

Congenital Hypoplasia of the Posterior Arch of the Atlas: Case Report and Extensive Review of the Literature

Atlasın Arka Yayının Doğumsal Gelişim Bozukluğu: Olgu Sunumu ve Literatürün Geniş Gözden Geçirimi

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ABSTRACT

Congenital hypoplasia of the posterior arch of the atlas (C1), a developmental failure of chondrogenesis, is a rare anomaly and may range from partial clefts to total agenesis of the posterior arch. Ossification of the posterior arch usually occurs between the 3rd and 5th years of life. The incidence of posterior arch anomalies of the atlas is between 0.69% and 2.95%. For the evaluation of the patient, cervical lateral plain radiography, 2D or 3D reconstructed CT and MRI are very useful and important tools in initial diagnosis. Surgery is the treatment of choice in symptomatic compression. Excision of the posterior arch is performed during surgery. After the surgery, patients may be followed up for instability and treated as necessary. A patient, admitted to the emergency department with head and neck trauma after a traffic accident is presented in this article. C1 hypoplasia was determined after detailed imagining studies and the radiology department consulted. When upper cervical anomalies are found in a young patient, the patient should be evaluated in detail with advanced radiological studies to avoid misinterpretation as fractures, luxation, osteolysis or instability. Consulting a radiologist could help making an accurate diagnosis and deciding on current therapeutic interventions.

KEYWORDS: Atlas, Hypoplasia, Posterior, Arch, Congenital, Anomaly

ÖZ

Kondrojenetikteki doğumsal bozukluk sonucu oluşan atlasın arka yayının gelişimsel eksikliği nadir bir anomalidir ve kısmi yarı oluşumundan atlasın arka yayının tam yokluğuna kadar gidebilir. Arka yayın kemikleşmesi 3-5 yaşlar arasında olur. Atlasın arka yayı anomalilerinin insidansı %0.69-2.95 arasındadır. Hastayı değerlendirmek için servikal lateral direkt grafi, 2 ve 3 boyutlu rekonstrüksiyonlu bilgisayarlı tomografi ve magnetik rezonans inceleme çok yararlı ve önemli tanı araçlarıdır. Semptomatik bası varlığında cerrahi bir tedavi seçeneğidir. Cerrahide arka yayın eksizyonu uygulanmaktadır. Cerrahi sonrasında hastalar instabilite için takip edilmeli ve gerektiğinde tedavi edilmelidir. Bu makalede, trafik kazası sonrası acil servise baş ve boyun travması ile gelen bir hasta sunulmaktadır. Detaylı radyolojik çalışmalar sonrası, atlas hipoplazisi saptanmış ve sonrasında hasta radyoloji bölümüne konsülte edilmiştir. Genç hastada üst servikal anomaliler saptandığında; kırık, luksasyon, osteoliz ve instabilite hasta ayrıntılı yanlış yorumlardan kaçınmak için hasta radyolojik incelemelerle ayrıntılı değerlendirilmelidir. Radyoloji uzmanına yapılan konsültasyon kesin tanıya ve doğru tedavi edici girişimlerin yapılması için karar verilmesine yardım olmaktadır.

ANAHTAR SÖZCÜKLER: Atlas, Hipoplazi, Arka, Yay, Konjenital, Anomali

INTRODUCTION

Congenital hypoplasia of the posterior arch of the atlas (C1), a developmental failure of chondrogenesis, is a rare anomaly and may range from partial clefts to total agenesis of the posterior arch (12,21). These abnormalities were considered by some to be a "benign variation" and indeed, almost all of them are discovered incidentally (3,4,11,13). In general, such abnormalities are asymptomatic but neurological symptoms may occur after minor cervical trauma in patients with isolated partial agenesis of the posterior arch of the C1 (2,12,13,28). Clinical features vary from mild neck pain to neurological

deficits occurring after minor cervical spine or head injury (29,31).

We report a patient with hypoplasia of the posterior arch of the C1 who was admitted to our emergency department for head trauma without any neurological deficit.

CASE REPORT

A previously healthy, right handed, 26-year-old male patient was admitted to our emergency department with head trauma after traffic accident. The patient suffered from headache and neck pain. The general and cranial neurological examination

was unremarkable. There were no motor or sensory deficits. All reflexes were normal and plantar reflexes were downgoing. No clinical deterioration was found with neck flexion and extension. All the laboratory tests were normal. Lateral cervical spine radiograph revealed two hemiarches of C1 with a hypoplasia in the midline (Figure 1). Cervical 2D and 3D reconstructed computed tomography (CT) studies showed hypoplasia of the posterior arch of the C1 (Figure 2, 3A-B, 4). The patient was hospitalized for a night. A magnetic resonance

imaging (MRI) study was performed the following day and it also revealed the partial absence of the posterior arch of the C1 (Figure 5). MRI study did not show any neural alteration or other associated abnormalities, such as protrusion of disc material, compression of spinal cord, basilar invagination, Chiari malformation, a syringomyelic cavity or narrowing of the sagittal diameter at the level of C1 (Figure 6). The patient was managed conservatively with medical therapy and a cervical collar and his neck pain subsided after a week. He



Figure 1: Lateral cervical spine radiograph shows two posterior hemiarch of C1 (arrows).



Figure 2: Axial CT image reveals hypoplasia of C1 (arrow).

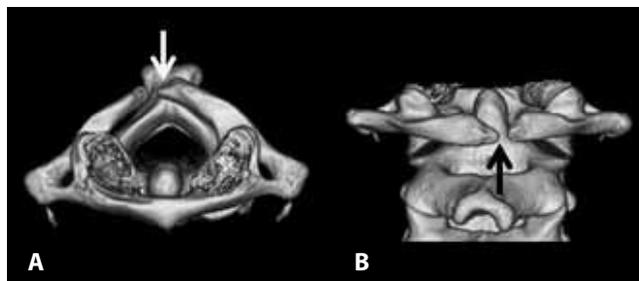


Figure 3: Three-dimensional reconstructed CT image of C1: **A)** Upper **B)** Posterior view shows hypoplasia of C1. White arrow in Fig 3A and yellow arrow in Fig 3B.

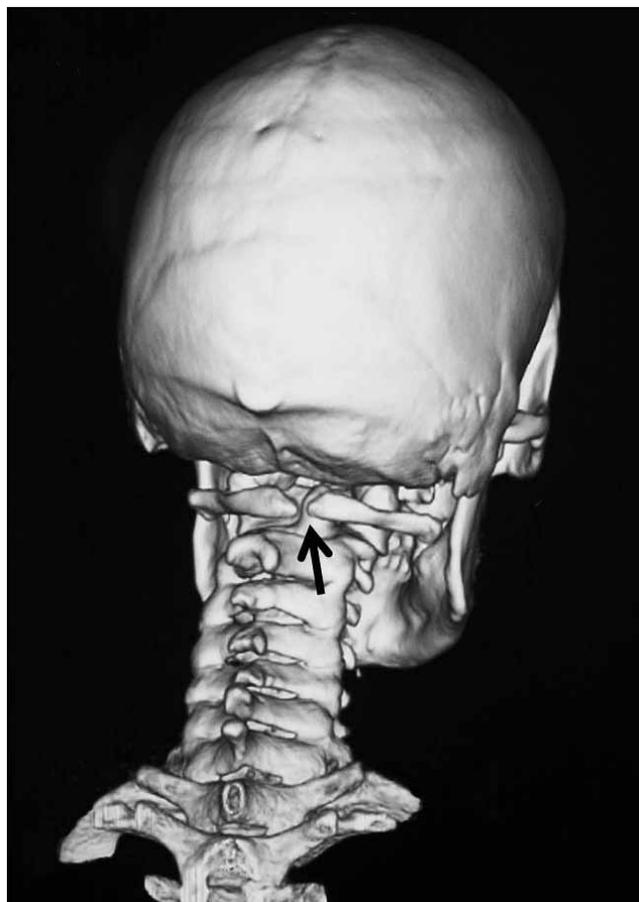


Figure 4: Three-dimensional reconstructed CT image of the C1 right posterolateral view. Note the hypoplasia of C1 (black arrow).



Figure 5: Axial T2-weighted MR image. The hypoplasia (black arrow) and two posterior hemiarch (white arrows) of C1 is hardly seen on the MR image. The spinal canal and cord appears normal.

remained asymptomatic when he was last examined a year later.

DISCUSSION

Understanding the embryology of the first and second vertebrae is the key to understanding developmental anomalies of the upper spine. The C1 derives from the primitive fourth occipital and first cervical sclerotomes. It has three primary ossification centers in the embryological period: an anterior center for formation of the anterior tubercle and two lateral centers from which the lateral masses and posterior arch form (23). In 7th week of gestation, the lateral centers extend dorsally to form the posterior arch (Figure 7). An additional center may be present posteriorly in the midline, forming the posterior tubercle of the C1 in second year of life in about 2% of the population (3,6,7,15,17,27,29). During maturation, the anterior arch of the C1 derives from usually one midline, but occasionally two or more ossification centers are seen radiologically in the first year of life. Ossification of the posterior arch usually proceeds perichondrally from two centers in the lateral masses, towards the midline, and fusion occurs between the 3rd and 5th years of life; incomplete fusion of the two hemiarches may be normal in the first 5-10 years (9). However, incomplete fusion may persist in 3 to 5% of patients. The anterior center usually fuses with the two lateral centers between 5 and 9 years age (23). In rare cases, a separate ossification center forms in the midline and fuses secondarily with the lateral masses forming the posterior arch; a posterior midline ossified tubercle develops if this center fails to fuse with the hemiarches (9). At least two different anomalies can develop during the ossification: 1) median clefts of the posterior arch of C1, and 2) varying degrees of posterior arch dysplasia (6,17,29).



Figure 6: Sagittal T2-weighted MR image. There is no spinal cord compression.

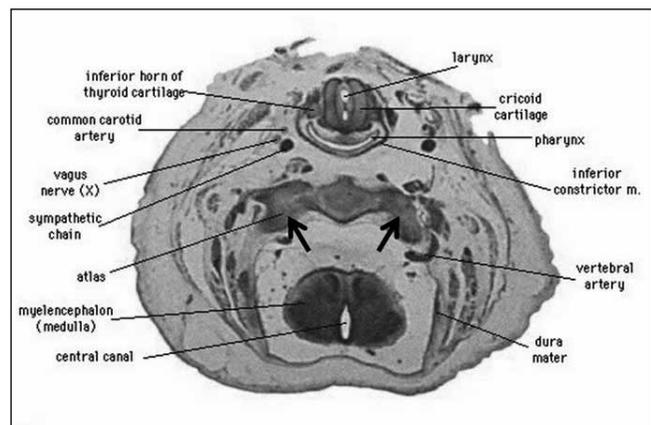


Figure 7: The lateral ossification centers of atlas vertebrae (arrows) extend dorsally to form the posterior arch in an 8-week-old human embryo. These centers fuse on the midline posteriorly to form the posterior arch of atlas (with the permission of M. Hill, Ph.D.: <http://embryology.med.unsw.edu.au/www/human/lowpower/HumC/C3L.htm>)

Different theories have been proposed to explain congenital defects of the C1 posterior arch (2,11,18,22,23,24,31). However, the exact mechanism remains obscure. Congenital absence or hypoplasia of the posterior arch of the C1 may be associated with several disorders, such as the Arnold Chiari malformation, gonadal dysgenesis and the Klippel-Feil, Down and Turner syndromes respectively (3,15,16,29). Martich et al. found hypoplasia of the posterior arch in 26% of 38 children with Down syndrome. If hypoplasia of the C1 is found in a young patient, the authors recommend ruling out Down syndrome, gonadal dysgenesis, achondroplasia or Turner syndrome (4,10,14,29,30). It is commonly accepted that defects of the posterior arch of the C1 are due to a lack of chondrification rather than a primary disturbance of ossification (2,9).

The reported incidence in a large study of 1613 autopsies with regard to presence of congenital aplasia is 4% for the posterior arch and 0.1% for the anterior arch (16). It has been reported that hypoplasia of the posterior arch of C1 may increase the risk of atlantoaxial subluxation in about 26% of children aged 2-3 years (14,16). Patient presented in the literature are predominantly children or women in the second or third decade of life (1,16,24). In Martich's study, clefts of the posterior arch were found in 45 of 1613 cadaveric dissections, 97% of which were median clefts (14). Şenoğlu et al. reported their findings which are obtained from 1104 patients by cervical spine CT scans: 37 patients had posterior arch defects, and of these, 29 (2.6%) had median clefts. Therefore, type A defects represented 78.4% of all posterior arch defects. Furthermore, they determined such defects in 2 (1.2%) of 166 dried C1 specimens and 1 (1.19%) of 84 fresh human cervical segments (23). The exact incidence of posterior arch anomalies of C1 is unknown but according to Currarino's study, the incidence is 0.69% (2). In Şenoğlu's study, the overall incidence (CT scan + dried specimens + fresh specimens) of the posterior atlantal arch in 1354 evaluated cases is 2.95% (40 cases). On the other hand, the anterior arch defects are even less common than posterior and the rate is reported as 0.1% in several studies (7,10,14,23,29).

A complete classification of congenital anomalies of the posterior arch of the C1 is proposed by Currarino et al (2). Plaut and Lawrence & Anderson first published an article about a developmental abnormality of C1 in 1937. A total of 41 articles have been published about posterior defects of C1 in the literature so far (Table I). In 1975, Richardson et al. were the first to propose a mechanism by which neurological deficits occur. They theorized that the isolated posterior fragment moved anteriorly and traumatized the dorsal spinal cord when, with extension, the distance between the occiput and the spinous process of the axis caused inward buckling of the ligaments (11,20). According to the classification of Currarino et al. there are five types of anomalies (Figure 8) (Table II): Type A defects are defined as failure of the posterior midline with a small gap remaining (that is, failure of the two lateral ossification centers to unite posteriorly in the midline); type B, unilateral clefts, ranging from a small defect to a complete absence of one hemiarch; type C, bilateral defects with preservation

Table I: Whole Literature Analysis of C1 Hypoplasia Between Years 1937 and 2009

Author	Year
Plaut	1937
Lawrence & Anderson	1937
Ghislanzoni	1953
Fiorani-Gallotta & Luzzati	1955
Garber	1964
Desgrez et al.	1965
Holsten	1968
Dalinka et al.	1972
Logan & Stuard	1973
Richardson et al.	1975
Schulze & Buurman	1980
Galindo & Francis	1983
Raininko et al.	1984
Freidman & Jacobs	1985
Dorne et al.	1986
Nakamura et al.	1987
Spadora et al.	1987
Motatenau et al.	1991
Martich et al.	1992
Russell	1993
Chigira et al.	1994
Currarino et al.	1994
Torremán et al.	1996
Devi et al.	1997
Phan et al.	1998
Hierholzer et al.	1999
Sharma et al.	2000
Hosalkar et al.	2001
Urasaki et al.	2001
Berger & Alexandrou	2002
Pérez-Vallina et al.	2002
Torriani & Lourenço	2002
Gangopadhyay & Aslam	2003
Lee et al.	2003
Klimo et al.	2003
Garg et al.	2004
O'Sullivan & Mcmanus	2004
Schröder et al.	2005
Saiguchi et al.	2006
Pasku et al.	2007
Senoglu et al.	2007
Tan et al.	2007
Sabuncuoğlu et al. (present case)	2009

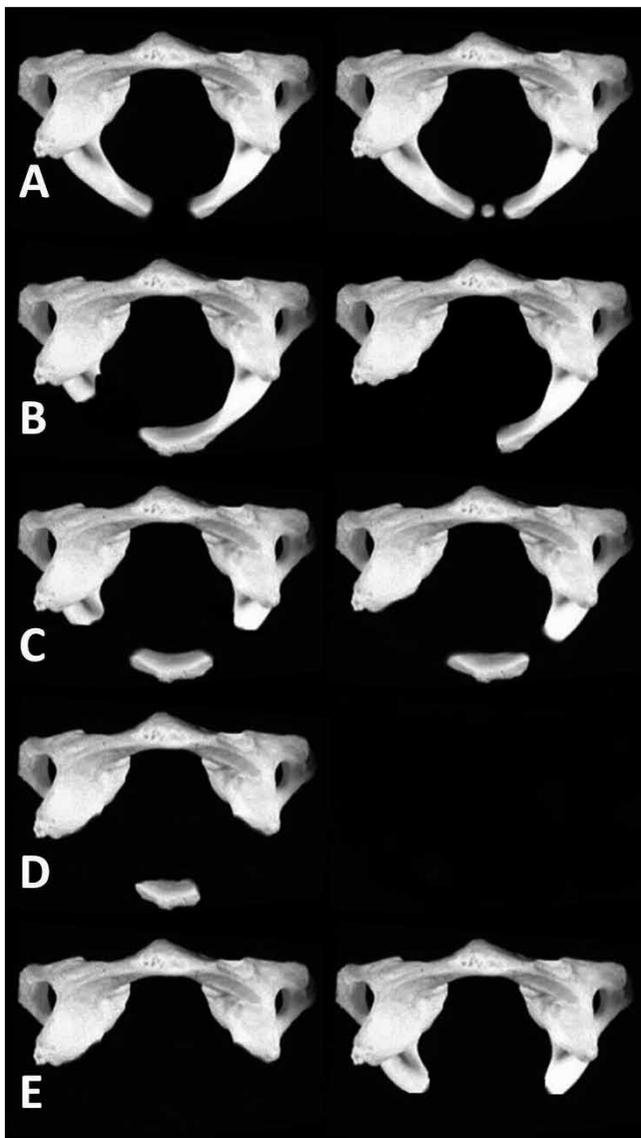


Figure 8: The figure shows the types of Curarino's classification in hypoplasia of C1.

Table II: Currarino's Morphologic Classification for C1 Hypoplasia

Type	Description
A	Failure of posterior midline fusion of the two hemiarches
B	Unilateral defect
C	Bilateral defects
D	Absence of posterior arch, with persistent posterior tubercle
E	Absence of the entire arch, including the tubercle

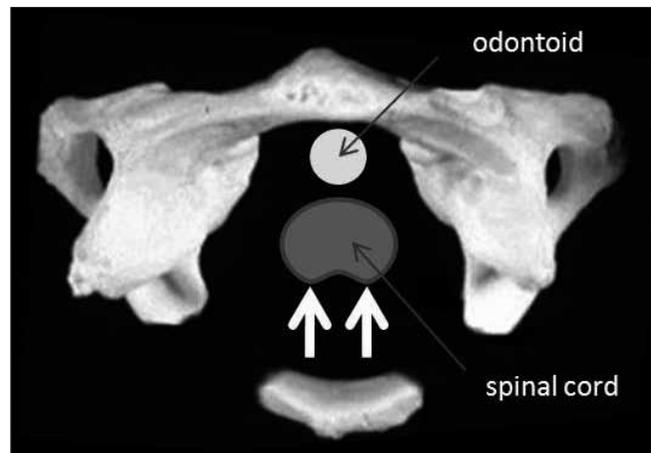


Figure 9: Spinal cord compression by the inward mobility of the isolated posterior bony fragment (arrows) can lead to some neurological symptoms in the patient. The presence of an posterior isolated bony fragment is a potential risk factor for neurological morbidity.

of the most dorsal part of the arch; type D, absence of the posterior arch with persistent posterior tubercle and type E, the complete absence of the posterior arch and posterior tubercle (2). The frequency of only type A is nearly 3-4% of individuals and these make up more than 90% of all posterior arch defects of C1 (2). This type is difficult to diagnose from lateral films of the cervical spine, and its diagnosis cannot be made with certainty in the first 5 to 10 years of life when two hemiarches of C1 may still be unfused normally (2). Patients with types C and D, have a free-floating posterior tubercle at the apex of the arch. It is hypothesized that the patient has both error of chondrification as well as the rare fourth ossification center to form this anomaly (11). A free-floating posterior tubercle is a problem in patients with C1 hypoplasia. This defect seems to suggest that the presence of an isolated posterior bony fragment somehow predisposes the patients to neurological morbidity. Richardson et al. tried to explain the symptoms of intermittent quadriplegia with compression of the cord by the inward mobility of the isolated posterior bony fragment during extension of the cervical spine (Figure 9) (20,24). Syring formation can be seen in these patients. This hypothesis supports the notion that the cord impingement occurred during extension, when the cord is expected to move superiorly in relation to the posterior arch. On the basis of the literature, the presence of an posterior isolated bony fragment is a potential risk factor for neurological morbidity rather than as an inconsequential normal variant (24).

Curarino et al, have also subdivided the patients into five clinical groups: 1) asymptomatic incidental findings, 2) neck pain or stiffness after trauma to the head or neck, 3) chronic symptoms referable to the neck, 4) various chronic neurological problems, and 5) acute neurological symptoms after minor cervical trauma (2,16).

Cervical lateral plain radiography, 2D or 3D reconstructed CT and MRI are very useful and important tools for evaluation of the patient. Congenital anomalies of the craniocervical junction are common incidental findings on cervical plain radiography; however, some may cause confusion when routine radiographs show apparently abnormal findings. These anomalies should be kept in mind to avoid misinterpretation as fractures, luxation or osteolysis (2,5,8,9,19). Cervical lateral plain radiography with flexion and extension views may help to detect cases with mechanical instability where there is increased risk of spinal cord injury and determine the mobility of the isolated tubercle (21,24,26). CT also provides excellent contrast between the nonossified and ossified portions of the arch of C1, which can enable precise determination of the extent of the abnormality (29). Additionally CT scans can be particularly useful for smaller defects and 2D or even 3D reconstruction might display better the topography of the upper cervical spine, especially to clinicians (9). MRI study has advantages in evaluating the spinal cord, adjacent neural structures and paraspinal soft tissue (29). Although MRI has not been performed in most of the case reports due to absence of neurological symptoms, an MRI study is needed in some symptomatic cases to determine the secondary changes within the spinal cord (2,16,29). These changes possibly represent focal myelomalacia, cord edema or a presyrinx state (12). It is possible that cord compression could have been directly visualized had the MRI study been performed with the neck extended (24).

Our patient may be classified as type A, clinical subgroup 1, according to Currarino's morphologic and clinical classification (2). Absence of ossification of the posterior arch of the C1 is asymptomatic and is generally detected as an incidental finding like our patient, although it may cause neck pain in some patients (29). Autopsies and surgical explorations have shown that dense fibrous bands bridge the bony gaps and account for good stability of the upper cervical spine. In the latter, hypoplasia of the posterior arch of C1, which may increase the risk of atlantoaxial subluxation, was seen in 26% of 38 children aged 2-3 years (16,29). Compensatory hypertrophy of the anterior arch of C1 and of the spinous process of C2 is usually found (15,29). Other coexisting abnormalities, including clefts of the anterior arch, atlantoaxial rotator subluxation, anterior atlantoaxial subluxation, downward projection of the posterior border of the foramen magnum and cervical myelopathy are reported in the literature (29). This anomaly can also stimulate basilar invagination, Jefferson's fracture and occipitalization of the C1 (15,25,29).

Congenital hypoplasia of the C1 with posterior tubercle can be managed conservatively or by surgical resection. Patients with this anomaly should be advised to avoid contact sports. Currarino et al., describing a variety of neurological problems, observed that it is often not clear whether the symptoms are in fact attributable to C1 defect, but recommended that certain sports should be avoided if such defects are shown (2,9). Due to previous case reports and review of the literature,

type C and D should undergo surgery at an early stage to prevent cumulative damage to the cord (24). Surgery is the treatment of choice in symptomatic compression. Excision of the posterior arch is curative. Patients may be followed up for instability and treated as necessary (4). Posterior fusion is a common surgical procedure in cases of atlantoaxial instability, provided the posterior elements of both vertebrae are intact. Posterior fusion involves the occiput and lower spinal segments, limiting the mobility of the upper cervical spine, if the posterior arch of C1 is abnormal (29).

In conclusion, most congenital anomalies of upper spine are determined incidentally in asymptomatic patients. Once it has been found in young patient, the patient should be evaluated in detail with advanced radiological studies to avoid misinterpretation as fractures, luxation, osteolysis or instability. Consulting a radiologist could help making an accurate diagnosis and deciding on current therapeutic interventions.

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