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LETTER TO THE EDITOR

Carotid agenesis and absence of bifurcation, asymptomatic and incidental, during the study of cognitive impairment: a case report☆

Agenesia carotídea y ausencia de bifurcación, asintomáticas e incidental, durante el estudio de deterioro cognitivo: a propósito de un caso

Introduction

Carotid vascular anomalies (carotid agenesis and absence of the carotid bifurcation) are a very rare finding; they are mostly asymptomatic but are occasionally associated with neurological disorders derived from the collateral circulation.

These interesting anomalies and variants of normality may be observed incidentally during non-invasive neuroimaging studies performed for other reasons.

Material and methods

We present a case of this rare vascular anomaly identified during the analysis and assessment of mild cognitive impairment.

After a head CT scan (Fig. 3) performed to assess cognitive impairment, we detected a congenital absence of the right carotid canal, and decided to expand the study with MRI angiography studies.

A cranial MRI angiography revealed a small-calibre right common carotid artery running on to the external carotid artery, with no visible carotid bifurcation. The right internal carotid artery (ICA) was completely absent (Figs. 1 and 2). The right middle cerebral artery (MCA) was fed by the posterior circulation. As a variant of normality (bovine arch), we observed a common origin of the right brachiocephalic truncus arteriosus and the left common carotid artery. The left ICA originated from a common segment and subsequently split into 2 anterior cerebral arteries. The common, internal, and external carotid arteries were permeable on the left side. Both vertebral arteries were permeable, with the left artery being dominant (Fig. 2). Dolichomegabasilar artery was also detected. At the parenchymal level, we observed minimal chronic ischaemic damage.[1,2]

Results

Congenital absence (complete agenesis) of the ICA is a very rare anomaly with incidence < 0.01%.[3,4]

It may be bilateral, but unilateral and left-sided cases are more frequent (3/1).

The precise cause is unknown; it is believed to be due to a mechanical or haemodynamic alteration during embryonic development, between the 3rd and 5th weeks of fetal life.[4]

The origin of the external carotid artery is controversial. Some authors argue that it shares an origin with the internal carotid artery, at the third aortic arch, whereas others suggest that the external carotid artery originates independently at the aortic sac, as it develops normally in most patients with absent ICA.

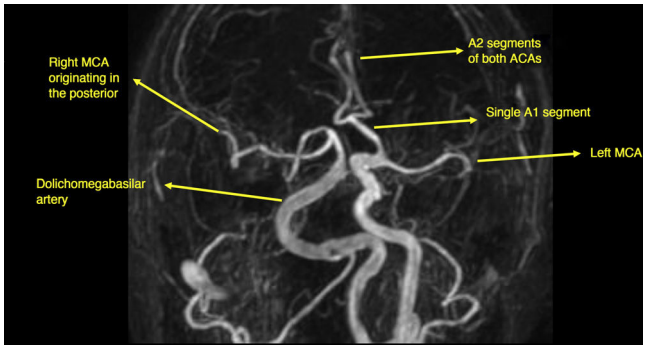

Figure 1 Intracranial MRI angiography.

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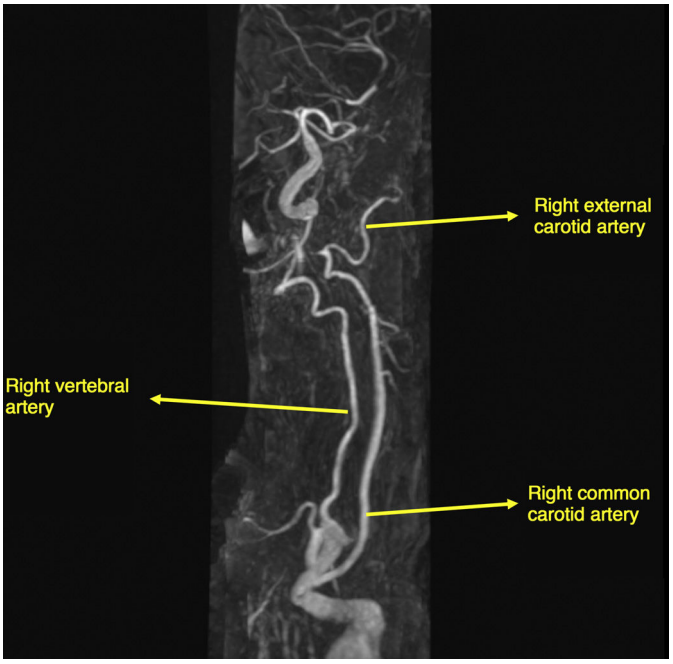

Figure 2 MRI angiography of the supra-aortic trunks.

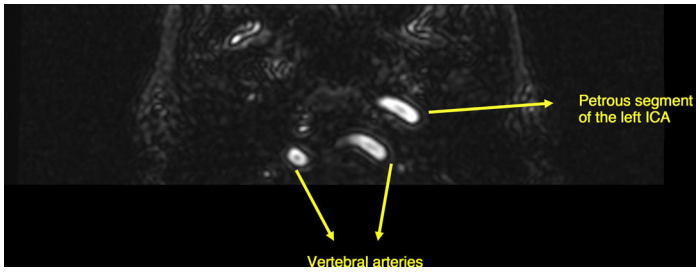

Figure 3 Head CT scan.

Discussion

Congenital absence of the ICA is normally asymptomatic, due to the presence of compensatory collateral circulation that is able to maintain perfusion of the brain; it is usually detected incidentally during imaging studies or after an ischaemic or haemorrhagic stroke.

There are no defined clinical symptoms; it has been associated with recurrent headache, blurred vision, hearing loss, Horner syndrome, epilepsy, hemiparesis or intracranial haemorrhage due to ruptured cerebral aneurysm, cerebrovascular disease, cognitive impairment, etc.[5]

Absence of the ICA is accompanied by the development of collateral circulation from the circle of Willis, transcranial branches originating at the external carotid artery, or persistent embryonic vessels.

In 1968, Lie published a classification of 6 patterns of collateral circulation (type A to F).[6]

Type A refers to unilateral absence of the ICA. The anterior cerebral artery (ACA) of the affected side is fed from the posterior communicating artery (PCoA) via the anterior communicating artery (ACoA) and the MCA.

In the fetal type, the most frequent form, the ACA of the affected hemisphere is supplied from the contralateral ICA via the ACoA, whereas the MCA is supplied from the basilar artery via the PCoA.

Conclusions

In this case, we describe the peculiar association of 2 rare congenital anomalies (absence of carotid bifurcation and internal carotid agenesis) and other variants of vascular normality in the same patient (bovine arch and dolichomegabasilar artery), with predominance and unusual features on the right side.

Although our patient was asymptomatic and we identified no association with her cognitive impairment, it is important to identify this phenomenon in some patients due to its association with cerebral aneurysms in thromboembolic disease, especially if contralateral carotid surgery is under consideration.[7]

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Characteristics of epilepsy secondary to mutations in the *PNKP* gene☆

Características de la epilepsia secundaria a alteraciones en el gen *PNKP*

Dear Editor:

PNKP gene mutations cause neurodevelopmental disorders with varying degrees of epilepsy, psychomotor retardation, cerebellar atrophy, and peripheral neuropathy[1]. Different phenotypes have been described in the literature:

1 Microcephaly, seizures, and developmental delay (MIM #613402). The condition, first described by Shen et al. in 2010, follows an autosomal recessive inheritance pattern. Patients present congenital microcephaly, early-onset epilepsy rapidly progressing to developmental and epileptic encephalopathy, and intellectual disability.[1—4]
2 Ataxia-oculomotor apraxia 4 (MIM #616267). First described by Bras et al. in 2015, it is characterised by ataxia and oculomotor apraxia secondary to cerebellar atrophy. Patients frequently present axonal sensorimotor polyneuropathy, but do not present microcephaly or epilepsy.[3,5,6]
3 In recent years, cases have been reported of patients with intermediate phenotypes[3,4,7—10]:

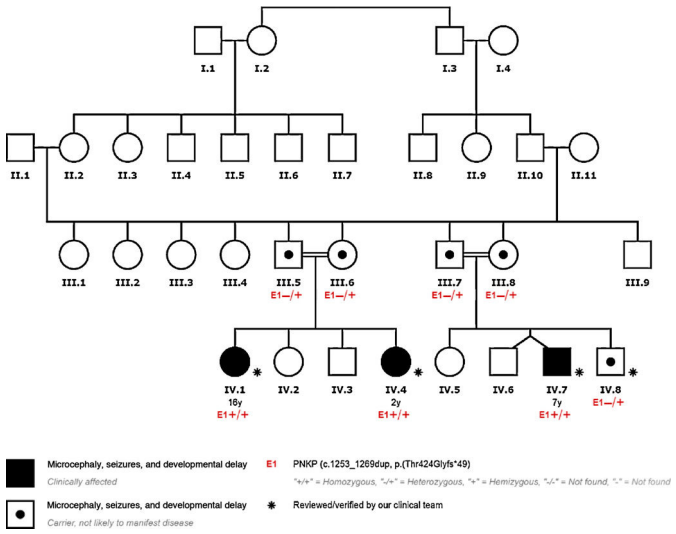

Figure 1 Pedigree chart of our patients' family.

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