Imaging Findings of a Patient with Incomplete Phenotypical Expression of the PHACES Syndrome

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Summary

We present imaging findings of a patient with an incomplete form of the PHACES syndrome with dolichosegmental intracranial arteries as the predominant component, and discuss the etiopathological and clinical significance of this finding.

Introduction

PHACES syndrome, once considered a very rare congenital disorder with around 150 cases presented to date, has recently become a popular syndrome with a surge of papers in recent years describing its various aspects.

The name of the syndrome is an acronym derived from various findings which may include posterior fossa malformations, hemangiomas, cardiac defects and coarctation of aorta, eye abnormalities and sternal and abdominal raphe defects¹⁻⁶. Recently a specific angiographic feature: dolichoectatic dilatation with tortuosity of intracranial arteries was first described in detail by Baccin et Al in two patients with PHACES syndrome⁷.

We present a case without most of the phenotypical aspects of this syndrome but vascular involvement predominant features which include dolichoectasia of the right supraclinoid internal carotid artery (ICA) and M1 segment of middle cerebral artery (MCA).

Case Presentation

A ten-year-old girl with a nonspecific medical history and family history presented with progressing lower extremity pains. Cranial MR and MRA demonstrated a persistent trigeminal artery on the right side with elongation and complex coiling of the right supraclinoid ICA and M1 segment of the MCA. A drainage vein that may have been consistent with an AVM was absent. A1 segment of the right anterior cerebral artery was absent as well. Another interesting finding was the presence of dilated perivascular spaces in the right basal ganglia. The patient lacked any posterior fossa or cerebral malformation, with a normal corpus callosum (figure 1). A right-sided arcus aorta with an aberrant left subclavian artery was seen on thorax CT (figure 2). Kommerell's diverticulum as a remnant of the left arcus aorta was also present. DSA confirmed all these findings (figure 3A-C). Though the patient seemed to be free of a facial hemangioma, on rigorous questioning, an operation history due to hemangioma was elucidated (figure 3D).

Discussion

The association of cervical hemangiomas with vascular and nonvascular intracranial malformations was first mentioned by Pascual-Castroviejo in 1978¹. The name "cutaneous he-

Figure 1 MRI and MRA of the patient. A) Transverse T2W image, arrows indicate bizarre vascular structures. B) Coronal T2W section, same pathology, different view (arrow). C) Another transverse T2W section for the depiction of dilated perivascular spaces on the right side (arrows). D) MRA, arrows indicate the complex coiling and dolichoectasia of the right supraclinoid ICA and M1 segment of MCA. More distal part of MCA and branches are completely normal.

mangioma-vascular complex” was proposed to the constellation of the findings by the same author in 1996². It was Frieden et Al who designated the acronym “PHACE” derived from the major findings: posterior fossa malformations, hemangiomas, arterial abnormalities, coarctation of the aorta and cardiac defects and eye anomalies³. Notification of stenoses of intracranial arteries by Burrows et Al and also association with sternal cleft in some cases prompted the appellation “PHACES syndrome”⁴. This new designation has been approved by the scientific community and is more widely used⁵.

Our case constitutes an interesting example due to the diversity of the clinical presentations of the PHACES syndrome and presumed to be an incomplete phenotypical expression. However, the patient lacks other significant features of the syndrome and does not meet the diagnostic criteria fully; especially complex tortuous intracranial arteries which shows a

close resemblance to the findings recently described in detail by Baccin et Al in two patients with PHACES and the history of a facial hemangioma prompted the diagnosis⁷. Their first case was complete in phenotypical expression and though the second case lacked posterior fossa malformations, a ventricular septal defect was present and was therefore considered phenotypically incomplete.

Our case is a milder form of the syndrome with some other findings accompanying the aforementioned abnormalities: a persistent trigeminal artery and a right sided arcus aorta with an aberrant course of the left subclavian artery.

Four different types of arterial anomalies have been associated with PHACES including: agenesis of cervical carotid arteries, persistent embryological vessels, arterial stenoses and dolichoectatic type intracranial arteries, the latter not being reviewed extensively in the literature⁷.

A special emphasis is mandatory with regard to the meaning of so-called dolichoectasia. Dolichoectasia means tortuous, elongated and dilated arteries and alternatively is often called dilatative arteriopathy. In adults dolichoectasia has been reported to be associated with various pathologies consistent with stroke, subarachnoid hemorrhage, cranial nerve palsies, mid-brain compression, and obstructive hydrocephalus^{8,9}. However in children it is usually asymptomatic. Dilatative arteriopathy can be classified into two types as: segmental and non-segmental. Nonsegmental arteriopathy indicates involvement of all intracranial arteries commonly due to atherosclerosis in adults and various inflammatory, genetic and degenerative processes in children and adolescents such as Marfan's disease, Ehlers-Danlos, AIDS, and sickle-cell disease. Segmental arteriopathy is the result of some vulnerability inherent to a specific portion of a vessel getting exposed to a generalized trigger⁷.

Segmental dilatative arteriopathy is the type seen in our patient can be considered secondary to segmental dysplasia.

In their paper, Baccin et Al proposed that different forms of arterial involvement may have been related to plurisegmental developmental anomaly of the neural crest involved in embryological dorsal aorta and cerebrofacial structures and timing of the revealing trigger may determine the type of resulting arterial anomaly⁷.

Although there is no clue to the timing of the insult and severity of the disease sometimes leading to incomplete expression, an interesting point related to our case is the presence of the vascular components without nonvascular components of the syndrome. It may raise the question of whether or not we are facing a variant form of the syndrome comprised of only vascular components or "vascular PHACE".

Lastly, since the dolichosegmental arteries in this syndrome are similar to AVM's on MRI when they are overlooked, correct diagnosis is crucial to avoid unnecessary attempts at treatment. These dolichoectatic vessels, as mentioned before, are considered segmental dysplasias and not to be associated with intracranial hemorrhage.

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