Innoscience Journal
ISSN 3005-8953
Open Access

Case Reports

Simultaneous Congenital Fusion Defect of Anterior and Posterior Arch of Atlas (C1): Case Report.

Pánuco-Romero, Juan de Dios*[1]; Pérez-Espinoza, Cristo Jassiel[1]; Aragon-Argote, Cindy Jaqueline[1]; Pánuco-Romero, Iris Xiomara[1].

*Correspondence: juandediosr7@icloud.com | ORCID: 0009-0005-0044-2030

[1]Neurosurgery Department, Regional Hospital "Manuel Cárdenas de la Vega" ISSSTE, Culiacán, Sinaloa, México.

Keywords: Atlas Posterior Arch, Spine abnormalities, Atlas anomalies, C1 hypoplasia, Craniocervical junction, Atlas Anterior Arch, Case Report.

Abstract: The atlas (C1), the most important cervical vertebra, occupies a crucial anatomical and functional position. It is distinguished by its unique structure, composed of anterior and posterior arches and two lateral masses, each with a transverse foramen for the passage of the vertebral artery at its segmental transition. Congenital anomalies of the posterior arch of the atlas are rare (0.69–4%) and can range from clefts to hypoplasia or aplasia. The combination of simultaneous anterior and posterior arch defects is rarely described in the literature. This case exemplifies the need to differentiate congenital malformations of the atlas from acquired pathologies, as well as to evaluate for dynamic compromise of the craniocervical junction and rule out spinal cord and/or upper cervical nerve root involvement.

Cite as: Pánuco-Romero JD, Pérez-Espinoza CJ, Aragon-Argote CJ, Pánuco-Romero IX. Simultaneous Congenital Fusion Defect Of Anterior And Posterior Arch Of Atlas (C1). Case Report. Innoscience Journal. 2025 Jul 10;3(1):6–9. DOI: 10.17613/0yg7q-9yj59

Published: March 28, 2026
Funding: This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors.
Conflicts of Interest: The authors declare that this manuscript was approved by all authors in its current form and that there are no conflicts of interest.

INTRODUCTION

The craniovertebral junction (CVJ) comprises the occiput, atlas (C1), axis (C2), and their supporting ligaments, along with soft tissue structures including 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 anomalies of the posterior arch of the atlas are rare, occurring in approximately 0.69–4% of the population, and may vary from clefts to hypoplasia or aplasia. According to the Currarino Classification (types A–E), Type A (midline cleft) is most prevalent, accounting for approximately 90% of congenital posterior arch defects (Fig. 1).

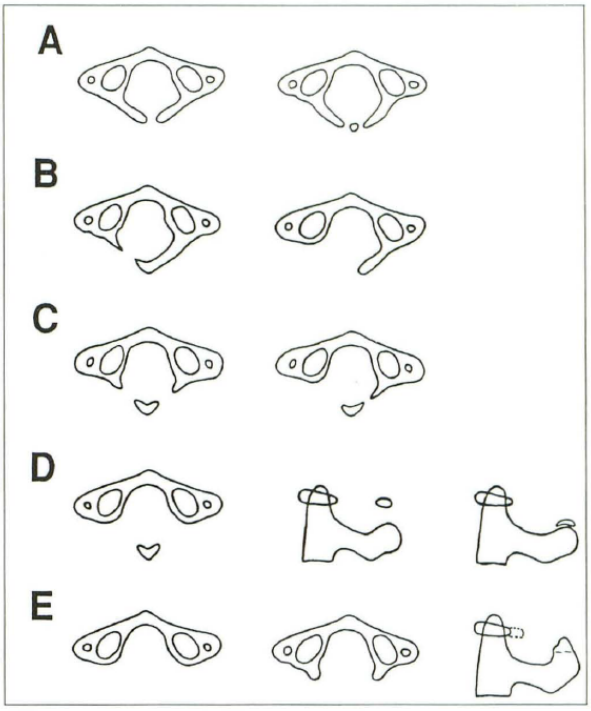

Figure 1. Currarino Classification of congenital anomalies of the posterior arch of the atlas (types A–E). Type A: midline cleft of both ossification centers; Type B: unilateral defect; Type C: bilateral clefts with preserved dorsal aspect; Type D: absence of posterior arch with unattached tubercle; Type E: complete absence of posterior arch and tubercle.

Anterior C1 fusion defects are substantially less common than posterior defects; these clefts typically occur at or just off the midline. The combination of simultaneous anterior and posterior arch defects is particularly uncommon, with few cases reported in the medical literature. Many patients are misdiagnosed, as the problem often goes undetected until imaging is performed following trauma or other causes of neck pain. This case report presents a patient with C1 fusion defects without myelopathy or radicular compromise, and discusses the clinical significance of this anatomical variant.

CASE REPORT

A 34-year-old woman presented to the neurosurgery clinic referred by physical medicine and rehabilitation with a chief complaint of chronic neck pain and mechanical low back pain. She had no history of trauma and denied weakness in any extremities or evidence of pyramidal tract involvement.

Clinical Examination: General and cranial neurological examination was normal. Cervical mobility was unrestricted with no clinical worsening on flexion or extension. Palpation revealed tenderness in the upper trapezius muscle with ++/++ spasm. Manual muscle testing revealed intact strength: C5 5/5, C6 5/5, C7 5/5, C8 5/5, and T1 5/5. Sensory examination was normal across dermatomes. Lower extremity examination demonstrated full range of motion without pain.

Imaging Findings: Computed tomography and magnetic resonance imaging revealed complex alterations of the CVJ characterized by anterior arch fusion defect of C1 (Fig. 2), hypoplasia of the posterior arch of C1 with compensatory hypertrophy of the craniocervical ligament complex (Fig. 3), and hyperostosis of the C2 spinous process with changes in the ligamentary complex (Fig. 4).

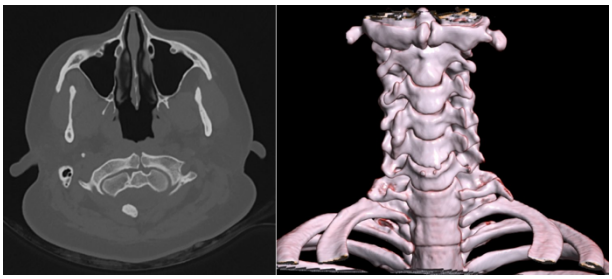

Fig. 2. Axial bone CT and 3D reconstruction images showing a anterior fusión defect of Atlas.

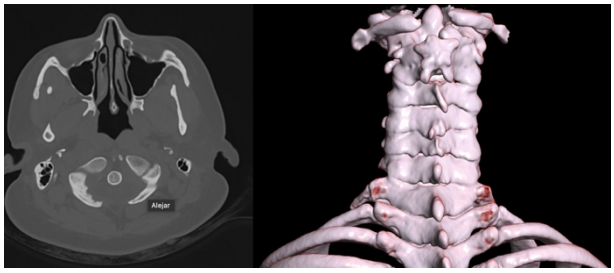

Fig. 3. Axial bone CT and 3D reconstruction images demostrate a posterior arch midline hipoplasia, and spinous process of C2 compensatory hyperostosis.

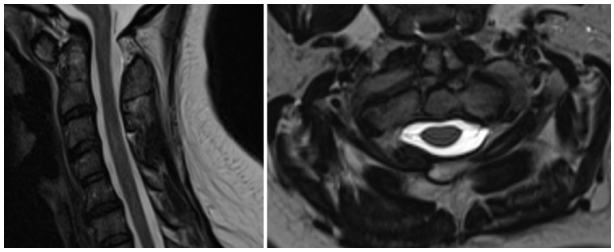

Fig. 4. Midline Sagital, and Axial T2 MRI demostrates craniocervical and atlantoaxial ligamentary complex hypertrophy.

Craniometric Assessment:
• Anterior Atlantal Dental Interval (AADI): 2.4 mm (normal; <3.0 mm indicates instability)
• McRae Line: The dens did not cross the line (normal)
• Clark Station: II (medium, indicating normal spinal canal width)
• Atlantoaxial Interval: 3.9 mm (normal stability)

DISCUSSION

Embryologically, the atlas develops from three primary ossification centers. The anterior arch does not develop until after birth, with ossification centers typically visible in the first year of life. Fusion of the anterior arch occurs between ages 3 and 5 years, with complete fusion possible up to age 9 years. The posterior arch develops from two lateral centers that extend posteriorly around week 7 of gestation. In approximately 2% of the population, a fourth ossification center forms the posterior tubercle by age 2 years.

The majority of congenital C1 anomalies are asymptomatic and discovered incidentally on imaging. Some are associated with syndromic conditions, including Down syndrome and Klippel-Feil syndrome. Congenital posterior arch defects are more frequently documented than anterior arch defects, with rare instances of combined anterior and posterior defects.

Our patient exemplifies Type A classification according to Currarino (midline cleft of the posterior arch). However, the presence of a simultaneous anterior arch fusion defect is notable, as current classification systems do not fully account for this combination. Recent work by Baena-Caldas et al. (2024) proposed an expanded classification system introducing Types F and G to encompass anterior arch defects and their combinations with posterior defects (Fig. 5). Our case would fall under Type G-1 (combined midline defects in both anterior and posterior arches, or "bipartite atlas").

Absence or hypoplasia of the posterior arch frequently results in compensatory hypertrophy of the anterior arch of C1 and the spinous process of C2, a pattern observed in this patient. Dynamic compromise of the craniocervical junction occurs in a minority of cases and may present with myelopathy, radiculopathy, or cervical degenerative disc disease. The absence of symptoms or neurological deficit in our patient despite these anatomical abnormalities underscores the importance of comprehensive craniometric assessment to evaluate stability and potential cord compression.

This case emphasizes the need to differentiate congenital malformations of the atlas from traumatic fractures, which may present with similar imaging appearances. Traumatic history must be carefully assessed. Close clinical follow-up is essential to identify any emergence of symptoms, which would guide decisions

regarding conservative management or surgical intervention.

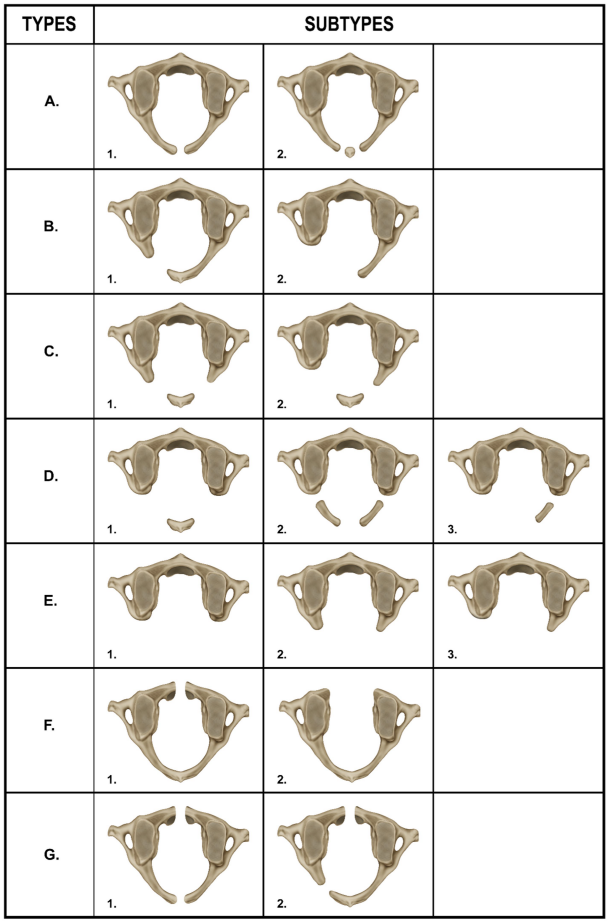

Fig 5. Curricano Classification modified by Baena-Caldas et. al. (2024)

CONCLUSIONS

Structural abnormalities of the spine can occur at multiple levels with varied imaging and clinical manifestations. This case demonstrates a rare combination of anterior and posterior arch fusion defects of the atlas. Careful differential diagnosis is essential to exclude traumatic etiologies. Comprehensive craniometric evaluation is necessary to assess stability and rule out spinal cord compression. Close clinical follow-up is warranted to monitor for symptom emergence and guide subsequent management decisions.

DECLARATIONS

This case report was conducted in accordance with the principles of the Declaration of Helsinki. Written informed consent was obtained from the patient for publication of this case report and accompanying medical images. The patient was informed that their identity would remain confidential and that the case would be used for academic and scientific purposes only.

The authors declare that they have no competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

This research received no specific grant from any public, commercial, or not-for-profit funding agency.

AUTHOR CONTRIBUTIONS

Pánuco Romero JD: Case presentation, literature review, manuscript preparation. Pérez Espinoza CJ: Case management, imaging analysis, clinical follow-up. Pánuco Romero IX: Case coordination, clinical evaluation. Aragón Argote CJ: Clinical contribution, manuscript review. All authors read and approved the final manuscript.

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