

A report of two cases of familial occipitalization of the atlas in a father and his daughter

ABSTRACT

Occipitalization of the atlas (OA) (atlantooccipital assimilation) is a rare congenital anomaly that results in fusion of the C0-C1 complex. Here, we report the two cases of familial OA occurring in a father and his daughter and describe the pathologic features of this rare developmental anomaly. A 70-year-old father and his 30-year-old daughter seeking conservative care for musculoskeletal complaints presented to a chiropractic clinic. Radiographs of the cervical spine demonstrated OA. This represents a novel example of this anomaly in two patients with a direct familial relationship.

Keywords: Assimilation of the atlas, atlantooccipital assimilation, congenital anomalies, occipitalization

INTRODUCTION

Occipitalization of the atlas (OA) (also referred to as assimilation of the atlas, atlantooccipital fusion, occipitocervical synostosis, or atlantooccipital nonsegmentation/assimilation) is a congenital osseous/fibrous failure of segmentation, resulting in complete or partial fusion (partial fusion being less common) of the occiput-atlas articulation (C0-C1 complex).^[1-5] OA is a rare congenital anomaly of the craniocervical junction (CCJ), occurring in 0.08%–2.76% of persons, with a global incidence of 3.63%.^[6] OA affects males and females equally.^[4,5,7-11] OA is not always an isolated anomaly; additional associated anomalies commonly found are hypoplasia of the basiocciput, basilar invagination, occipital vertebra, spondyloschisis of the atlas, and vertebral synostosis (block vertebrae).^[1,11]

In addition, OA has been reported with various syndromes such as Goldenhar, Klippel–Feil, and Pierre–Robin, as well as with craniosynostosis, platybasia with clivus reduction, and atlantoaxial dislocation.^[1-3] Although usually asymptomatic, patients with OA may present with headaches, neck pain, limb paresthesia/pain, weakness, tinnitus, disrupted vision, and lower cranial nerve palsies.

CASE REPORT

Two patients initially presented with complaints unrelated to the cervical spine. A 70-year-old father seeking maintenance care regarding a chronic low back complaint and his 30-year-old daughter who began treatment in 2007 for cervical and thoracic spine pain presented. Following a motor vehicle collision in May 2019, the daughter had cervical radiographs to assess for traumatic injury. The radiographs revealed congenital OA [Figure 1]. The daughter initially presented with cervicothoracic pain and headaches before the accident. The daughter’s primary complaint after the motor vehicle collision was regarding increased cervical and thoracic pain. The daughter also reported a secondary complaint of headaches, not exacerbated by the accident.

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The father was receiving treatment for low back pain, with no mention of cervical complaints during the 30 years of care. Due to the father's age, a full spine radiographic series was performed during a reexamination in 2014 to rule out contraindications to chiropractic manipulative therapy. Retrospective review of her father's radiographs obtained in 2014 revealed that he also had occipitalization [Figure 2].

The patients were informed of their anomalies and the clinical significance. Neither patient exhibited severe neurologic manifestations. Informed consent was provided by patients to carefully observe for manifestations and to have flexion–extension radiographs and/or computed tomography/magnetic resonance imaging examinations in the future, if warranted.

Recent flexion–extension radiographs of the daughter were obtained in 2020, which demonstrated complete lack of joint space between the posterior arch of the atlas and the occiput. The atlantodental interval (ADI) measures within normal limits on both flexion and extension views. The ADI findings in conjunction with the absence of atlantooccipital joint space, is indicative of occipitalization of the atlas. Radiographs were negative for any evidence of spinal instability [Figure 3].

The development of the CCJ is a complex process that can give rise to various congenital and acquired malformations.^[2] The atlas develops from three primary ossification centers: (1) each for the anterior and posterior arch and one for each of the lateral masses.^[2,12] The development of the atlas involves the fourth occipital and first cervical sclerotomes. It is theorized that OA results from failed segmentation of these sclerotomes.^[2,3,6,10,13] Congenital defects/anomalies of the branchial (pharyngeal) arch commonly accompany OA because they develop simultaneously with sclerotome segmentation.^[2] These anomalies include nonunion of ossification centers leading to anomalous ossicle development, resulting in interparietal (or inca) bones and preinterparietal bones.^[12] Clinically, these ossicles may be mistaken for skull fractures and can complicate surgical interventions.^[12]

Because OA is often asymptomatic, OA and its related anomalies tend to be clinically undetected, and as such, the anomalies are usually discovered incidentally on radiographs.^[2,3,5,8,9] Many associated conditions such as basilar invagination, atlantodental dislocation, and foramen magnum stenosis can result in cervicomedullary compression, resulting in neurologic impairment (via compression of the spinal cord, tonsillar herniation, or syringomyelia).^[2,6-10] In some cases, the brainstem compression can become so severe, to the point where surgical intervention is needed.^[3,7] Often, the onset of neurologic



Figure 1: Lateral radiograph of a 70-year-old male which displays complete assimilation of the atlas (along with an articulation between the fused segment and the posterior arch of C1)

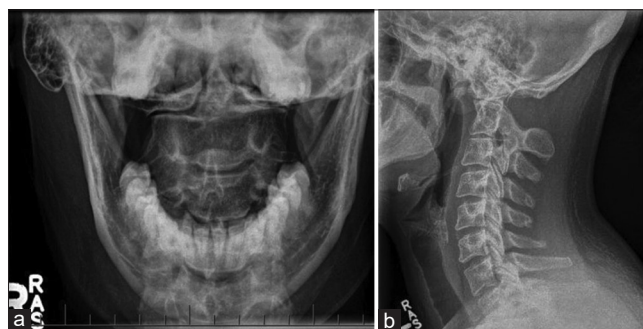


Figure 2: (a) An APOM radiograph of a 30-year-old female presenting with complete atlantooccipital assimilation of the atlas (lack of visible joint space between the C0-C1 junction). (b) A lateral radiograph of a 30-year-old female demonstrating complete fusion of the posterior arch of the atlas, and the base of the occiput

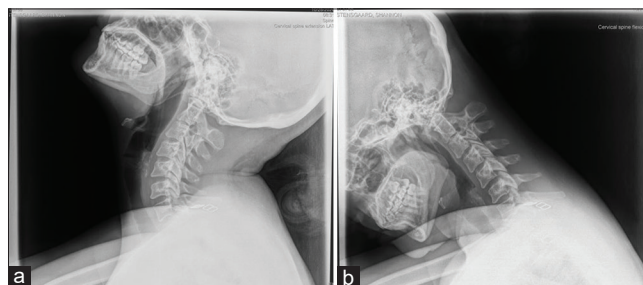


Figure 3: (a) Lateral extension radiograph of a 30-year-old female which displays complete assimilation of the atlas. (b) Lateral flexion radiograph of a 30-year-old female which displays complete assimilation of the atlas. The atlantodental interspace measures within normal limits on both flexion and extension views

symptoms will not present until later in life, typically occurring first during the 2nd–4th decades.^[1,3,4,11,13] These neurologic deficits may initially manifest as transitory headaches, progressing to more significant neurological syndromes (and occipitocervical instability).^[4,11,13] Patients with OA may also present with specific physical findings such as low hairline, torticollis, cervical motion restrictions, and decreased overall neck height.^[3] An important

complication of OA is reduction in transverse and sagittal diameter of the foramen magnum.^[2]

Radiographic features include loss of visible joint space between the occipital condyles and C1, basilar invagination, and synostosis. OA can alter joint biomechanics, varying the force required to fracture the assimilated regions, resulting in instability.^[14] Patients with associated synostosis of C2–C3 commonly present with instability at other levels in the cervical spine, and as such should have flexion–extension views to determine the stability of the cervical region before conservative treatment.^[15]

The surgical management of OA and its sequela (neurological deficits, and segmental instability) include arthrodesis (fusion) via screw–rod instrumentation for instability.^[6-8] Moreover, surgical management with or without decompression surgery is determined on a case-by-case basis.^[6,7]

CONCLUSION

To our knowledge, the occurrence of familial OA involving two direct family members has never been reported. This may suggest a genetic/hereditary component to the condition or may just be due to chance. Further research is needed to be certain.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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