms of atlas arch defects.

CVJ craniometry is a useful tool for identifying congenital abnormalities involving the occiput, atlas, and axis. It is widely recognized that when the tip of the dens protrudes more than 5 mm above Chamberlain's line, it can result in compression of the medulla [11]. In the present case, we found that the DOCL distance was 7.5 mm above the Chamberlain's line, indicating that the medulla may be subject to compression. According to Smoker and Khanna [11], the CCA is a measurement used to assess ventral spinal cord compression. In a normal range, the CCA typically ranges between 150° in the flexion position and 180° during neck extension [11]. In this specific case, the CCA measured 139°, indicating a potential predisposition to ventral spinal cord compression. Previous reports have linked basilar invagination and spinal cord compression to AS [3]. Another important measurement on the sagittal plane is the AADI, which assesses atlantoaxial subluxation or dislocation. On radiographs, the AADI should ideally be less than 3 mm to indicate normal alignment [12]. In this particular case, the AADI measured less than 3 mm, suggesting the absence of atlantoaxial instability or subluxation. Furthermore, the simultaneous presence of AS, AAD, and PAD may not occur in isolation but can be part of a syndrome or a spectrum of abnormalities. The reduction in bone volume due to the presence of AAD together with a PAD may reduce the structural integrity of the C1 vertebra. Above all, it is crucial to note that these possibilities remain speculative, as clinical confirmation is not feasible. Upon conducting further literature reviews, we discovered that GDF6 is expressed within the developing vertebral interspace. Settle et al. [13] provided evidence of GDF6's role in the fusion of vertebrae. Interestingly, mutations in GDF6 have also been linked to vertebral segmentation defects in Klippel-Feil syndrome [14], a condition that presents similarly to what is observed in the present case.

Two additional anatomical variations were also observed including FNM and ITF. These variations are asymptomatic and not usually associated with any significant clinical symptoms or functional impairments. The FNM is an impression located on the pharyngeal surface of the clivus found in 0.9%–7.6% as reported by Bayrak et al. [15]. Two theories have been proposed to explain the origin of this variant. The first suggests that it is a remnant of the notochord, while the second theory proposes that the FNM is a residue of an opening related to emissary veins [15]. The ITF

is described as a gap in the antero-lateral forming a groove for the vertebral vessels found in around 7% of individuals [16]. It was identified on the right side of the subject in the present study (Fig. 2). Radiologists should recognize ITF to avoid confusion with fractures and other anomalies during the interpretation of radiographs and CT scans.

The present study reported a concomitant occurrence of AS, CAD, and multiple variations at the craniocervical junctions. To our knowledge, this case is extremely rare and no similar case has not been reported previously.

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Conceptualization: AS, TS, LY. Data acquisition: PT, AS, TS. Data analysis or interpretation: PT, WK, NK, KT, KR. Drafting of the manuscript: AS, TS. Critical revision of the manuscript: PT, NK. Approval of the final version of the manuscript: all authors.

Conflicts of Interest

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

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