Acta Orthop. Belg., 2007, **73**, 282-285

CASE REPORT

Congenital posterior atlas defect associated with anterior rachischisis and early cervical degenerative disc disease : A case study and review of the literature

Dritan PASKU, Pavlos KATONIS, Apostolos KARANTANAS, Alexander HADJIPAVLOU

From the University of Crete Heraklion, Greece

A rare case of a wide congenital atlas defect is reported. A 25 year-old woman was admitted after complaints of radicular pain in the right arm. Radiographs incidentally revealed aplasia of the posterior arch of the atlas together with anterior rachischisis. A review of the literature is presented and a possible association with early disc degeneration is discussed.

Keywords : spine ; congenital disorders ; computed tomography ; MR imaging ; disc degeneration.

INTRODUCTION

Malformations of the atlas are relatively rare and exhibit a wide range including aplasia, hypoplasia and various arch clefts (*2, 15*). The reported incidence in a large study of 1,613 autopsies with regard to presence of congenital aplasia is 4% for the posterior arch and 0.1% for the anterior arch (*5-8*). The onset of ossification of the posterior arch of the atlas occurs during the seventh week of intrauterine life proceeding perichondrically from two centers located in the lateral masses. Complete fusion of the posterior arch is expected to occur between 3 and 5 years of age. At least two anomalies can develop during the ossification process : 1) median clefts of the posterior arch, and 2) varying degrees of posterior arch dysplasia (*4, 16*). Generally, the patients are asymptomatic and most of them are discovered incidentally. We describe the case of a young woman with a previously undiagnosed posterior atlas defect coexisting with an anterior rachischisis, presenting with radicular arm pain resistant to conservative therapy. In addition, a review of the literature is presented with emphasis on the possibility of the association between the atlas defect and early disc degeneration.

CASE REPORT

A 25 year-old woman presented with neck pain radiating to the right arm over the last 5 days. She also reported intermittent neck and arm pain for the past 4 years. The patient had consulted in our hospital for an episode of cervical pain one year previously without arm pain but was discharged from the emergency department without any radiological examination. Her symptoms deteriorated with neck flexion, with pain referred to the upper thoracic

■ Dritan Pasku, MD, Resident in orthopaedic surgery.
■ Pavlos Katonis, MD, Assistant Professor in orthopaedics.
■ Alexander Hadjipavlou, MD, Professor and Chairman.
Department of Orthopaedic Surgery and Traumatology, University of Crete Heraklion, Greece.
■ Apostolos Karantanas, MD, Associate Professor of radiology.
Department of Radiology, University of Crete, Heraklion, Greece.
Correspondence : Dritan Pasku, 2 G. Gennimatas, 71414, Tsalikaki, Heraklion, Crete, Greece.
E-mail : paskudr@hotmail.com.

No benefits or funds were received in support of this study

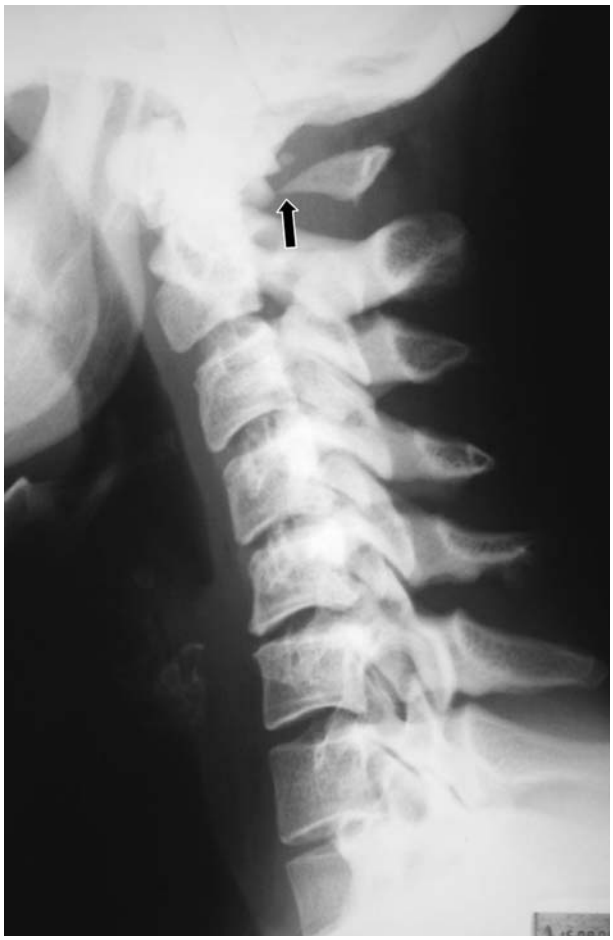

Fig. 1. — Lateral radiograph of the cervical spine reveals aplasia of the posterior arch of the atlas (arrow) with an isolated posterior bony fragment.

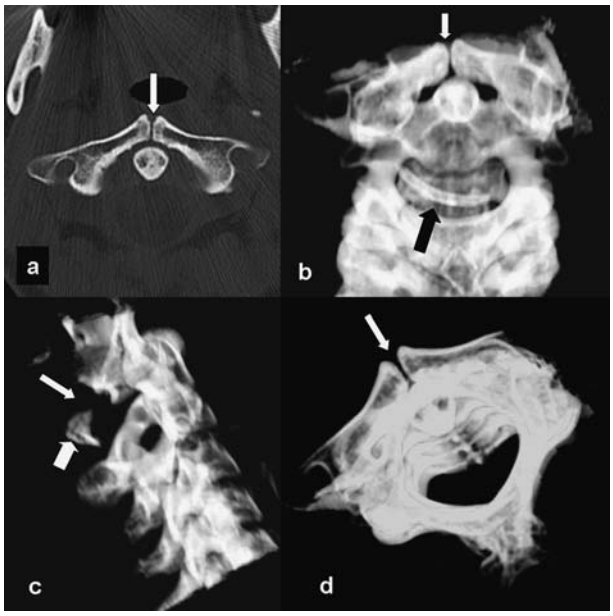

Fig. 2. — Multidetector computed tomography of congenital atlas dysplasia. a) The axial high resolution image shows the anterior rachischisis (arrow). b) 3D reconstruction shows the anterior rachischisis (arrow) and the isolated tubercle (black arrow). c) The lateral 3D reconstruction shows to better advantage the complete atlas cleft (arrow) and the tubercle (thick short arrow). d) Anterior rachischisis is obvious from another 3D reconstruction (arrow).

region. Physical examination revealed a minor motor deficit of the brachioradialis and triceps muscles on the left side. No sensory loss was revealed and reflexes were normal bilaterally. The Hoffmann, Romberg, Babinsky and Lhermitte signs, were negative.

Plain radiographs and a multi-detector computed tomography scan of the cervical spine revealed aplasia of the posterior arch of the atlas with a posterior bone tubercle (figs 1, 2). Anterior rachischisis of C2 was also found. The patient was admitted to the orthopaedic department with a soft collar and analgesics. The MR imaging study performed the following day revealed a moderate disc herniation at C4-C5 level, a small disc herniation at C5-C6 level and confirmed the absence of the posterior arch of the atlas. The patient was managed conservatively.

DISCUSSION

The embryology of the atlas is unique as it is the only vertebra to develop from only two lateral ossification centers, one in each lateral mass. These nuclei fuse on the midline posteriorly to form the posterior arch of the atlas. The anterior arch results from the anterior fusion of the nuclei with a dense band of tissue known as the hypochordal bow. In 20% of newborns a third ossification center exists in the anterior arch. A failure in ossification of the anterior part leads to rachischisis (*2*). A separate ossification center develops in the posterior cartilaginous cleft during the second year of life. This center is responsible for the complete fusion of the posterior arch of the atlas. A failure of chondrogenesis in this phase may lead to a disturbed

Table I. — Classification of congenital malformations of the atlas according toVillas *et al* (*18*)

Area 1 : Defect of formation of the posterior arch
Area 2 : Defect in union of the posterior arch with the superior articular facet
Area 3 : Defect of formation with hypoplasia or agenesis of the superior articular facet
Area 4 : Defect of formation of the anterior arch
Area 5 : Malformation of the atlanto-occipital junction
Area 6 : Malformation of the atlanto-axial junction

Table II. — Classification of the congenital defects of the posterior arch of the atlas according to Currarino *et al* (*3*)

Type A : Failure of posterior midline fusion with a small gap remaining
Type B : Unilateral clefts
Type C : Bilateral defects with preservation of the most dorsal part of the arch
Type D : Complete absence of the posterior arch with a persistent isolated tubercle
Type E : Complete absence of the entire posterior arch

ossification. This was supported by the findings at autopsies or intraoperatively, where it was found that connective tissue bridged the bony defects (*5-8, 10, 13*). Congenital absence or hypoplasia of the posterior arch of the atlas may be associated with several disorders, such as the Arnold-Chiari malformation, gonadal dysgenesis and the Klippel-Feil, Down and Turner syndromes respectively (*4, 12, 16*). On the other hand, it has been reported that hypoplasia of the posterior arch of the atlas may increase the risk of atlantoaxial subluxation in about 26% of children aged 2-3 years (*11*). Motateanu *et al* (*12*) reported an affected mother and daughter and Currarino *et al* (*3*) an affected mother and son, suggesting an autosomal dominant inheritance. The anomalies of the upper cervical vertebrae occur more frequently in individuals with cleft lip, cleft palate or both (*17*). The presence of a fixed torticollis may hide a hypoplasia of the atlas in childhood (*9*). Villas *et al* (*18*) presented in 1990 an anatomic classification of the defects of the atlas (table I). A complete classification of congenital anomalies of the posterior arch of the atlas is proposed by Currarino *et al* (*3*), based on seven of their own cases and 39 other cases described in the literature up to 1994 (table II). The incidence of a type A anomaly is estimated to be approximately 97% whereas only 0.69% of the general population has a type B-E anomaly (*3*). In their study Currarino *et al* (*3*) have 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. In the literature, all the case reports highlighted the role of the abnormality in the development of cervical myelopathy. In most of the published cases, MR imaging has not been performed due to absence of neurologic symptoms (*3, 16*). In symptomatic patients though, MR imaging is able to depict the secondary changes within the spinal cord such as myelomalacia, cord oedema or a presyrinx state (*14*). Richardson *et al* (*13*) presented an intermittent quadriparesis in a 15 year-old boy and suggested that the symptoms were secondary to compression of the cord by the inward movement of the isolated posterior bony fragment during extension of the cervical spine. Patients presented in the literature are predominantly children or women in the second or third decade of life (*1, 14*). All patients who presented with significant neurological findings had a type C or D anomaly (*3, 10, 13, 14*).

Our patient may be classified as type D, clinical subgroup 1, according to Currarino *et al* (*3*) ; the main symptoms in this case were radicular pain and neck stiffness. MR imaging revealed disc degeneration with herniation at the C4-C5 and C5-C6 levels. There were no risk factors in our patient to contribute to the disc disease such as profession, dancing and contact sports activities. The possible association between congenital abnormalities of the atlas and early disc degeneration has not been addressed in the literature. One patient, a 30-year-old man, presented by Sharma *et al* (*14*) presented with a disc protrusion at C5-C6. One possible explanation for the early degenerative disc disease might be the altered stability of the upper cervical spine resulting in increased forces applied to the lower levels.

Dynamic MR imaging in order to show cord compression by the isolated bone tubercle might be

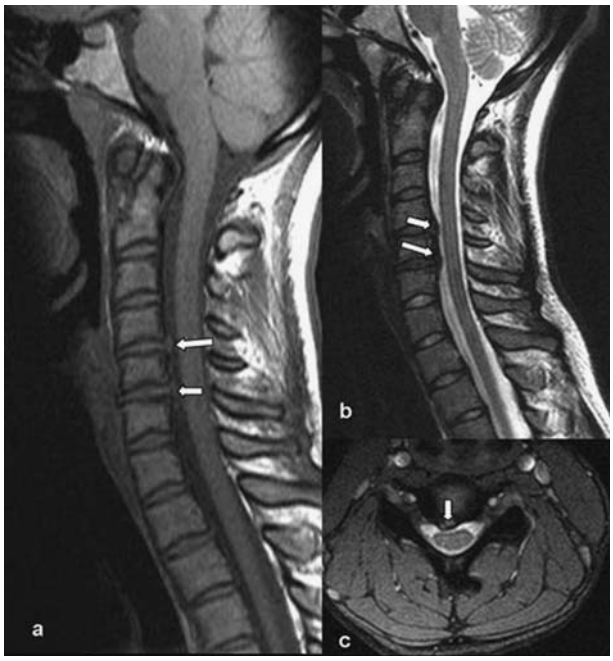

Fig. 3. — MR imaging with sagittal T1-weighted (a) and sagittal T2-weighted (b) images, show the disc herniations at the C4-C5 and C5-C6 levels (arrows). The axial 2D multi-echo gradient echo image at the C4-C5 level (c) shows the extruded disc herniation to provoke moderate displacement of the thecal sac (arrow).

of help to select patients who should avoid contact and other strenuous sports.

CONCLUSION

We have described a rare case of congenital posterior atlas defect associated with anterior rachischisis and early degenerative disc disease, studied with MR imaging and multi-detector computed tomography. Anomalies of the atlas are rare and in general asymptomatic, but physicians must be familiar with their clinical presentation, occasionally complicated by dynamic cord compression. Secondary changes involving disc degeneration should be confirmed in prospective studies.

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