Case Report

Congenital malformed posterior arch of atlas with fusion defect: a case of developmental canal stenosis causing cervical myelopathy

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Abstract: Congenital anomalies of the posterior arch of the atlas (PAA) are usually asymptomatic and diagnosed incidentally. Very rarely, they present with cervical myelopathy, usually being associated with partial aplasia or agenesis of PAA. We describe a 44-year-old lady with cervical myelopathy secondary to a malformed PAA with developmental atlas-level spinal stenosis and a congenital posterior fusion defect with persistent midline cleft showing significant non-osseous fibro-cartilaginous hypertrophy, causing critical cord compression. Spinal decompression by en-bloc wide excision of anomalous arch with occipito-cervical fusion was performed. Post-operatively, the patient's neurology improved gradually over 12 months, with radicular symptoms being the earliest and gait disturbance being the last symptom to resolve. At 24 months, she was asymptomatic with imaging showing good spinal cord decompression at the level of atlas. Developmental atlas stenosis with hypertrophic posterior arch fusion defect is an unusual cause of cervical myelopathy, which can be effectively treated with decompression with/without stabilization. Being aware of such an entity can avoid diagnostic dilemma and facilitate prognostication of outcomes, accurate surgical planning in the stenotic segment thereby ensuring effective management of these patients.

Keywords: Cervical atlas; congenital defects; spinal cord compression; surgical decompression; spinal fusion

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Introduction

Cervical stenosis has been reported in 4.9% of the adult population, 6.8% of the population 50 years of age or older and 9% of the population 70 years of age or older, involving mainly the subaxial cervical spine (1). Upper cervical stenosis is a relatively rare entity, being either congenital or acquired in origin. While the acquired causes vary from infection and trauma to rheumatoid arthritis; congenital causes include os odontoideum, congenital clefts or aplasias of the arches of atlas, hypertrophy of the dens, lateral masses or the transverse atlantal ligament, either of which may rarely present as cervical myelopathy (2-4). Congenital anomalies of the posterior arch of the atlas (PAA) although rare, are well documented, being usually asymptomatic and diagnosed incidentally (5-7). Anomalous hypertrophy of the PAA presenting as cervical myelopathy is usually associated with partial aplasia or agenesis of PAA (4). We hereby present a case of congenital malformed PAA with posterior midline fusion defect and secondary developmental canal stenosis at the level of atlas, presenting with cervical myelopathy.

Case presentation

A 44-year-old Indian housewife, presented to the clinic with complaints of persistent neck pain for 6 weeks, which had gradually progressed to radiate into the right half of the body, associated with tingling and numbness in the right half of body. There was no history of trauma, any joint stiffness, fever, weight loss or stigmata of tuberculosis. Examination demonstrated terminal restriction in all ranges of neck motion. Neurological assessment revealed myelopathic gait with bilateral upper limb and lower limb

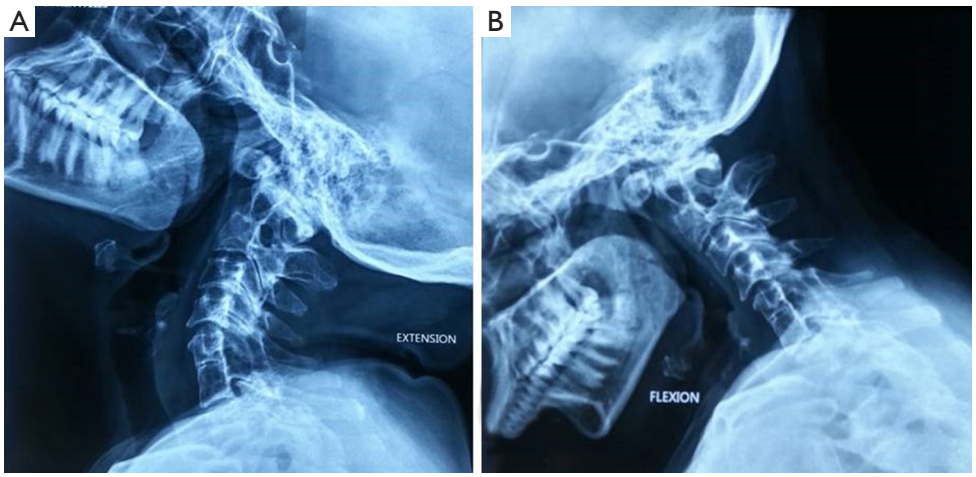

Figure 1 Pre-operative lateral radiographs (A) extension and (B) flexion views showing no evidence of atlanto-axial instability, normal looking PAA and fused C2–C3 and C6–C7 vertebrae. PAA, posterior arch of the atlas.

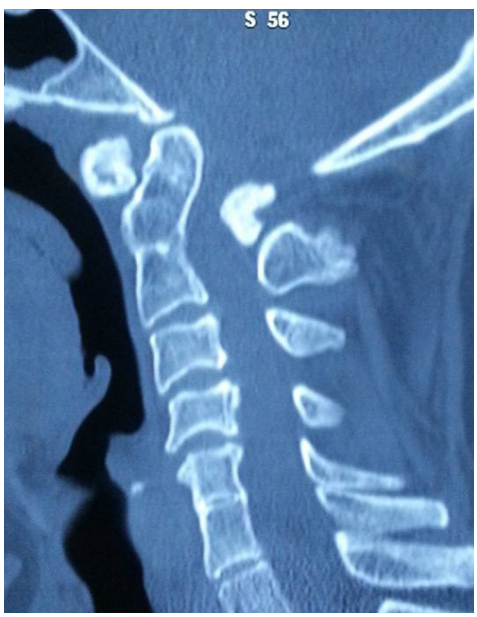

Figure 2 Sagittal CT showing the bony posterior arch projecting into the spinal canal, severely reducing the SAC. Note the sagittal SAC measured from the posterior surface of dens to the anterior-most part of PAA was 6.36 mm. CT, computed tomography; SAC, space available for cord.

hyperreflexia, bilateral Babinski sign and Hoffman's reflex present.

Radiographs showed congenital fusion of C2–C3 and C6–C7 vertebrae, well maintained coronal and sagittal vertebral alignment without radiographic instability (*Figure 1*).

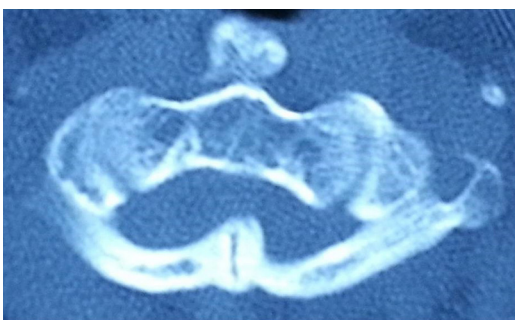

Figure 3 Axial computed tomography at level of atlas showing the malformed incurved PAA, with failure of fusion of the two hemi-arches with a midline cleft. There is osseo-cartilaginous hypertrophy of the midline fusion defect indenting sharply into the spinal canal. PAA, posterior arch of the atlas.

Computed tomography (CT) showed malformation of PAA with failure of posterior fusion of both laminae with a midline cartilaginous cleft at the site of the posterior tubercle (*Figures 2,3*). Bilateral hemiarches were abnormally incurved into the spinal canal, losing their normal smooth rounded contour; together with hypertrophy of posterior midline cartilaginous fusion defect, this caused severe spinal canal stenosis at the level of atlas. The mid-sagittal spinal canal diameter at the level of atlas was 6.36 mm. The odontoid process was well formed and the lateral masses of atlas were normal, indicating a pure posterior compressive canal stenosis.

Magnetic resonance imaging (MRI) showed posterior

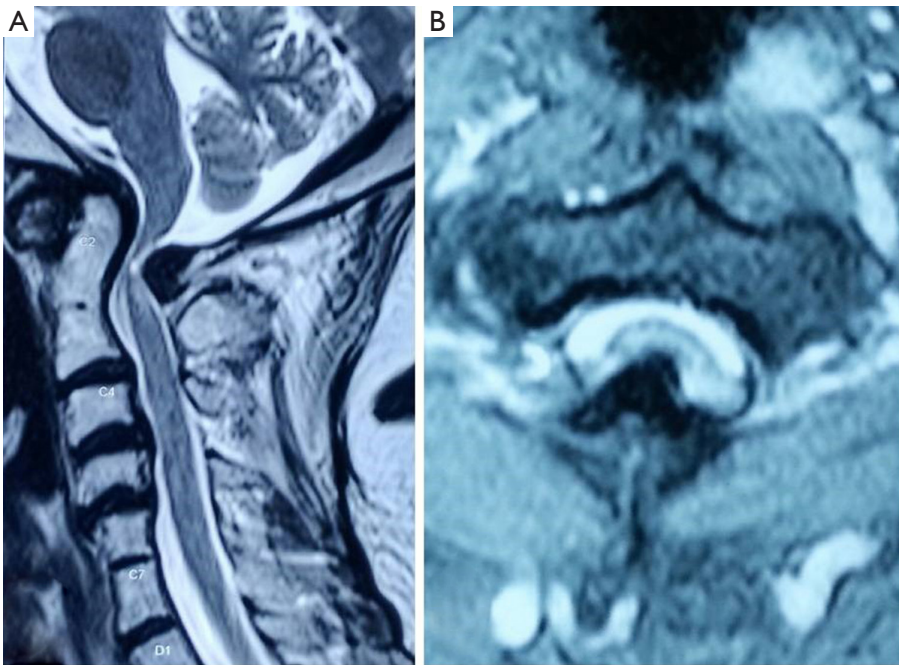

Figure 4 (A) T2W mid-sagittal and (B). Axial view MRI shows severe cord compression due to posterior mass, sharply kinking onto the cord. Note the hypointense signal intensity of the posterior mass is similar to the disc spaces, without any bony marrow, suggesting fibro-cartilaginous tissue. Hyperintense spinal cord signal changes suggestive of cord compression and myelopathy. T2W, T2-weighted; MRI, magnetic resonance imaging.

cord compression due to the anomalous arch with sharp kinking of the cord at the atlas with >50% reduction in the space available for cord (SAC) (*Figure 4*). Axial images revealed a sharp midline hypertrophic mass of the posterior tubercle of the atlas critically indenting into and compressing the spinal cord. The midline compressive mass was hypointense on T2-weighted images without any marrow enhancement seen, with similar intensity as the intervertebral discs, suggestive of a non-osseous fibro-cartilaginous tissue.

En-bloc excision of the anomalous arch with occipito-cervical fusion was performed via posterior midline approach using occipital 'T' plate, lateral mass screw fixation in C3 and C4 and autologous iliac crest bone grafting (*Figure 5*). The excised arch was noted to be abnormally curved inwards into the canal, with loss of smooth rounded contour, tightly compressing onto the cord (*Figure 6*). Meticulous wide dissection was performed under microscopic visualization to excise the arch en-bloc Post-excision the cord was noted to be well pulsatile without any evidence of dural injury.

Post-operatively, the patient had improvement in neck pain and myelo-radicular symptoms, which gradually disappeared over next 3 months. The gait improved and neurology returned to normal at 12 months. CT scan and MRI done at 20 months show adequate canal decompression, the implants holding well *in situ* with a well formed posterior bilateral lateral fusion mass (*Figures 7,8*). At 24 months follow-up, the patient showed an excellent clinical outcome without any obvious complaints or disability.

Discussion

During embryogenesis, the body of the atlas vertebra develops from the primitive fourth occipital and first cervical sclerotomes (3,5). Seventh week of intrauterine life sees the emergence of three primary centres of ossification, one forming the anterior arch and two forming the lateral masses (8). The PAA ossifies perichondrally from the lateral mass ossification centres, extending dorsally towards the midline, being nearly fused at birth except for a few millimetres of cartilage in the midline posteriorly, which represents the posterior tubercle (9). Reported in 0.5–2%

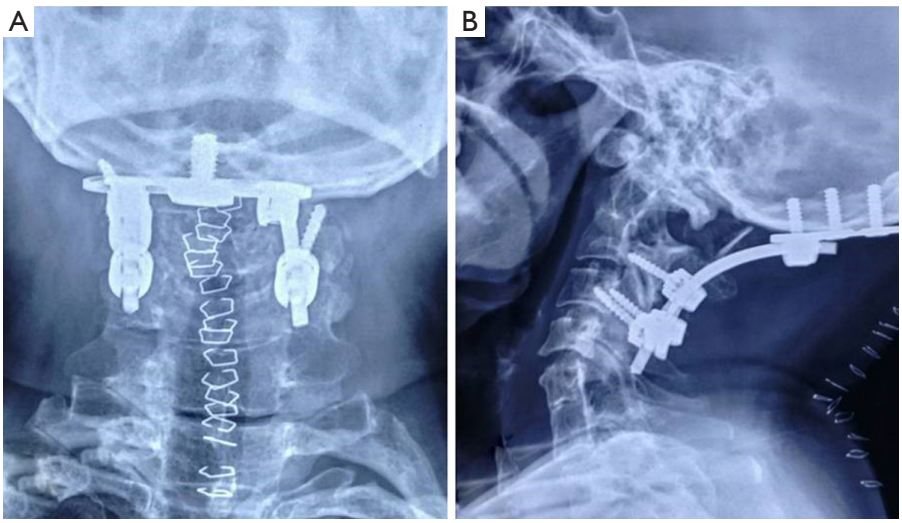

Figure 5 Immediate post-operative radiograph (A) anteroposterior and (B) lateral views, showing occipito-cervical fixation with occipital T plate and cervical lateral mass screws.

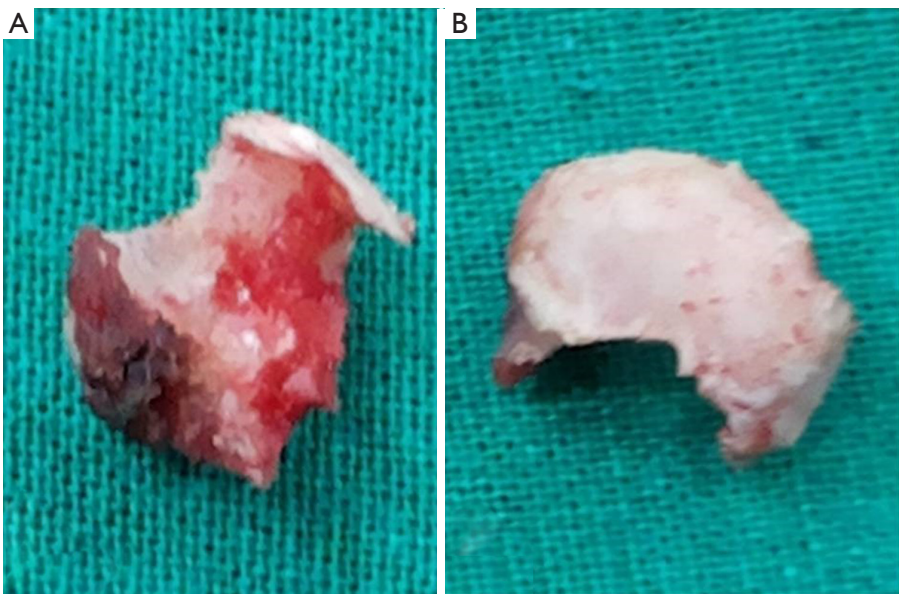

Figure 6 (A) and (B): the excised anomalous arch of atlas showing sharply angulated contour with outer smooth surface.

of the population, an additional center may be present posteriorly in the midline, forming the posterior tubercle of the atlas during the second year of life (8-10). Complete fusion of the PAA is expected to occur around 5 years of age (range 3–13 years) (3,5,6).

Congenital anomalies of PAA have a wide spectrum of presentation, ranging from failure of posterior midline fusion with a cartilaginous cleft to complete aplasia of PAA. Incidence of PAA defects has been reported between 0.69% to 4% (2,3,8,11-13). Currarino (14) classified these defects into five types, as shown in *Table 1*. The present case represents a Currarino type A defect of failure of posterior midline fusion of two anomalous hemi-arches, with a hypertrophied cartilaginous mass at the posterior tubercle. Type A defect is reported to be the most common type (90%) of congenital posterior arch defect (5,8,14). The

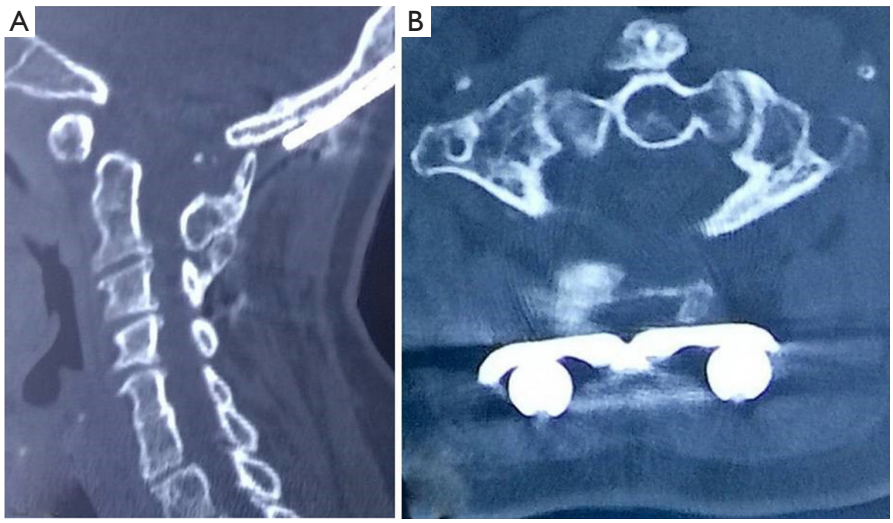

Figure 7 Twenty months post-operative CT (A) sagittal and (B) axial image, showing the excision of the compressive mass, wide laminectomy defect with adequate spinal decompression and restored SAC, with posterior implant *in situ*. CT, computed tomography; SAC, space available for cord.

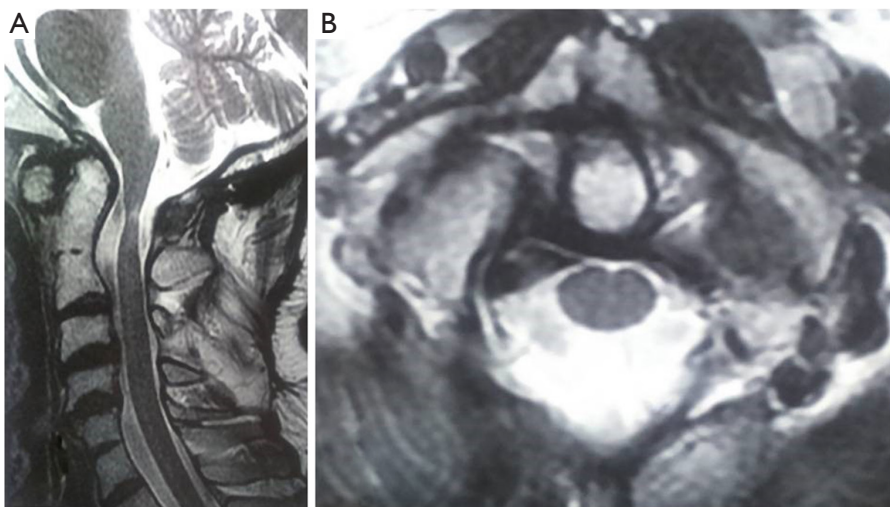

Figure 8 Twenty months post-operative T2W magnetic resonance images, (A) sagittal and (B) axial view, showing good cord expansion with smooth CSF flow around the cord, with patchy residual cord signal intensity changes seen.T2W, T2-weighted; CSF, cerebrospinal fluid.

anomalous posterior tubercle represents either an unfused persistent cartilaginous cleft of the bilateral hemilaminae or a hypertrophic cartilaginous remnant of the posterior tubercle ossification centre. The present case remains distinct as the midline anomalous osseo-cartilaginous mass was severely hypertrophied to cause spinal canal stenosis at the level of atlas causing cervical myelopathy due to the malformation.

Cervical stenotic myelopathy is a common presentation in the subaxial cervical spine involvement (2,15). The mid-sagittal canal diameter at the level of atlas in the present case was 6.36 mm, whilst the normal sagittal diameter at the level of atlas is reported to be 16–25 mm (2,3,16). While significant cervical cord compression occurs if the sagittal canal diameter is <14 mm at the level of atlas; myelopathic signs and symptoms develop when the diameter falls <10 mm (17,18). Such a degree of severe stenosis, as seen in the present case, predisposes the patient to a progressive

Table 1 Classification of congenital anomalies of the posterior arch of the atlas according to Currarino *et al.* (14)

Type	Definition
A	Failure of posterior midline fusion of the two hemi-arches
B	Unilateral cleft
C	Bilateral cleft
D	Absence of the posterior arch with persistent posterior tubercle
E	Absence of the entire arch including posterior tubercle

myelopathy as well as a high risk of severe spinal cord injury even after trivial trauma (12,17).

The patient presented with pain radiating to one half of the body with tingling and numbness in the same half with upper motor neuron lesion signs in all four limbs. Almost a third of the patients with PAA defects are reported to remain asymptomatic (6); in the symptomatic patients, the presentation varies from neck pain and occipitalgia (9), to neurological paraesthesia and frank myelopathy (5,6,11) and to extremely crippling reports of progressively deteriorating quadriparesis with respiratory distress (2). Although the arch defect is of congenital origin, manifestation is usually delayed into adulthood. Degenerative spondylotic changes occurring with age, at the level of the atlas, are thought to precipitate the manifestation of neural compression (17,19). Despite the lack of a specific history of injury in the past, trauma plays an important causative role in the pathogenesis of neurological complications. Microtrauma to the malformed PAA causing periosteal reaction and hypertrophy of the cartilaginous fusion defects is postulated to be responsible for progressive hypertrophy of the PAA (4).

Stenosis at the level of the atlas is reported in literature (*Table 2*) as isolated case reports, with the reported causes varying from clefts or aplasias of anterior or posterior arches of atlas, os odontoideum, lateral mass hypertrophy, ossification of transverse atlantal ligament and hypertrophy of the dens, either alone or in combination (2-4,12,14,17). Das *et al.* reported cervical myelopathy due to unilateral bony lateral mass hypertrophy, with hypoplastic posterior hemiarch of atlas, a Currarino type B defect, treated with transoral + posterior decompression and occipito-cervical fusion (3). They noted that even in the absence of complete arch formation or failure of fusion, there remains a fibrous constricting band in place of the actual arch, which needs to be divided during posterior decompression (3). The posterior midline hypertrophied mass, seen in our patient, represents such a fibro-cartilaginous hypertrophied tissue in place of the actual PAA which failed to fuse in the midline during the course of perichondral ossification. Kasliwal *et al.* reported a case of cervical myelopathy secondary to unilateral PAA hypertrophy, which was treated with surgical decompression (4). The present case differs primarily in the fact that the hypertrophy was at the posterior failure of fusion defect, causing a midline compression of the cord, in contrast to the unilateral arch hypertrophy in the case reported by Kasliwal *et al.*

Bhattacharjee *et al.* reported complete neurological recovery following posterior surgical decompression in acute onset spinal stenosis secondary to PAA fusion defect with a bony bar (2). Intra-operatively, they found an abnormally incurved arch into the spinal canal, severely compressing the spinal cord (2). Gross examination of excised mass, in the present case, had similar characteristics of an abnormally inward contoured arch with narrowed antero-posterior sagittal diameter in addition to the midline cartilaginous hypertrophy of the posterior arch fusion defect. This represents developmental canal stenosis at the level of atlas due to inadequate posterior migration of the arches, causing antero-posterior narrowing of the canal, thereby reducing the SAC significantly. The severity of the developmental stenosis depends on the degree of hypoplasia of the PAA, which in turn is decided by the amount of dorsal extension of the arch arising from the two lateral ossification centres during embryogenesis (17). Thus, an accurate pre-operative CT scan evaluation of the characteristics of the anomalous arch is essential to ensure careful dissection and adequate decompression while avoiding inadvertent iatrogenic neurological or dural injury in using instrumentation in a developmentally stenotic spinal segment. Occipito-cervical fixation was done to achieve fusion as the anomalous curved PAA needed a wide laminectomy to achieve complete decompression and the dissection involved stripping of suboccipital stabilizing ligamentous attachments to C2 spinous process; both factors entail a risk of post-operative

Table 2 Summary of patients with developmental canal stenosis at the level of atlas with cervical myelopathy reported in literature

Author	Age	Sex	Clinical findings	Radiological findings	Treatment	Results
Bhattacharjee *et al.* (2)	10	Male	Progressive spastic quadriparesis (left > right) with respiratory distress following trivial fall	Fusion defect of PAA with bony bar with cervical stenosis	Surgical decompression with anomalous arch excision	Immediate improvement in respiratory function, complete recovery by 1 year
Kasliwal *et al.* (4)	26	Female	Posterior cervical headaches, intermittent tingling and numbness involving Right arm, trunk and leg. Subtle right sided hemiparesis, exaggerated DTR's, Hoffmann's sign positive on right side	Right posterior C1 arch lesion with cord compression	Posterior decompressive C1 hemi-arch excision	Good clinical outcome at 3 months follow-up
Phan *et al.* (19)	80	Male	Bilateral intrinsic hand muscle weakness, hypertonia, hyperreflexia, Hoffmann's sign positive, spastic gait	Hypoplastic C1 arch with significant spinal canal stenosis	C1 arch excision with partial C2 laminectomy with foramen magnum decompression	Subtle neurological improvement with improved gait
	75	Male	Spastic gait, hyperreflexia, motor weakness in all 4 limbs, urinary disturbance, bilateral C4 hypoesthesia, Hoffmann's sign positive	Hypoplastic C1 arch with canal stenosis at the level of atlas	Posterior decompressive C1 arch excision	Motor improvement in all 4 limbs, bladder control regained, able to walk without assistance
Hsu *et al.* (17)	38	Male	Tingling in abdomen and perineum on flexing his neck, bilateral hand numbness for 2 months, DTR's 3+ in UL and 4+ in LL, no motor deficit	Incurving of PAA with stenosis at the level of atlas, with a dural sac diameter of 6.23 mm	Posterior arch excision with duraplasty	Improvement in the hand numbness and he tingling sensation in the abdomen and perineum
Sawada *et al.* (20)	38	Male	Weakness of right UL and both LL, bilateral LL spasticity, subtle sensory deficit	Hypoplastic atlas with spinal canal stenosis	Posterior decompressive laminectomy of the atlas	Clinical improvement wit resolution of weakness and spasticity
Komatsu *et al.* (21)	56	Male	Progressive gait disturbance, LL spasticity, motor weakness in all 4 limbs, hypertonia in all limbs, LL hyperreflexia, urinary difficulty	Hypoplasia of atlas with marked segmental compression of spinal cord	Posterior decompressive excision of PAA, C2 laminoplasty with foramen magnum decompression	Improved walking, resolution of urinary incontinence, recovery of power in bilateral UL
Okamoto *et al.* (22)	77	Male	All 4 limb paraesthesia, LL spasticity, exaggerated DTR, Hoffmann's sign positive, gait disturbance	Hypoplastic PAA with developmental canal stenosis at the level of atlas	Posterior decompressive C1 arch excision with occipito-cervical sublaminar wiring	Improvement in gait and fine movements
Present case	44	Female	Persistent neck pain radiating to Right half of body, tingling and numbness in right half of body, myelopathic gait, hyperreflexia in all 4 limbs, Hoffman's sign positive	Hypoplastic C1 arch with failure of posterior midline fusion with developmental canal stenosis, associated congenital C2- and C6–7 fused vertebrae	Posterior C1 arch excision with occipito-cervical fixation	Immediate improvement in myeloradicular symptoms, gait improved, normal neurology at 12 months

PAA, posterior arch of atlas; DTR, deep tendon reflexes; LL, lower limbs; UL, upper limbs.

atlanto-axial instability or occipto-cervical kyphosis. C1–C2 Magerl-type transarticular screw fixation could not be used due to anomalous fused C2–C3 vertebrae; C1 arch excision precluded the use of Brooks or Gallie's type sublaminar wire based fusion construct. Sabuncuoglu *et al.* extensively reviewed the literature on hypoplastic PAA and concluded that the excision of anomalous PAA is curative, and if the PAA anatomy is abnormal, the posterior fusion should involve the occiput and lower cervical segments (8).

In conclusion, posterior arch of atlas congenital defects are a complex constellation of abnormalities with abnormal arch morphology, midline cleft hypertrophy and arch fusion defect. Though infrequently symptomatic, these can present as cervical myelopathy; being aware of such an entity avoids diagnostic delays and treatment dilemmas as the stenotic segments are prone to severe deficit even with trivial trauma. Posterior laminectomy is effective for cord decompression, and fusion is indicated in cases with potential post-operative instability or in presence of other congenital defects.

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None.

Footnote

Conflicts of Interest: The authors have no conflicts of interest to declare.

Informed Consent: Informed consent was obtained from the individual participant included in the study for the purpose of publication.

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