Childs Nerv Syst
DOI 10.1007/s00381-014-2439-y

CASE REPORT

Spinal arteriovenous metameric syndrome in a neonate presenting with congestive heart failure: case report

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Received: 11 February 2014 / Accepted: 10 May 2014

Abstract
Background Spinal arteriovenous metameric syndrome (SAMS) is a combination of more than two separate vascular malformations in the same embryonic metameres. This syndrome, also known as Cobb syndrome, is rare, especially in the neonate.
Case description A neonatal girl with a birthmark in the occipital and posterior nuchal regions presented with severe heart failure on the day of birth. The large arteriovenous fistulas in the left hypoglossal canal and in the posterior nuchal region were embolized with detachable coils on the postnatal days 5 and 18, which improved heart failure markedly. The associated intramuscular arteriovenous malformation in the posterior neck was left untreated because large arteriovenous fistulas had been occluded. She grew up without any neurological deficits and developed with normal milestones until the latest follow-up of 8 years old.
Conclusion To our knowledge, this is the first case with SAMS in a neonate presenting with congestive heart failure. Presence of a birthmark in a neonate presenting with congestive heart failure may suggest the possible underlying high-flow vascular malformations in the same metamere.

Keywords Cobb syndrome · Heart failure · Neonate · Spinal arteriovenous metameric syndrome

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Introduction

Spinal arteriovenous metameric syndrome (SAMS), also known as Cobb syndrome, is a rare embryonic metameric syndrome, in which cutaneous, muscular, and/or bony vascular lesions as well as paraspinal and/or spinal vascular lesions are found in the same metameres. Less than 80 cases with this syndrome have been reported in the literature [1, 2, 4, 7–10, 12–16]. Classically, a birthmark (a cutaneous veno-capillary malformation) and a spinal lesion [arteriovenous malformation (AVM) or arteriovenous fistula (AVF)] are found in the same or neighboring metameres [6]. However, conceptually, it is not necessarily required to have both cutaneous and spinal vascular malformations. Any combinations of vascular malformations in the same metameres are possible. Their symptoms are usually related to the spinal vascular lesions, i.e., pain or sudden/progressive neurological deficits due to hemorrhage, mass effect, or ischemia (venous hypertension or steal phenomenon). To our knowledge, this syndrome had not presented with congestive heart failure in the neonatal period. We report a neonate who was initially diagnosed as having a high-flow hemangioma but finally proved to be multiple AVM/AVFs in the craniocervical and cervical regions in the same metameres.

Case presentation

A neonatal girl was the first product of the apparently healthy parents. Pregnancy was uneventful. The patient was delivered by caesarian section at the gestational period of 40 weeks and 0 day because of fetal distress. Birth weight was 3,082 g, and the Apgar scores were 8 and 9 at 1 and 5 min after birth, respectively. The birthmark with a marked swelling in the occipital and posterior nuchal regions was apparent from birth, and it was warm and pulsating (Fig. 1). Vascular bruits

Published online: 21 May 2014

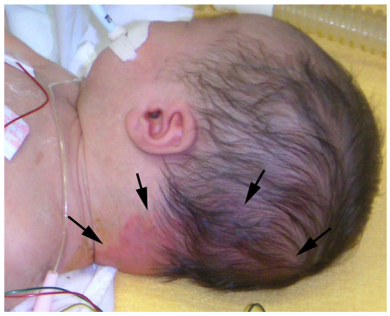

Fig. 1 Photograph of the occipital and posterior nuchal pulsating mass on the postnatal day 4. *Arrows* indicate the margin of the birthmark and swelling

were audible around the birthmark. Motor weakness was not apparent, and bladder and bowel functions seemed to be normal. Family history was not remarkable.

Congestive heart failure was present from birth. Chest X-ray showed an enlarged heart with a cardiothoracic ratio of 65 %. Contrast-enhanced CT on postnatal day 3 showed the enhanced mass in the occipital and posterior nuchal regions, indicating a vascular-rich mass (Fig. 2). There was no intracranial abnormality on CT. This patient was transferred to us on day 4 for the treatment of heart failure caused by presumed high-flow "hemangioma."

On day 5, this patient underwent a diagnostic angiography, which was followed by transarterial embolization. Under general anesthesia, control angiography was performed using a 4-French diagnostic catheter. Full understanding of the angioarchitecture of the lesions was difficult at the first intervention because of the restricted volume of the contrast and angiographic runs due to a compromised renal function. Angiography showed direct AVFs in the left hypoglossal canal, in the left high cervical epidural region, and in the posterior nuchal region. There was no intradural AVM (Figs. 3 and 4). In addition, there were intense muscular stains due to nuchal intramuscular AVM. Staged reduction of blood flow in the AVFs was intended. First, a microcatheter was advanced to the

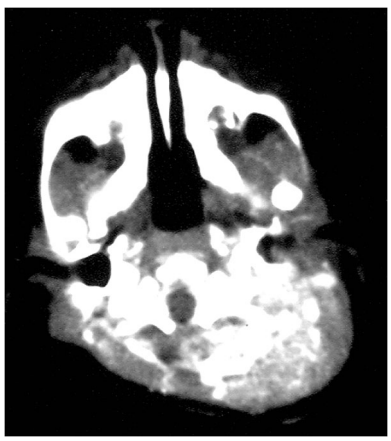

Fig. 2 Contrast-enhanced CT on day 2 shows a large enhancing mass in the left occipital and nuchal regions

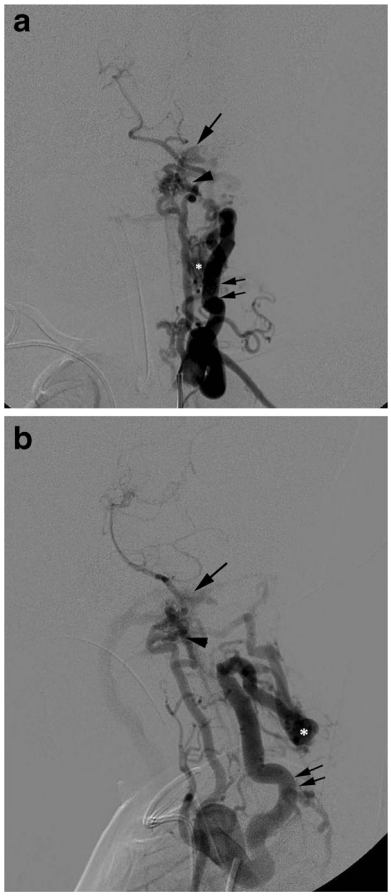

Fig. 3 Left subclavian arteriogram (early arterial phase) (**a** left anterior oblique view, **b** lateral view) showing the direct arteriovenous fistula in the left hypoglossal canal (*single arrow*) and at the C2 epidural space (*arrowhead*). *Double arrows* indicate the large left deep cervical artery. *Asterisk* indicates the shunt point of the posterior nuchal arteriovenous fistula

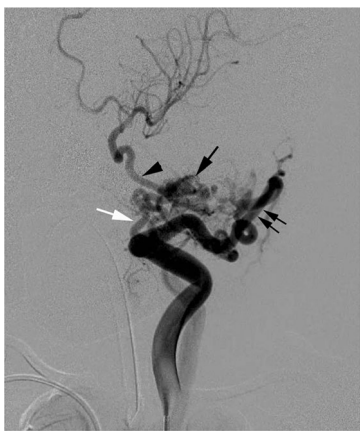

Fig. 4 Left common carotid injection (left anterior oblique view) shows the ascending pharyngeal artery (*white arrow*) supplying the arteriovenous fistula in the left hypoglossal canal (*single black arrow*). Occipital artery (*double black arrows*) supplies the arteriovenous fistula as well as the nuchal muscular arteriovenous malformation. Note the small left internal carotid artery (*arrowhead*) in comparison to the large occipital artery as well as the ascending pharyngeal artery

left anterior meningeal artery (odontoid artery) originating from the left vertebral artery at the C2/3 vertebral level. The tip of the microcatheter was further advanced into the left anterior condylar vein in the left hypoglossal canal. Transarterial embolization using detachable platinum coils was performed to the AVF at the left anterior condylar vein. Then, the microcatheter was introduced into the left anterior condylar vein through the hypoglossal branch of the left ascending pharyngeal artery. Again, coil embolization of the AVF in the anterior condylar vein was performed. The exact location of the AVF in the posterior neck could not be determined in the first session. This embolization controlled heart failure for about 2 weeks. The second embolization was required due to deterioration of heart failure.

On day 18, the patient underwent the second transarterial coil embolization for the posterior mid-cervical AVF, which was located within the cervical muscles and was fed by the left deep cervical artery (Figs. 5 and 6). This embolization resulted in marked improvement of heart failure. The patient was discharged on day 30 with a moderately enlarged heart due to the residual small AVFs. A low dose of diuretics to control mild cardiac overload was prescribed until the age of 2.5 years old. This patient grew normally without any focal neurological deficits. At the age of 4, follow-up diagnostic angiography showed no large remaining AVFs, but there were still a small epidural AVF fed by the C2 segmental artery of the left vertebral artery and intramuscular AVMs mainly fed by the left deep cervical artery and left occipital artery (Fig. 7). MR angiography at the age of 7 showed enlarged left deep cervical and occipital arteries feeding the intramuscular AVMs (Fig. 8). At the latest follow-up at the age of 8, she was neurologically normal with normal developmental milestones. No cervical bruits were audible. No cardiac overload was observed. Cardiothoracic ratio on X-ray film was 50 %. There remained a port-wine stain at the occipital and posterior nuchal region without swelling.

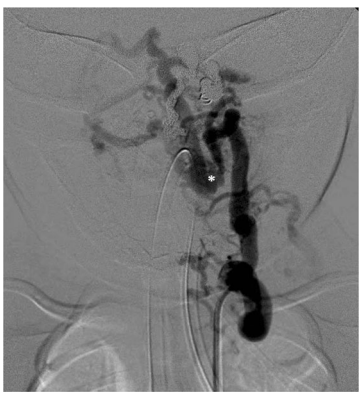

Fig. 5 Left deep cervical artery injection (frontal view) after the coil embolization of the arteriovenous fistula in the hypoglossal canal demonstrating the arteriovenous fistula in the posterior nuchal region. *Asterisk* indicates the shunt point

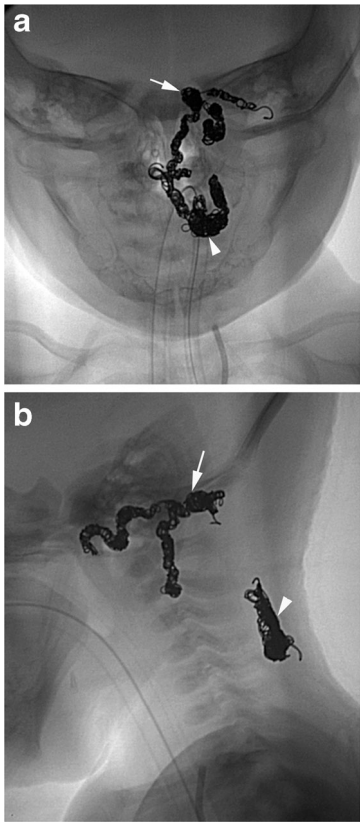

Fig. 6 Plain X-ray film (**a** frontal view, **b** lateral view) shows the deposited coils in the left hypoglossal canal (*arrows*) as well as the feeding arteries and in the posterior nuchal region (*arrowheads*)

Discussion

In the process of vasculogenesis, endothelial cells derive from mesoderm, and the tunica media derives from the neural crest cells or mesodermal cells. These cells corporate to form vessels in the given metameres after migration to their own target regions. When embryological adverse events occur before the migration, more than two separate vascular malformations in the same metamere could be formed. This explains the pathogenesis of SAMS.

SAMS was first described in 1915 by Stanly Cobb, who was a resident of Harvey Cushing at that time [2]. Thus, this syndrome has long been called Cobb syndrome. Original description of Cobb syndrome was the association of the spinal and cutaneous angiomas. Cutaneous lesions are occasionally not obvious on usual inspection but could be detected after special maneuvers including stroking and warming [13]. There was no distinct definition of "angioma," which may include capillary and venous malformations, AVM, AVF, and their combination. Since many of the reported cases were

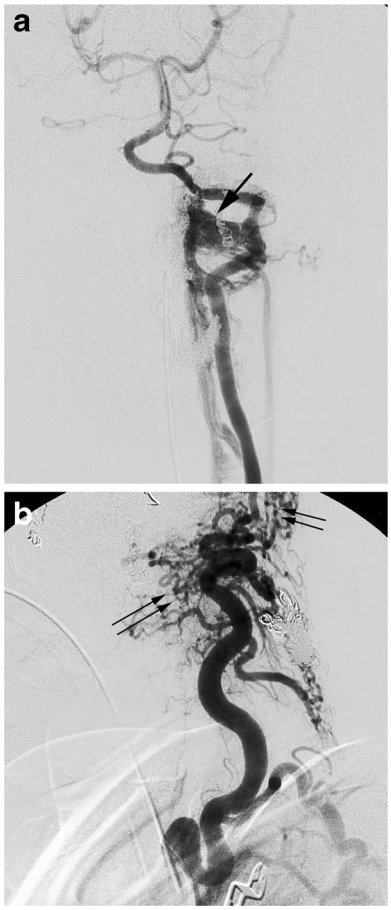

Fig. 7 Follow-up angiography at the age of 4. Left vertebral injection (**a** frontal view) shows the remaining epidural arteriovenous fistula (*arrow*) at C2 level. Left deep cervical injection (**b** lateral view) shows the remaining muscular arteriovenous malformations (*double arrows*)

diagnosed at the time when spinal angiography and magnetic resonance imaging were unavailable, their true pathologies of the lesions were not always evident.

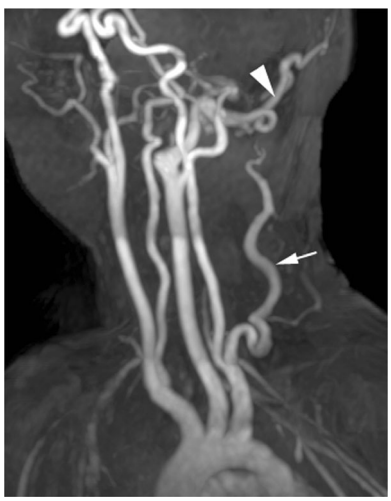

Fig. 8 Follow-up MR angiography at the age of 7 showing the left occipital artery (*arrowhead*) as well as left deep cervical artery (*arrow*), supplying the nuchal muscular arteriovenous malformation

There have been several reports on SAMS in infants [1, 4, 10, 16], but none presented with a congestive heart failure. Congenital cerebrovascular diseases, which present with congestive heart failure in the neonatal period, include the congenital AVFs in the brain, such as the vein of Galen aneurysm and dural sinus malformation as well as less frequently pial AVF [7]. Some infantile hemangioma in the proliferating phase presents with heart failure in the neonatal period because of high blood flow in the tumor. Hepatic hemangiomas are frequently reported to cause heart failure in the neonates [5]. Thus, when the neonate presents with severe heart failure, one should consider these cerebral vascular malformations and systemic vascular lesions in addition to the congenital heart diseases. Birthmark may suggest the associated vascular malformations and prompts to seek for such underlying high-flow AVM/AVFs.

For the diagnosis of SAMS, CT/CT angiography and MR imaging/MR angiography are the first-line diagnostic tools because of their inherent less invasiveness, but the selective catheter angiography is required to define the nature and angioarchitecture of the spinal lesions, which may be followed by the interventional procedures. Differentiation between the AVM/AVFs and infantile hemangiomas is crucial because treatment is different [11]. The former requires the occlusion of the shunt points or nidi, but the latter has a nature of spontaneous involution with time or requires flow reduction by steroid or propranolol treatment and/or embolization only in the selected patients [15].

Niimi et al. [12] reported the largest series of SAMS, in which 28 SAMS patients (19 %) were included among 148 patients with spinal cord AVMs. They were 24 nidus-type AVMs and 4 intradural AVFs. Characteristics of their SAMS were nidus-type lesions, female dominance (M:F=1:2.5), and younger age in comparison to non-SAMS patients with sporadic spinal cord AVMs. The lesions were almost evenly distributed along the spinal column. Their clinical manifestations were intradural hemorrhage (79 %, either subarachnoid hemorrhage or hematomyelia) in most cases. Interestingly, male dominance (M:F=2:1) was reported in SAMS by Rodesch et al. [14] in 2002.

The case presented here was unique in that SAMS is associated with congestive heart failure in the neonate, and the AVFs were located at the craniocervical junction (in the hypoglossal canal and high cervical epidural space at C2 level) and within the posterior neck. In adults, AVF in the hypoglossal canal usually presents with intractable tinnitus and less frequently hypoglossal palsy [3]. Transvenous coil embolization is a treatment of choice for the AVF in this location in adults. Our case is the first and youngest one, which presented with an AVF in this location in a neonatal period. Coil embolization in our patient did not cause a procedural complication of hypoglossal palsy.

In conclusion, a birthmark in a neonate with congestive heart failure may suggest the underlying high-flow AVF in the

same metamere, i.e., SAMS. This prompts further neuroradiological examination for it.

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