
CASE REPORT

Congenital Posterior Arch Defect of the Atlas: Report of a Rare Anomaly

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ABSTRACT

Congenital defects of the posterior arch of the atlas are rare, with a reported prevalence of 0.95 to 4%. The defects have been classified into five types (A-E), with type A being most common and types B-E being found only in 0.69% of the general population. Although most patients are asymptomatic, some may present with chronic neck pain, headache, Lhermitte's sign, transient quadriparesis or even acute neurological deficits. The defects may also mimic a Jefferson fracture leading to a wrong diagnosis. Despite its rarity, knowledge of this congenital posterior arch defect of the atlas is important for avoiding diagnostic errors and to provide information on its management. We are therefore reporting a case with this congenital defect discovered as an incidental finding and provide a literature review about this rare anomaly.

Key Words: Cervical atlas; Congenital abnormalities; Musculoskeletal abnormalities; Spinal fractures; Tomography, X-ray computed

中文摘要

先天性寰椎後弓缺陷：一宗罕有病例的报告

朱志揚、陳德文、馮啟邦、鄭志成

先天性寰椎後弓缺陷症非常罕見，據文獻報告，發病率只有0.95%至4%。這症狀被分為A至E五種類型，其中A型最常見，而B至E型只佔一般人口的0.69%。雖然大部分患者並沒有病徵，有些患者卻出現慢性頸部疼痛、頭痛、拉密特徵（Lhermitte's sign）、暫時性的四肢輕癱或急性神經功能缺損。寰椎後弓缺陷症的病徵可能會與Jefferson骨折相似，引致誤診。雖然寰椎後弓缺陷症很罕見，認識此症對於治療及避免誤診很重要。所以特此報告這宗因偶然而發現的寰椎後弓缺陷症病例，並對此病症作文獻回顧。

INTRODUCTION

Congenital defects of the posterior arch of the atlas are rare.¹⁻³ The reported prevalence ranges from 0.95 to 4%.¹ These congenital defects have been classified into five different types by Currarino et al,⁴

and are considered to be benign, since most patients are asymptomatic.^{1,2,5,6} However, their detection is nevertheless clinically important, since at times they can cause chronic neck pain or even myelopathy.^{6,7} Some of the defects can mimic a Jefferson fracture and lead

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to misguided clinical management.⁸ Knowledge of this congenital defect is useful in order to avoid diagnostic errors, especially in patients who have suffered cervical trauma.⁶ We therefore report a case with anomaly as an incidental finding and provide a review of the literature about this rare anomaly.

CASE REPORT

A 34-year-old male construction site worker, with good past health, was struck by metal rod over his left face during work. Left periorbital swelling, depression, and laceration over left cheek were noted. He also complained of diplopia on bilateral gaze. There was no neck pain or tenderness.

Plain computed tomography (CT) of the brain and facial bones was performed, which revealed no definite intracranial abnormality. However, a left zygomaticomaxillary complex fracture was noted, with fractures of left maxillary antrum, inferior rim and floor and the lateral rim and wall of the left orbit, and left zygomatic arch, but there was no entrapment of orbital content. The right posterior arch of atlas

was incidentally found to be absent, likely due to a congenital anomaly. Anteriorly, there was an unclosed right transverse foramen of the atlas (Figure 1). No other abnormality was detected in the upper cervical spine.

An urgent operation was performed with open reduction of malar and zygomatic fractures, repair of the orbital floor fracture with bone grafts and bone grafting for the left zygoma. The patient recovered uneventfully, never complained of neck pain and no neurological deficit was detected.

DISCUSSION

The atlas has three primary ossification centres, with an anterior centre forming the anterior tubercle and two lateral centres forming lateral masses and posterior arches.^{1-3,7,9} In 2% of the population, there is a fourth centre forming a posterior tubercle.^{3,7,9-11} During the seventh gestational week, the lateral centres extend dorsally to form the posterior arch.^{2,3} At birth, the posterior arches are nearly fused except for several millimeters of cartilage.^{1,7,8} Complete fusion of posterior arches is expected to occur between the ages of 3 and 5

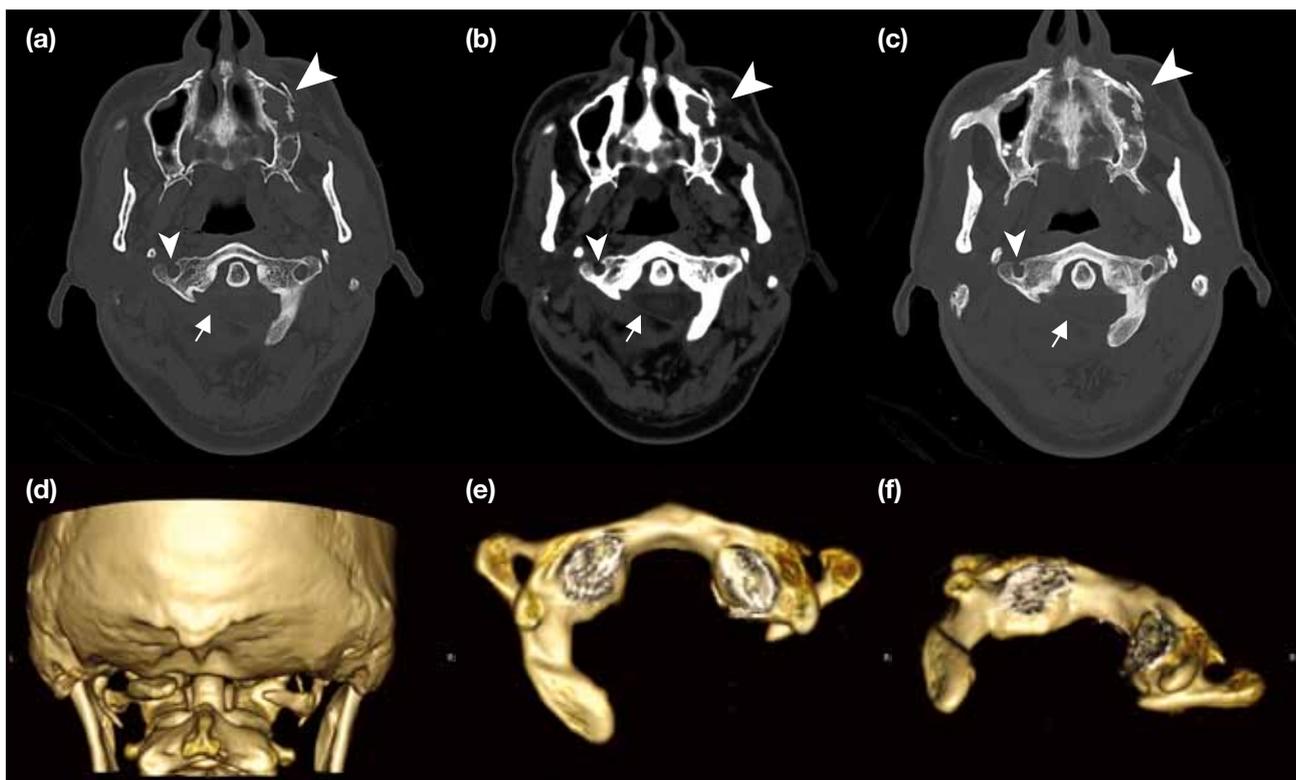


Figure 1. Computed tomographic scans at C1 level (a) in bone window, (b) in soft tissue window, and (c) maximum-intensity-projection image of bone window show congenital defect of the right posterior arch of atlas (arrows) and anteriorly unclosed right transverse foramen of atlas (small arrowheads). Left zygomaticomaxillary complex fracture is also seen (large arrowheads). (d) 3-Dimensional (3D) volume-rendered image of the skull base in posterior view shows that the odontoid process of C2 vertebra can be clearly seen because of the posterior arch defect of atlas. (e and f) 3D volume-rendered images of isolated C1 vertebra (superior view and superior oblique view) clearly demonstrate the posterior arch defect and anteriorly unclosed right transverse foramen of atlas.

years.^{1,10,11} The anterior centre usually fuses with the two lateral centres at about 5 to 9 years of age.^{1,3,7,9} At times, however, no ossification centre arises in the anterior arch, instead the arch forms from ventral extensions of the lateral masses.⁸

Defects of the posterior arches are thought to occur because of a failure of local chondrogenesis rather than subsequent ossification.^{1,7,9,10} This hypothesis of the underlying cause is supported by the fact that the region of atlas devoid of the posterior arches is found to be crossed by connective tissue rather than by cartilage.^{7,10}

The various congenital defects of the posterior arch of the atlas were first summarised by von Torklus and Gehle¹² into six forms: total aplasia, aplasia with persistent posterior tubercle, aplasia with paramedian

unilateral remnant of the arch, bilateral remnant of the arch with cleft formation, hemiapsia, and unilateral posterior arch partial aplasia.^{8,13}

Subsequently, Currarino et al⁴ modified and then developed a new classification system for congenital defects of the posterior arch of atlas, which is now widely used (Figure 2):

- Type A — Failure of midline fusion of the two hemiarches, i.e. failure of the two lateral centres to unite posteriorly;
- Type B — Unilateral cleft ranging from a small defect to the complete absence of one hemiarch and posterior tubercle;
- Type C — Bilateral clefts of the lateral aspects with preservation of the most dorsal part of the arch;
- Type D — Absence of the posterior arch with a persistent posterior tubercle; and
- Type E — Absence of the entire arch including the tubercle.

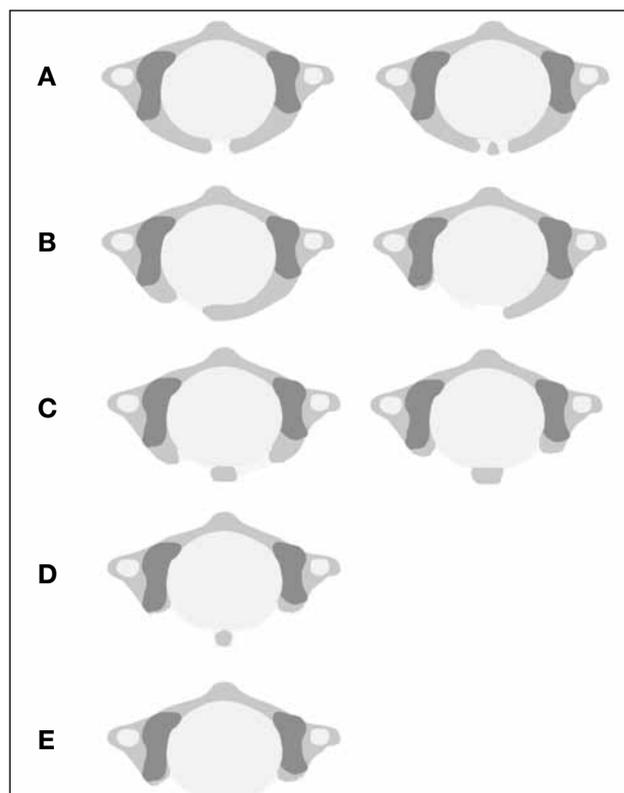


Figure 2. Classification of posterior arch defects of atlas by Currarino et al⁴ (modified from references 1 and 4). Type A: failure of posterior midline fusion of the two hemiarches. In some cases, a small separate ossicle may be seen within the gap. Type B: unilateral cleft; the cleft can range from a small gap to a complete absence of the hemiarch and posterior tubercle. Type C: bilateral clefts of the lateral aspects of the posterior arches; the most dorsal part of the arch is preserved. Type D: complete absence of the posterior arch with a persistent isolated tubercle; this anomaly is conceivably a more extensive form of bilateral clefts. The bilateral lateral parts of the posterior arch are absent, except for the posterior tubercle. Type E: absence of the entire posterior arch including the tubercle.

Type A is the most common type, affecting 4% of the general population.^{1,3} Other types (B-E) are found to affect 0.69% of the general population.⁴ According to the classification, our patient had a type B congenital defect, with complete absence of the right neural arch of the atlas and no posterior tubercle.

Anterior arch defects are even less common than defects of the posterior arch, with a reported prevalence of 0.09 to 0.1%.^{1,3} In most patients, the anterior cleft is associated with anomalous development of the posterior arch.¹ A rare entity called a bipartite atlas, with both anterior and posterior arch defects, has also been described.¹

Despite the rarity of posterior arch defect of the atlas, a number of diseases associated with this anomaly have been reported.⁹ Examples include: Arnold-Chiari malformations, gonadal dysgenesis, Klippel-Feil syndrome, Turner and Down syndromes.^{3,9,11} Synostosis of C2, C3 and C4 vertebrae has also been noted.⁹ There is also a report concerning a mother and her daughter with the same type of posterior arch defects of atlas, indicating autosomal dominant inheritance.¹¹

In general, posterior arch defects of the atlas are considered benign anatomical variations.^{1,3} Most patients are asymptomatic, the congenital defect being found incidentally.^{1,3,9,10} Variable symptomatic presentations have also been reported, including chronic neck pain, headache, Lhermitte's sign (an electrical sensation

running down the back and into the extremities), and transient quadriplegia.^{1,3,6} More sinister presentations include acute neurological symptoms or deficits, as well as atlantoaxial instability.^{3,10} Therefore, identification of this rare anomaly is clinically important. Patients who are symptomatic usually suffer from types C or D defects, with isolated posterior bony remnants.^{3,6,7,14} It has been hypothesised that the isolated posterior remnant moves anteriorly and may traumatise the dorsal spinal cord during extension of the cervical spine.^{6,7,14} The movement of the bony fragment during extension has been well demonstrated, supporting such a hypothesis.¹⁴

These congenital defects are sometimes found in lateral radiograph in the context of trauma.⁶ Dynamic cervical radiographs are helpful for evaluating mobility of the isolated posterior tubercle.¹⁵ On the other hand, CT can provide excellent contrast between the non-ossified and ossified portions of the posterior arch of atlas, and is particularly useful for detecting small defects.¹¹ CT is also useful for differentiating this anomaly from cervical fracture.⁶ For patients with neurological symptoms, however, magnetic resonance imaging (MRI) should be performed.^{2,11} MRI not only demonstrates the bony anomaly, it can also directly demonstrate any secondary changes within the spinal cord, such as T2 hyperintensity in the spinal cord.¹³ Such changes could be due to focal myelomalacia, cord oedema, or a 'presyrinx' state.¹⁴

When no neurological deficit is found, treatment is normally conservative.^{2,6} Surgery is indicated when there is atlanto-axial instability and/or spinal cord compromise.^{3,6} Posterior spinal fusion may be performed in cases of atlanto-axial instability.¹¹ For patients suffering from neurological symptoms due to either type C or D defects following isolated posterior tubercle, surgical excision of the posterior tubercle has been reported to be curative.^{1,15} Early surgery is recommended for these patients, so as to avoid cumulative damage to the spinal cord.¹⁵ Notably, in some patients even a trivial trauma can lead to severe neurological deficits.¹ Some authors therefore recommend that an isolated posterior bony fragment should be viewed as a potential cause of significant neurological morbidity rather than merely a normal variant.¹⁴

CONCLUSION

Congenital defects of the posterior arch of atlas are uncommon and patients are mostly asymptomatic.

Nevertheless, some patients with isolated posterior tubercles may present with neurological deficits and secondary changes in the spinal cord. The latter subjects can benefit from early surgical intervention. Here we have described a patient with an incidental finding of a congenital defect in the posterior arch of atlas, who presented after trauma with zygomaticomaxillary fracture. Fortunately, no neurological deficit ensued. After review, we believe that this rare anomaly is not necessarily merely an incidental finding. Knowledge about this entity is important, when it comes to avoiding diagnostic errors and providing useful approaches to further management.

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