CASE REPORT

Multiple anomalies in the origin and course of vertebral arteries and aberrant right subclavian artery in a child with moyamoya syndrome

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Accepted 30 November 2017

SUMMARY

Here we report, for the first time, a combination of five-vessel aortic arch, anomalous origin of the right vertebral artery (VA) from the common carotid artery (CCA), an aberrant right subclavian artery (SCA), and bilateral symmetrical segmental agenesis of VAs. In this case report, we present a patient with moyamoya syndrome (MMS) and Down syndrome (DS) who has bilateral symmetrical segmental agenesis of VAs, left VA originating from aortic arch and anomalous origin of right VA arising from CCA in combination with an aberrant right SCA. Therefore, five vessels are originating from aortic arch. Here, we report, for the first time, a combination of five-vessel aortic arch with an aberrant right SCA and symmetrical segmental agenesis of both VAs. The possible embryological mechanisms of the anomalies as well as an relation with MMS and DS are discussed.

BACKGROUND

Possible variations in the origin of vertebral artery (VA) are well documented and analyzed in the literature.¹ However, common carotid artery (CCA) origin of the VA, combined with symmetrical bilateral segmental absence of VA and aberrant right subclavian artery (ARSCA) in the context of Moyamoya syndrome (MMS) without

accompanying segmental agenesis of the internal carotid arteries (ICAs) has not been reported yet.

CASE PRESENTATION

A 3-year-old girl with Down syndrome (DS) was admitted to a regional hospital with focal seizure and left arm weakness. Neurological examination showed left facial and arm paresis. The brain MRI showed an acute infarction in the right middle cerebral artery territory. MR angiography revealed high-grade stenosis at the cavernous segment of the left ICA and at the proximal V3 segment of both VAs. The patient was referred to our MM Center to clarify possible MMS.

INVESTIGATIONS

Cerebral angiography showed an occlusion of the left ICA distal to the ophthalmic artery origin with moyamoya (MM)-type vascular network in this area and significant stenosis at the supraclinoid and cavernous segment of the right ICA (figure 1).

In addition to these findings suggesting MM vasculopathy, the angiogram revealed the absence of the brachiocephalic trunk and separate origin of the right subclavian artery (SCA) and the right CCA from the aortic arch. The right SCA was originating just distal to the left SCA, with the left VA originating directly from the aortic arch between the left CCA and the left SCA (figure 2), whereas the right VA was originating from the proximal CCA (figure 3).

Selective left and right VA injections showed symmetrical short segment absence of proximal V3 segments with collateralization through a rete-type network, as well as through the deep and ascending cervical arteries and also anterior–posterior spinal arteries (figure 4).

DISCUSSION

Complex embryological evolution of VA, including fusion and partial regression of metameric arteries, accounts for the wide range of possible variations. The most common variant is direct aortic arch origin of the left VA.¹ Bilateral anomalous origin of the VA is uncommonly seen with a prevalence of 2.8%.¹ Tsai *et al* reported increased incidence of anomalous right VA origin in patients with anomalous origin of the left VA and ARSCA. It has been reported that concomitant appearance of anomalous origin of both VAs, with right VA originating

Figure 1 Angiography of right and left internal carotid arteries (ICAs) in lateral view showing stenosis at the supraclinoid and cavernous segment of the right ICA and occlusion of the left ICA distal to the ophthalmic artery origin with moyamoya vessels.

To cite: Beyaz P, Khan N, Baltasvias G. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-013464

Figure 2 (A and C) Angiography of aortic arch showing an ARSCA, which originates from the distal aortic arch. (B) Direct aortic arch origin of left vertebral artery (VA). (C) Note the absence of right VA in the right subclavian artery injection.

from the CCA and left VA originating from aortic arch, as in our case, is even rarer in patients with ARSCA.² The VA is established secondary to the process of longitudinal anastomosis (intersegmental arteries) of the cervical segmental arteries (SAs) and obliteration of the medial parts of these arteries. Normally, the first to the sixth cervical intersegmental arteries (CIAs) develop into the VA and the seventh SA makes the SCA. If longitudinal

Figure 3 Right common carotid artery (CCA) injection in lateral view demonstrating the right vertebral artery (thin arrow) originating from the proximal CCA (thick arrow).

Figure 4 (A and B) Selective left and right vertebral artery (VA) injections showing symmetrical short segment absence of V3 segment with rete-type network. (B, C, and D) Note also the reconstruction of right distal VA through deep cervical, ascending cervical, and anterior and posterior spinal arteries.

anastomosis of the right CIA stops between the sixth and seventh SAs, and the right-sided dorsal aorta is obliterated proximal to the seventh SA, then the right-sided SCA originates from the left-sided aorta distal to the left SCA and therefore the right VA originates from the right CCA.³

Brouwer *et al*⁴ convincingly discussed the possible mechanism of bilateral anomaly of VA origin, including the CCA origin, with ARSCA. They made a distinction in the description of SA as medial and lateral. In case of persistence of the lateral SA, the VA would originate from the cervical arteries, whereas in case of persistence of the medial SA, the VA would originate from the right dorsal aorta, the future proximal CCA.

Extracranial VA involvement is not typically expected in the course of MM disease; however, we have four anecdotal cases of MMS displaying segmental absences of extracranial VAs, with two of these showing symmetrical and bilateral absence with a well-developed collateralization as seen in our case. This raises the question of potential occurrence in embryonic stages as part of the MM vasculopathy or associated syndrome (agenesis) versus being rather an expression of the MM progressive vasculopathy alone (occlusion). In our case, this question cannot be answered with certainty; however, symmetry, bilaterality and

lack of progression point rather to a long-standing occurrence and therefore more to the scenario of agenesis.

Prevalence of MMS in patients with DS is higher than in general population.⁵ A potential etiological–pathogenetic link between the two entities or a common underlying chromosomal abnormality has been proposed.⁵ Mishra *et al*⁶ reported a case with a very similar arterial disposition but without segmental absence of the VA and same clinical background, MMS with associated DS, that they named MM “disease”. In the current report, we preferred the term MMS based on the association with another medical condition (DS).⁷ The fact that the two cases shared the same background shows that DS might play a significant role in the incidence of these arterial variations, an assumption further supported by the known predisposition for vascular dysplasia of patients with DS.⁸ On the other hand, Akira *et al* have reported that in MM disease there is a high frequency of associated cerebral arterial variations mostly in the posterior circulation.⁹ However, no distinction was made between MM disease and MMS and one cannot exclude that

some of the included cases were not MMS with an associated DS or another disease.

Contributors PB: identified the case, performed literature review, interpreted and analysed angiographic images and wrote the manuscript. GB had substantial contributions to the design of the work, critically revised the draft paper, final approval of the paper to be published and was scientific advisor. NK had contributions to the design of the work, revised the paper, maintained patient’s consent and ethics approval.

Competing interests None declared.

Patient consent Obtained.

Ethics approval Informed consent for publication was obtained in accordance to our ethics committee review board approval (BASEC no. 2016-00880).

Provenance and peer review Not commissioned; externally peer reviewed.

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Learning points

- Here, we report an extremely rare combination of arterial anomalies in association with moyamoya syndrome (MMS) and Down syndrome in a child, pointing at a potential embryological basis.
- This case shows arterial anomalies are not rare in the course of MMS and in order to identify the relationship between arterial variations, MMS and various associated diseases, we need to focus on mechanisms of embryological maldevelopment and more data are needed as supportive evidence.

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