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OLGU SUNUMU / CASE REPORT

Atlanto-Occipital Hypertrophy in Eight Years – Old - Girl: An Uncommon Case

Sekiz Yaşında Bir Kız Çocuğunda Atlanto-Oksipital Hipertrofi: Nadir Bir Olgu

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ABSTRACT

Introduction: The craniovertebral junction is an anatomical structure consisting of the occiput, atlas and axis surrounding the foramen magnum and differing functionally and developmentally from other spinal components. Atlanto-occipital region hypertrophy is one of the rare anomalies seen in this junction.

Case Presentation: In this article we presented an eight years old female patient admitted to our clinic with complaints of dizziness, facial numbness and vertigo. The patient's physical and neurological examination were normal. Magnetic resonance (MR) imaging revealed hypertrophic exostotic growth in the left lateral atlanto-occipital joint and some other malformations including cerebellar tonsil hypertrophy, type 0–1 Chiari syndrome, vascular loop syndrome, and cerebellar indentation due to hypertrophic exostotic structuring. The patient's MR angiography revealed encurvation in the right vertebral artery and severe hypoplasia in the left anterior cerebral artery (ACA) 1st segment.

Conclusion: The presented case is of particular importance among craniovertebral pathologies due to its rarity in childhood. Craniovertebral junction anomalies observed in the pediatric population should be carefully evaluated and investigated for underlying pathologies, and larger case series are needed to establish appropriate approaches.

Keywords: Atlanto-occipital junction, hypertrophy, child

ÖZ

Giriş: Kraniyovertebral bileşke; foramen magnumu çevreleyen oksiput, atlas ve aksisten oluşan, diğer spinal yapılardan fonksiyonel ve gelişimsel olarak farklılık gösteren anatomik bir yapıdır. Atlanto-oksipital bölge hipertrofisi, bu bileşkede görülen nadir anomalilerden biridir.

Olgu Sunumu: Bu makalede, kliniğimize baş dönmesi, yüzde uyuşma ve vertigo şikâyetleriyle başvuran sekiz yaşında kız hasta sunulmuştur. Hastanın fiziksel ve nörolojik muayenesi normaldi. Manyetik rezonans (MR) görüntülemede sol lateral atlanto-oksipital eklemde hipertrofik ekzostotik büyüme ve buna ek olarak serebellar tonsil hipertrofisi, tip 0–1 Chiari sendromu, vasküler loop sendromu ve hipertrofik ekzostotik yapılanmaya bağlı serebellar bası gibi bazı diğer malformasyonlar saptandı. Hastanın MR anjiyografisinde sağ vertebral arterde eğrilik ve sol anterior serebral arterin (ACA) 1. segmentinde ciddi hipoplazi izlendi.

Sonuç: Sunulan olgu, çocukluk çağında nadir görülmesi nedeniyle kraniyovertebral patolojiler arasında özel bir öneme sahiptir. Pediatrik yaş grubunda görülen kraniyovertebral bileşke anomalilerinin dikkatle değerlendirilmesi, altta yatan patolojiler açısından araştırılması ve daha geniş olgu serileriyle desteklenmesi gerekmektedir.

Anahtar Sözcükler: Atlanto-oksipital bileşke, hipertrofi, çocuk

Introduction

The craniovertebral junction (CVJ), consists of atlas (C1), axis (C2) and the occiput which surrounds foramen magnum. It is functionally and developmentally different from other spinal bone components. Unilateral hypertrophy of the atlanto-occipital and atlanto-axial joints, which are functional structures of the craniovertebral junction, is a very rare condition. It may cause neurological deficits such as chronic pain and spinal deformities which usually become symptomatic in adulthood (1). Here, we present a girl with atlanto-axial hypertrophy whose symptoms initiate at very early age.

Case Presentation

An eight years and one month old girl admitted to our clinic with complaints of dizziness, facial numbness and vertigo, which is present for one years. The patient also had knife stabbing headache, lasting 2-3 minutes and not accompanied by vomiting, which had been present for about a month and occurred 2-3 days

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in a week, especially after returning from school. In addition, the patient, who had been complaining of dizziness with vertigo lasting 2-3 minutes for about a month, occasionally complains of not being able to run fast although doing regularly sport activities.

History of the patient showed that she was born on time with C/S after a normal pregnancy and hospitalized for about 15 days with a diagnosis of hypoglycemia due to poor sucking after birth. She was discharged after being fed with nasogastric tube (NG) because of several low blood sugar levels. Then she fed from a regular feeding bottle. The patient's mental and motor development stages were normal. The patient's mother has a history of panic attacks and her father has a history of differential disease. The patient's physical and neurological examination revealed no abnormal findings other than a grade 2 systolic murmur. The laboratory findings including hemogram, liver and renal function tests, thyroid functions, vitamin B12, folic acid, ferritin levels were normal except for low vitamin D level of 14,12. (25-80 ng/L) Magnetic resonance (MR) imaging showed exostotic growth in the left lateral atlanto-occipital joint (Fig. 1) a 9 mm choroid plexus cyst, type 0-1 Chiari syndrome (Fig. 2) and linked multiple cystic structures approximately 4.6 x 5.9 mm, close to the anterior region of the cyst. The grey-white matter signal distribution of the cerebellar hemispheres was normal. MR angiography of the brain showed vascular loop syndrome. Encurvation and hypoplasia were found in the right vertebral artery (Fig. 3) and also A1 segment of the left anterior cerebellar artery was hypoplasic (Fig. 4). EMG and ECHO were normal and the patient was followed up intermittently.

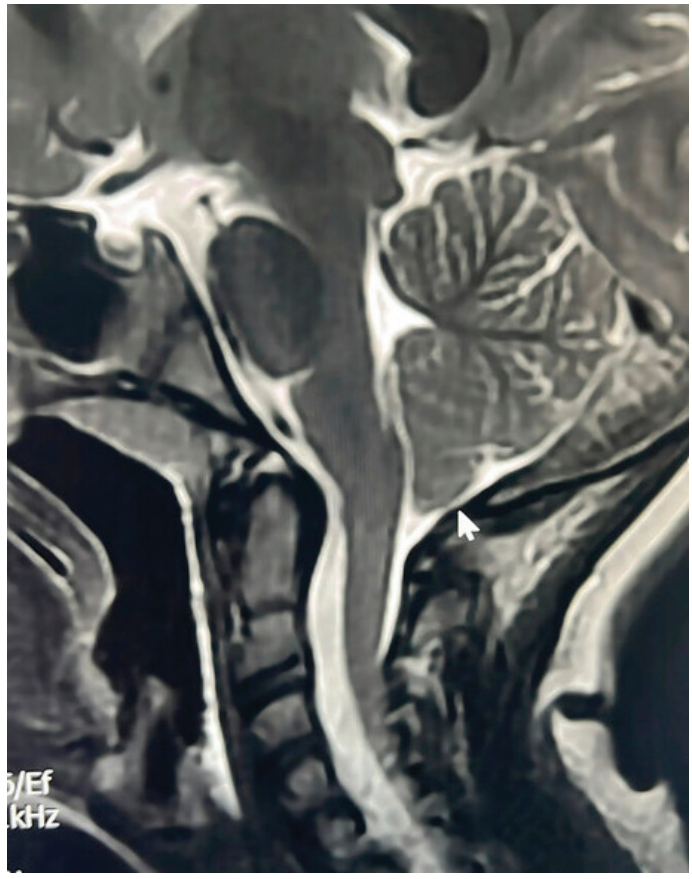

Figure 1. Hypertrophy of the left atlanto-occipital joint.

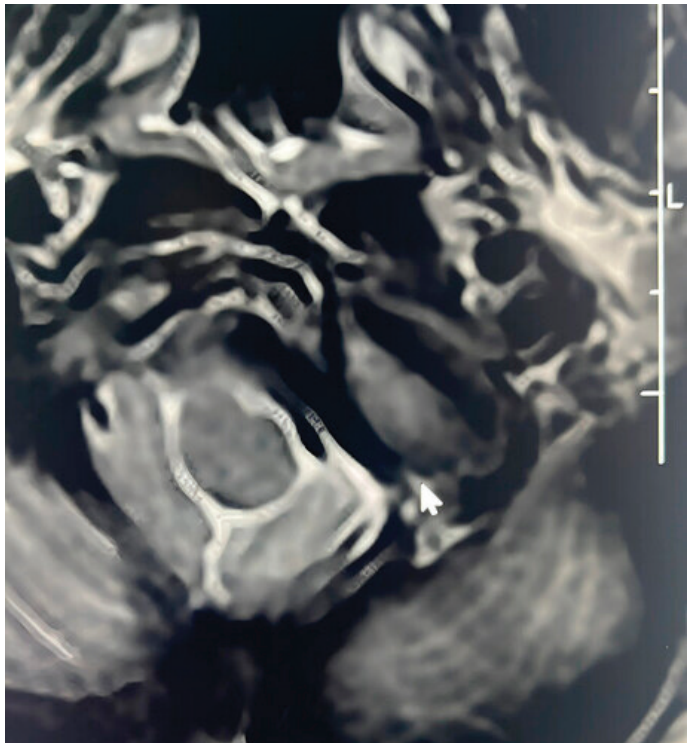

Figure 2. Hypertrophy of the cerebellar tonsil and minimal indentation at the level of foramen magnum. Type 0-1 Chiari malformation.

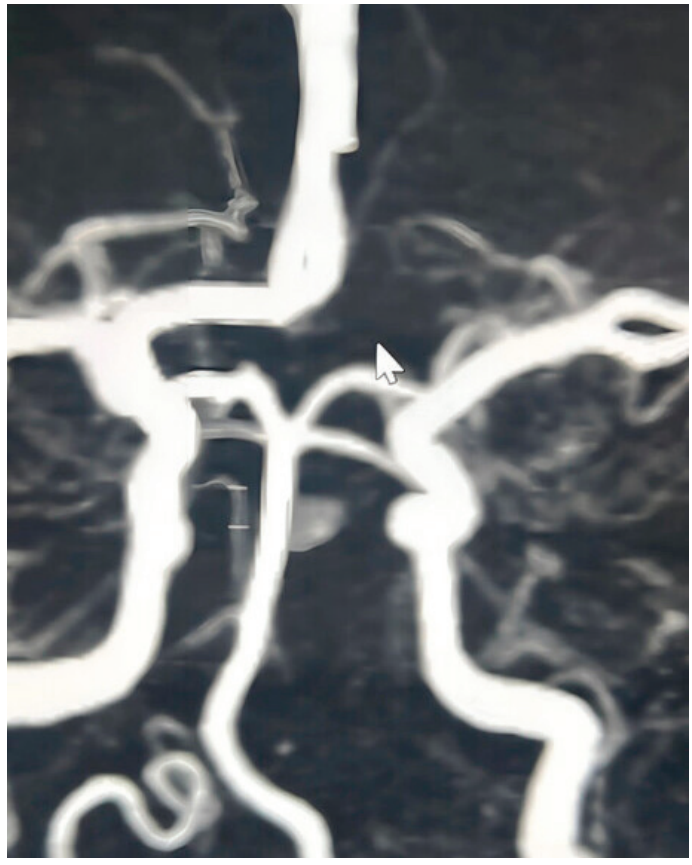

Figure 3. MRI angiography revealed hipoplasia of the right vertebral arteria.

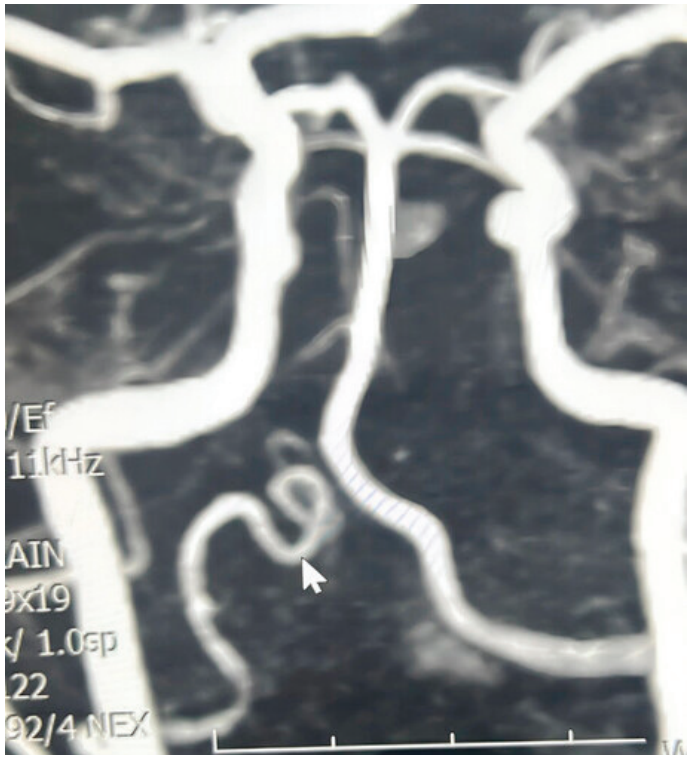

Figure 4. MRI angiography evaluation showed severe hypoplasia of the left anterior cerebral arteria (ACA) 1st segment.

Discussion

Congenital anomalies are commonly encountered in the craniovertebral junction (CVJ) consist of the occiput, atlas (C1) and axis (C2) region due to unique embryologic development. Atlanto-occipital and atlanto-axial region anomalies, which are craniocervical junction pathologies, are rare pathologies. Malformations associated with this region have a potential of causing serious neurological and vascular deficit and may require surgery especially in the adult age group. The incidence of different types of CVJ anomalies varies with demographic environment and ill-defined genetic factors. The clinical features are generally delayed up to 2nd or 3rd decades, since they are subtle and often missed (2).

The first case report in the literature was published by Goel et al in 2006 (3). They described 4 patients aged between 21 and 54 years. It was reported that these patients had unilateral atlantalateral mass hypertrophy and this condition present with symptoms like slow progressively torticollis and myelopathy. They suggested that a hypertrophied mass may be due to a congenital abnormality or a slow growing pathological entity. In our case we thought that this hyperplasia may be a congenital abnormality because of its association with other abnormalities including Chiari malformation, right vertebral arterial and left cerebral arterial first segment hypoplasia and vascular loop syndrome.

Menezes and Feroy described patients with congenital CVJ anomalies causing neural compression usually early in first and second decades of life (4). Nath et al. (5) reported unilateral atlanto-occipital lateral mass hypertrophy with accompanying right atlanto-axial dislocation and Chiari 1 malformation in a 22-year-old patient with spastic quadriparasia and cervical myelopathy findings worsening over time.

In our case, hypertrophic exostotic structure in the left lateral aspect of the atlanto-occipital joint and cerebellar indentation due to this structure were found in an eight years old girl with complaints of headache, dizziness, numbness in the face and vertigo. The first difference that distinguishes our case from other cases is that our patient was in the paediatric age group, unlike previous cases in the literature. Another difference is that, while slow-developing myelopathy was the main symptom in other cases in the literature, in our case, neurological symptoms such as headache, facial numbness and vertigo. Also cerebellar symptoms which are presumed to be due to compression of the cerebellum by the hypertrophic exostotic structure are at the forefront. This situation shows us that this pathology may show a broad clinical spectrum and may present with different clinical findings in paediatric age.

The etiology of this pathology is controversial. Goel, et al. (3) stated that this pathology may be a congenital anomaly but could not establish a complete relationship. Nath, et al. (5) suggested that it may be related with malrotation and proatlas segmentation disorders in the developmental process. In our case, the presence of findings including choroid plexus cyst, encurvation and hypoplasia in the right vertebral artery and hypoplasia in the right cerebral artery seen on MR angiography can indicate that an underlying developmental pathology. However, the real cause is open to debate. Due to the different presentation of craniovertebral anomalies in different age groups and the insufficient number of cases in the literature (2).

Another important point in the literature is the treatment approach. There is no standardised approach to the treatment of atlanto-occipital and atlanto-axial joint hypertrophy. In adult cases in the literature, posterior decompression and stabilisation are recommended when it is necessary (3,5). We preferred clinical follow-up because our case was in the paediatric age group and did not resemble clinical and neurological findings. Identification of these patients may sometimes be difficult considering the rarity, early age of the findings, and difficulty in identifying the mass on radiological films. High resolution CT scans and MRIs may be required to visualise hypertrophy of the atlas and the structure of the cord.

Conclusion

The presented case has a special importance among the craniovertebral pathologies reported in the literature due to its rarity in childhood it can develop on different pathological bases and lead to different clinical pictures in both adults and children. Therefore, we believe that craniovertebral junction anomalies

seen in pediatric age should be carefully evaluated, investigated for other underlying pathologies, and patients should be followed up at regular intervals. But, larger case series are needed to make comments and approaches on this issue in childhood.

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