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Case Report

Transient ischemic attack in elderly patient with PHACE syndrome

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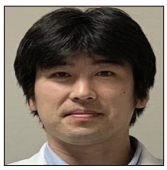

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ABSTRACT

Background: Posterior fossa brain malformations, hemangiomas, arterial anomalies, coarctation of the aorta and cardiac defects, and eye abnormalities (PHACE) is a rare congenital anomaly with a broad spectrum of clinical manifestations.

Case Description: We describe a 75-year-old male with PHACE anomaly, aortic anomaly, malformation of brain, aplastic right carotid artery, and cervical vasculopathy. He presented with a transient ischemic attack with the left hemiparesis, a rare clinical presentation of the PHACE syndrome. He had an uneventful recovery and recently completed a 2-year follow-up after the superficial temporal artery to middle cerebral artery anastomosis.

Conclusion: PHACE syndrome should be kept in mind, even in individuals of advanced age, in the instance of a TIA, especially in situations which may involve induced hypoperfusion.

Keywords: Aplasia of carotid artery, Dandy–Walker syndrome, PHACE syndrome, Transient ischemic attack

INTRODUCTION

PHACE is a neurocutaneous syndrome including malformations of the posterior fossa, facial hemangiomas, arterial anomalies, cardiac anomalies, aortic coarctation, and abnormalities of the eye.[4] All abnormalities are not necessarily present in each patient, and clinical presentation and course are variable. Arterial abnormalities usually involve cervical and cerebral vasculature. This progressive cerebral vasculopathy leads to increased risk for transient ischemic attack (TIA) in patients with PHACE.[3] Although the natural history of PHACE vascular anomalies remains unknown, their impact on long-term neurologic outcomes is particularly relevant, with up to 50% of patients becoming symptomatic due to progressive intracranial arteriopathy.[10] We describe an uncommon presentation of TIA in a 75-year-old male – a new clinical feature of rare adult PHACE syndrome.

CASE REPORT

A 75-year-old male presented with sudden onset of weakness in his left upper and lower extremities. Medical history confirmed that the patient was a full-term birth without trauma or history of hypoxia. Hemangioma on the right side of the neck was noted after birth. Routine laboratory examinations were unremarkable. Neuropsychological evaluation was normal.

Magnetic resonance imaging of the brain revealed multiple posterior fossa anomalies including agenesis of the cerebellar vermis with hydrocephalus and hypoplasia of the right cerebellar vermis. Accordingly, the patient was diagnosed with Dandy–Walker malformation [Figure 1]. Multiple complex cerebrovascular anomalies were noted as follows: narrowing of supraglenoid portion of the left internal carotid artery (ICA), bending of bilateral vertebral arteries, and narrowing of the basilar artery with calcification. Cerebral aneurysm was seen in the terminal portion of the left ICA [Figure 2a]. There was a complete absence of the right ICA from the origin with corresponding absence of the left bony carotid canal [Figures 2b and c].

Anomalous aortic arch anatomy was also seen. The left subclavian artery arose from the medial wall of the aortic arch, more proximal than expected. Bilateral common carotid arteries arose from the distal side of the left subclavian artery. The right subclavian artery had an aberrant course, with its proximal portion making a U-shaped bend [Figure 2d].

Single-photon emission computed tomography (SPECT) demonstrated reduced cerebral blood flow (CBF) in the border zones between the middle and posterior cerebral artery territories of the right hemisphere [Figure 3a]. We administered aspirin (100 mg/day) and edaravone (200 mg/day) for 2 weeks. The patient was ultimately referred for a superficial temporal artery to middle cerebral artery anastomosis. CBF of the right hemisphere was improved to normal level [Figure 3b]. He had an uneventful recovery and recently completed a 2-year follow-up. In subsequent examinations, no additional neurological events were reported.

DISCUSSION

PHACE syndrome was first reported in 1996. The acronym stands for a set of characteristic disorders that include posterior fossa malformations, hemangioma, arterial anomalies, coarctation of the aorta or cardiac defects, and eye abnormalities.[4] The diagnostic criteria for PHACE syndrome were published in 2009.[8] In our case, the aortic anomaly, right common carotid artery aplasia, cerebrovascular malformation, and Dandy–Walker syndrome were included as the major criteria. The etiology was not clear. PHACE syndrome shows a female-to-male predilection of 9:1 and has been associated with a mutation in an X-linked gene.[12] Genomic copy number variations have also been reported to occur in some chromosomes.[11] Cerebrovascular accidents in the pediatric population have a multifactor etiology, and patients with PHACE syndrome have several risk factors including possible vascular stenosis and occlusion with a subsequent reduction in blood flow and thromboembolism related to potential cardia and supra-aortic arterial lesions.[5] Hemangioma on the right neck at birth is reportedly associated with higher risk for cerebrovascular anomalies.[6] Arterial anomalies seem to be the most common noncutaneous abnormality in PHACE, occurring in as many as 57% of patients. A wide variety of congenital arterial anomalies is associated with PHACE, including persistent embryonic arteries, agenesis or hypoplasia of major arteries (such as the internal carotid and vertebral arteries), and "angiomatous" malformations of intracranial and extracranial blood vessels (such as aneurysmal dilatations or moyamoya phenomenon).[8] The neurologic

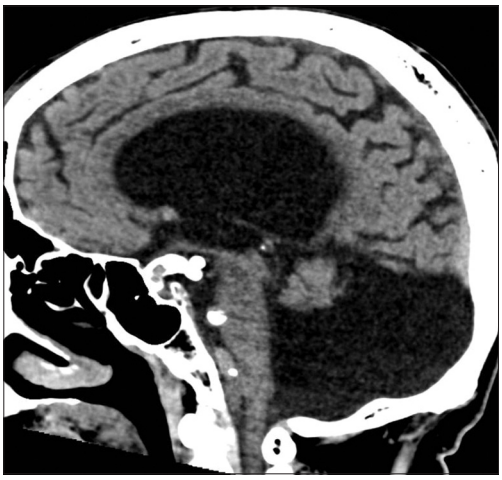

Figure 1: Sagittal computed tomography image of the brain demonstrates a posterior fossa anomaly with vermian hypoplasia, expansile arachnoid cyst, and hydrocephalus.

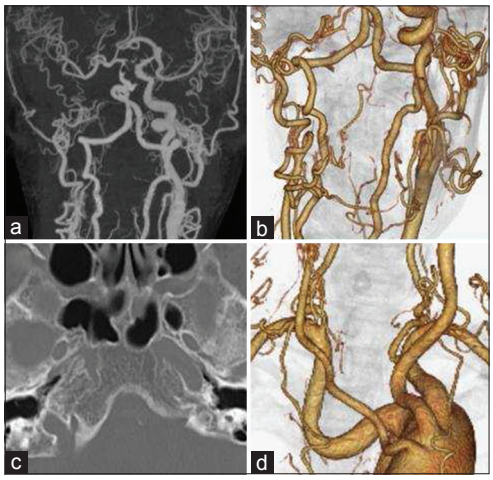

Figure 2: (a) Maximum intensity projections of computed tomography (CT) angiography demonstrate stenosis of the basilar artery and aplasia of the right common carotid artery. (b) Multiplanar reconstruction of CT angiography of the neck demonstrates aplasia of the right internal carotid artery. (c) Skull base CT scanning demonstrates an absence of the right carotid canal. (d) Abnormal origin and course of the left subclavian vessels are demonstrated. Bilateral common carotid arteries have arisen from the distal side of the left subclavian artery.

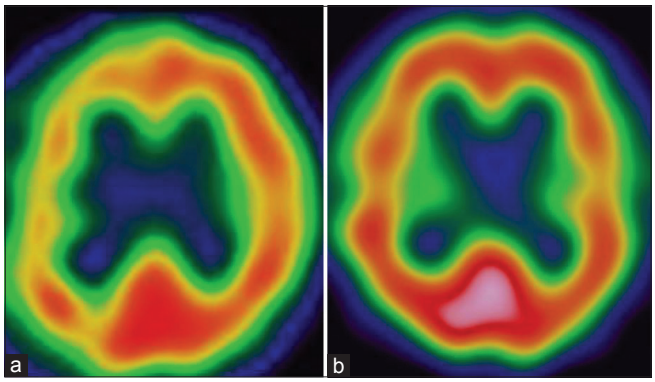

Figure 3: (a) Single-photon emission computed tomography demonstrates reduced cerebral blood flow in the border zones between the middle and posterior cerebral artery territories in the right hemisphere. (b) After extracranial–intracranial bypass, cerebral blood flow of the right hemisphere improves to the normal level.

signs and symptoms of PHACE syndrome typically present in infancy or childhood and may be related to congenital lesions of the cerebrum or cerebellum as the result of acute ischemic stroke. Acute ischemic stroke is rare in children and difficult to diagnose. There are many factors that may contribute to the under-recognition of TIA in infants with PHACE, including the fact that a clinical diagnosis of stroke is often delayed in children.[2,9] Heyer *et al.*[7] concluded that children with PHACE are at a significantly increased risk of developing ischemic stroke. However, there are scant data about the incidence of ischemic stroke, neurological sequelae, and long-term prognosis in adult PHACE patients.

Our patient presented at 75 years of age, and PHACE was not considered earlier when he was found to have a Dandy–Walker malformation. It is important to emphasize that a complete imaging evaluation be conducted in patients, even if they are of advanced age. In this patient, a new diagnosis of PHACE syndrome was given, according to the diagnostic criteria presented by Sullivan *et al.*[12] TIA was probably induced by aplasia of the right ICA and stenosis of the basilar artery. TIA has been explained by the hypoperfusion of the right cerebrum, in which carotid aplasia and basilar artery stenosis lead to a decrease in cerebral blood flow in the critical watershed territories as seen in a SPECT scan. Ischemic damage to the neurons may increase membrane instability, excitatory neurotransmitter release, neuronal excitability, and consequent movement disorders.[1]

CONCLUSION

PHACE syndrome is a rare congenital neurocutaneous syndrome. It usually presents during infancy and childhood. Here we present a rare case of adult PHACE syndrome with aplasia of the ICA. The patient experienced TIA which induced ipsilateral hypoperfusion. PHACE syndrome should be kept in mind, even in individuals of advanced age, in the instance of a TIA, especially in situations which may involve induced hypoperfusion. Details of birth history, complete surveys of the brain, and intracranial and extracranial vasculatures are strongly advised because these patients are at increased risk of TIA.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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