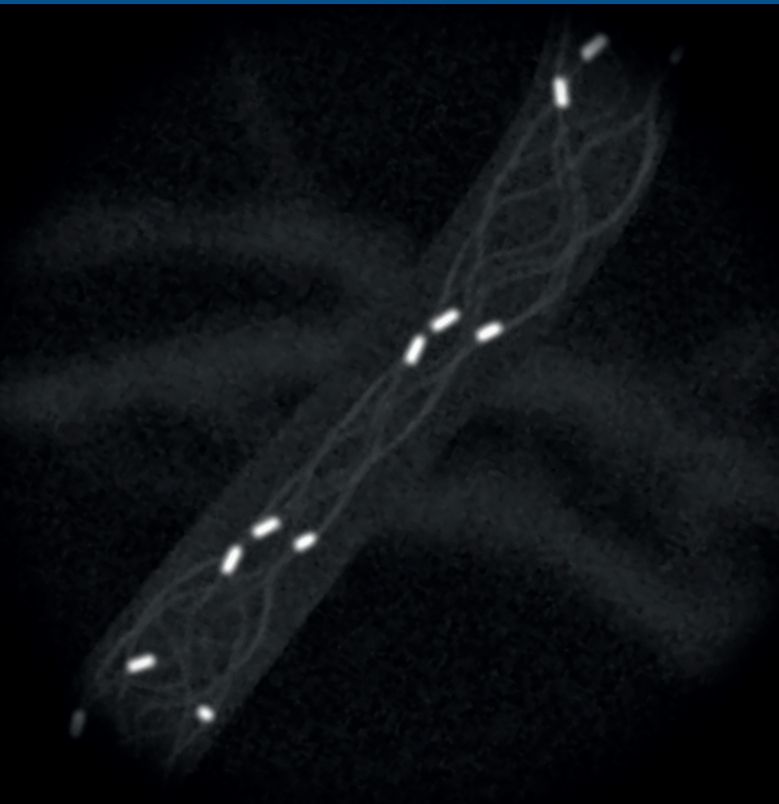


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Internal carotid artery agenesis: A case report and review of literature

Shuo Li¹, Kusum Hooda¹, Nishant Gupta² and Yogesh Kumar¹

Abstract

Congenital agenesis of the internal carotid artery is a very rare anomaly. Intracranial circulation in the involved internal carotid artery territory is maintained by collateral circulation from the contralateral internal carotid artery through the anterior communicating artery and from the vertebrobasilar system through the posterior communicating artery. Usually, patients with internal carotid artery agenesis are asymptomatic due to collateral circulation, but they may present with headache, seizures, or transient ischemic attack. Aneurysms have also been reported to be associated with this entity. Computed tomography angiography, magnetic resonance angiography, and digital subtraction angiography are commonly used modalities to make the diagnosis. Unenhanced skull base computed tomography will show the absence of carotid canal, thus differentiating from carotid hypoplasia. We report on a case of right internal carotid artery agenesis with discussion of embryogenesis, clinical presentation, and imaging findings.

Keywords

Cerebral collateral circulations, circle of Willis, internal carotid artery

Introduction

Absence or hypoplasia of the carotid artery are rare developmental anomalies. A review of literature has yielded less than 200 reported cases. Recognition of this rare anomaly is important, particularly when considering endovascular interventions in the event of thromboembolic events with revascularization. We describe a case of carotid agenesis which was discovered serendipitously, and review the currently available literature related to this condition.

Case description

A 50-year-old male with multiple comorbidities, including history of HIV/AIDS and heart failure with reduced ejection fraction (HFrEF), presented to the emergency department with weakness and weight loss. Due to the immunocompromised state of this patient, a contrast-enhanced head computed tomography (CT) was obtained by the emergency department due to an initial concern for central nervous system (CNS) infection such as toxoplasmosis.

The contrast-enhanced head CT demonstrated no abnormalities, such as cerebral infarction, and there was no abnormal enhancement to suggest cerebral infection. Incidentally, however, an absence of the right internal carotid artery (ICA) was noted (Figure 1). Axial CT imaging at a higher level near the cavernous sinus demonstrated a lack of the right

ICA. On careful review of the contrast-enhanced CT, the right middle cerebral artery (MCA) was found to receive collateral circulation from the vertebrobasilar circulation via the posterior communicating artery (Figure 2); the right anterior cerebral artery (ACA) received collateral circulation from the contralateral ACA via the anterior communicating artery. Examination of the head CT in bone window demonstrated an absence of the right internal carotid canal (Figure 3). This finding categorizes our case as one of carotid agenesis, rather than hypoplasia, and this is distinguished and discussed further below.

Subsequent CT of the chest demonstrated a relatively small right common carotid artery (CCA) caliber (Figure 4). No image of the carotid bifurcation was obtained.

The patient's focal neurological symptoms resolved during the hospital stay without any intervention and, therefore, no further characterization such as computed tomography angiography or magnetic resonance angiography was obtained.

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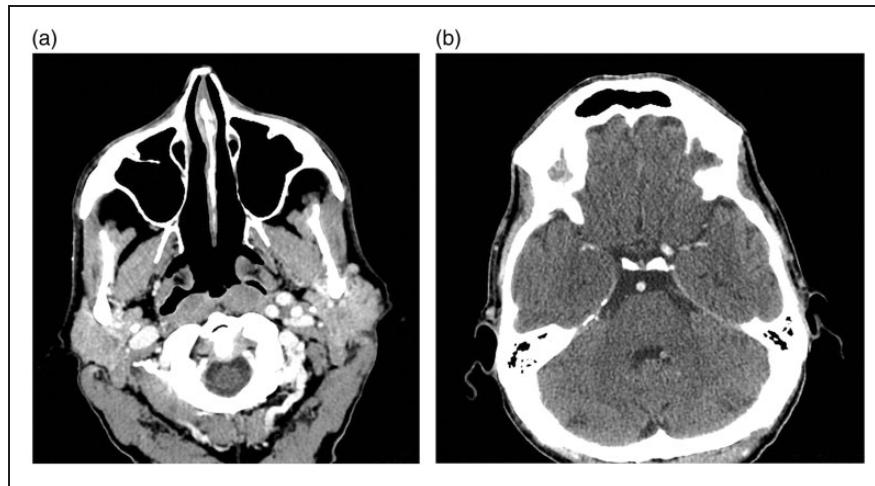


Figure 1. Axial CT image through the skull base on image (a) showing no right ICA. Axial CT image at a higher level near the cavernous sinus, image (b), demonstrates a lack of right intracranial ICA. CT: computed tomography; ICA: internal carotid artery.

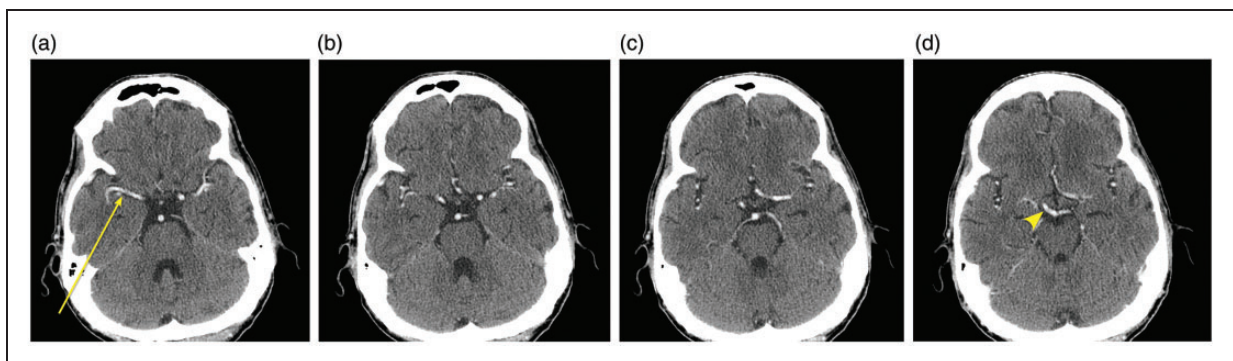


Figure 2. Consecutive axial CT slices (a) through (d) showing anastomosis and collateral flow of right MCA (arrow) from the ipsilateral PCOM (arrow head). CT: computed tomography; MCA: middle cerebral artery; PCOM: posterior communicating artery.

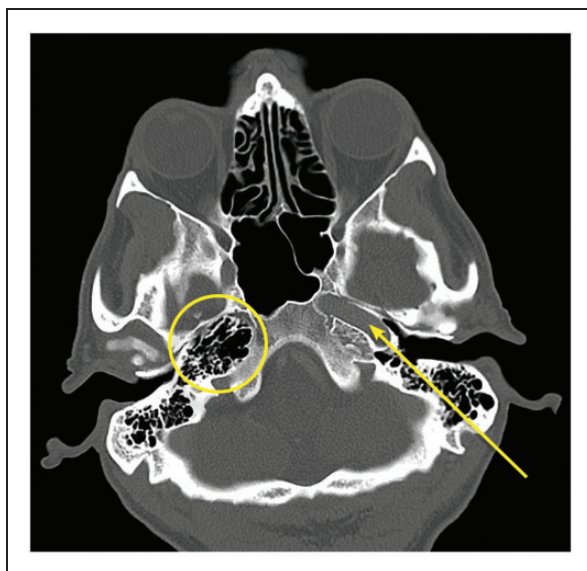


Figure 3. Axial CT scan in bone window at the skull base demonstrating a lack of internal carotid canal on the right (circle). Note the patent internal carotid canal on the left (arrow). CT: computed tomography.

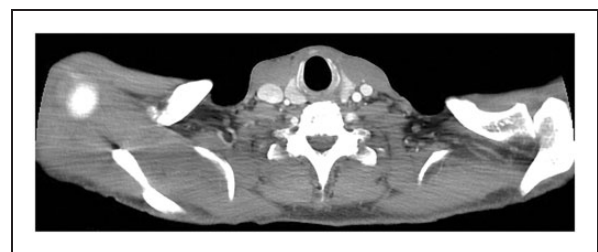


Figure 4. Axial computed tomography (CT) scan of the upper thorax showing a relatively diminutive of right common carotid artery.

Discussion

Embryology

The ICA is a terminal branch of the CCA. The ICA consists of seven segments according to the Bouthilier classification: cervical, petrous, lacerum, cavernous, clinoid, ophthalmic (supraclinoid), and communicating (terminal) segments.¹ According to Lasjaunias and others,^{2,3} the different segments arise from different

embryologic segments, specifically the following arteries: ventral pharyngeal hyoid, mandibular, primitive maxillary, trigeminal, dorsal ophthalmic, and ventral ophthalmic. A complete review of the embryological origin of the ICA, including each segment, is beyond the scope of this article. We offer a brief overview of the embryologic development of the ICA.

The ICA begins to develop in the 3 mm stage of embryological development.⁴ It develops from the first and third aortic arches and the paired dorsal aorta. The root of the ICA is formed from the third aortic arches. Intermediate portions of the ICA develop from the dorsal aorta between the first and third aortic arches. The distal ICA originates from the dorsal ends of the first aortic arch.⁵ In the 4 mm embryo, anterior and posterior divisions of the distal ICA form. The anterior division develops to supply the forebrain, and includes the ACA, MCA and anterior choroidal artery. The posterior division is the progenitor of the posterior choroidal artery, diencephalic, and mesencephalic arteries. The basilar artery arises from posterior extension of the posterior communicating arteries bilaterally in the 5–8 mm embryo. By the 14 mm embryo stage, the vertebral arteries have formed from anastomoses between cervical segmental branches of the dorsal aorta and unite with the basilar artery via upper cervical segmental branches. The CCAs begin forming following involution of the portion of the paired vertebral aortic roots between the third and fourth aortic arches in the 12–14 mm embryo. Portions of the third aortic arches proximal to the external carotid artery (ECA) become the CCAs (Figure 5).⁶ Some controversy exist regarding whether the ECA develops with the ICA. Radiographic evidence such as collateral circulation via the rete mirabile suggests that the ECA develops separately from the ICA.⁷ Intracerebral collateral circulation via the circle of Willis is complete in the 24 mm embryo stage, with the completion of the anterior communicating artery.

Agenesis of the ICA occurs due to abnormal regression of the first and third aortic arch, with the exact etiology being unclear.^{8,9} It appears that collateral circulation in ICA agenesis depends on when the insult to the ICA embryologic origin (proximal, mid, or distal ICA origin) occurs. For example, in cases of ICA agenesis that presents with collateral circulation via the circle of Willis, insult to ICA origin would occur after development of basilar artery (14 mm stage) but before completion of the circle of Willis (24 mm stage).

Anatomy

The first case of carotid agenesis was discovered on postmortem examination in 1787 by Tode.¹⁰ Subsequently, the first case of carotid agenesis diagnosed by angiography was reported by Verbiest.¹¹ Since then, there have been less than 200 case reports of carotid artery agenesis.

The terms of agenesis, aplasia, and hypoplasia are often used interchangeably in the setting of an absent or small ICA. Lie and Hage defined agenesis as the total absence of ICA; whereas hypoplasia and aplasia refer to cases where a portion/segment of the ICA is present, with a normal or even slightly enlarged ICA segment proximal to an abrupt narrowing.¹² Therefore, in agenesis the bony carotid canal is absent, whereas in aplasia and hypoplasia the bony carotid canal is present.

Six types of circulation to the ACA and MCA on the ipsilateral side of carotid agenesis were described by Lie and Hage in 1968.¹² Overall, the types of collateral circulation can be separated into three categories: collateral flow through the circle of Willis, through persistent fetal circulation, and through skull base arteries stemming from the ECA, the so-called rete mirabile (Figure 6).

In type A, there is unilateral absence of the ICA with collateral circulation to the ACA through the anterior communicating artery (ACOM) and the MCA through the posterior circulation via the posterior communicating artery (PCOM); our index case is type A. In type B, ipsilateral ACA and MCA on the side with absent ICA are supplied by the ACOM. In type C, there is bilateral carotid artery agenesis with supplies to the ACAs and MCAs through the posterior circulation via the PCOMs. In type D, there is agenesis of the cervical portion of the ICA with the cavernous portion of the ICA reconstituted by an anastomotic siphon from the contralateral cavernous ICA. Type E is unique in that there is hypoplasia of bilateral ICA rather than true absence with diminutive ACAs; there are prominent PCOMs with normal caliber of MCAs, suggesting that the majority of blood flow in the MCAs originates from the posterior circulation. Type E can be better referred to as carotid hypoplasia, where the carotid canal persists and there are remnants of vessel with the canal. Type F demonstrates distal reconstitution of ICAs via anastomosis with distal branches of ECAs. Since Lie's original description, an additional type of collateral in which the ICA and MCA are reconstituted by a persistent trigeminal artery (PTA) has also been described (the different types of PTA are not discussed here).¹³

Epidemiology and clinical presentation

As described above, carotid agenesis can be both unilateral and bilateral, with a higher proportion of cases being unilateral. There is also a predilection to left-sided carotid agenesis, with a reported ratio of cases of 3:1. The true incidence of carotid artery agenesis is unclear, with most cases such as our index case found incidentally by ultrasound, magnetic resonance imaging (MRI), or CT. Most cases are clinically silent, due to well-developed collateral circulations.¹⁴ In a retrospective review of cerebral MRI and angiograms, Ryan et al.¹⁴ found seven patients with either absence or

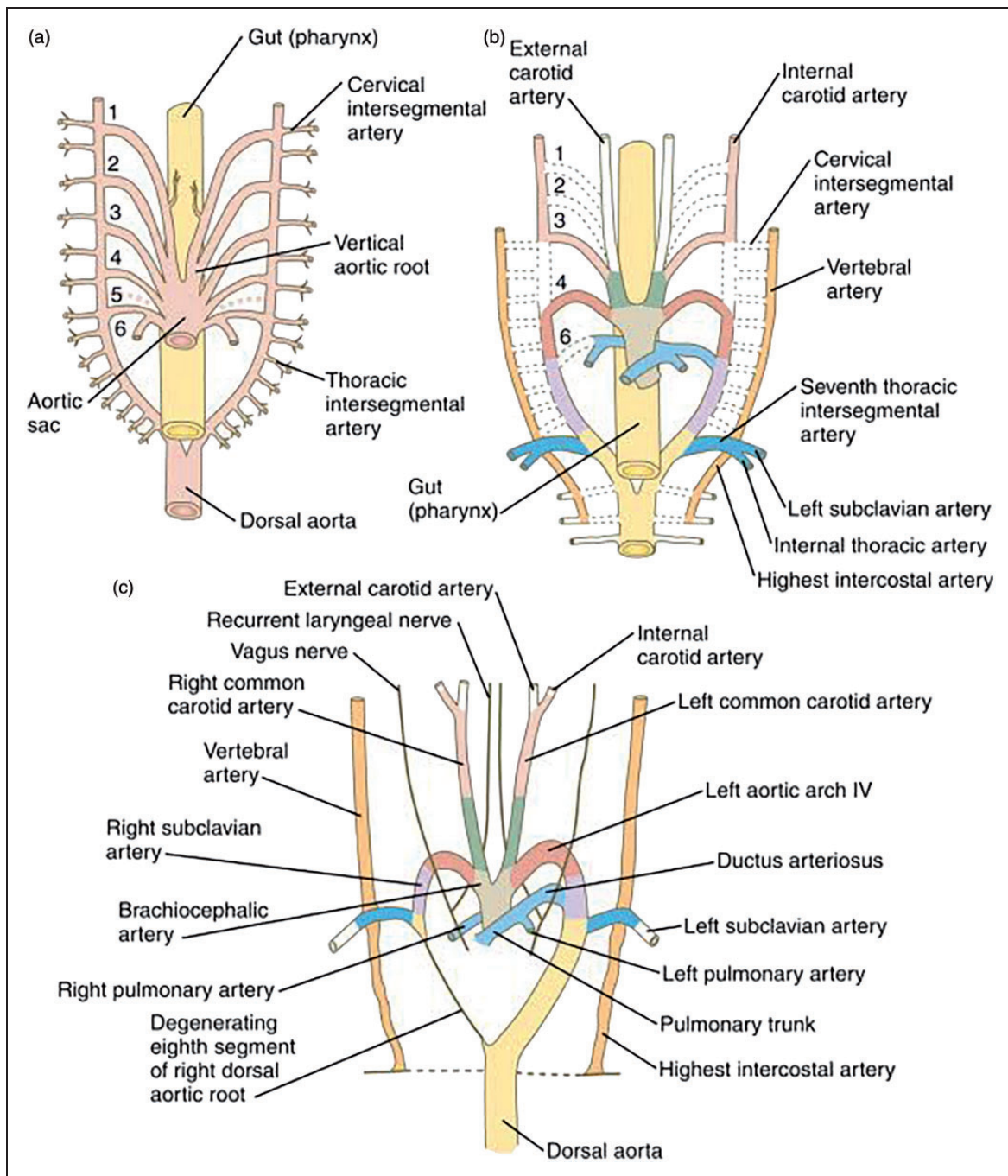


Figure 5. Diagram illustrating the overview of the aortic arches and the carotid arteries. A: multiple aortic arches are seen (numbered) as well as other smaller arteries. B: depiction of more developed aortic arches and carotid arteries, with regression of multiple vessels, including the first and second aortic arches. C: depiction of mature aortic arch and proximal carotids, color coded to correspond to A and B. Reproduced with kind permission from Elsevier.³²

hypoplasia of the ICA from more than 5000 examinations, with an incidence of 0.13%.

However, there are also other abnormalities which have been reported to be associated with carotid artery agenesis.^{15–18} Some of the earliest reports of carotid agenesis were found secondarily, after rupture of cerebral aneurysms in the contralateral circulation causing subarachnoid hemorrhage,^{19–22} with a significantly higher association of aneurysms seen in these patients.

Zink et al.²³ found 27.8% of cases involving carotid agenesis or hypoplasia are associated with intracerebral aneurysms, compared to a incidence of 2–4% in the general population. Increased incidence of cerebral aneurysms found by Zink et al. for patients older than 30 years suggests that the cerebral aneurysms are acquired rather than congenital.

Other clinical symptoms which have been reported in the setting of carotid artery agenesis include pulsatile

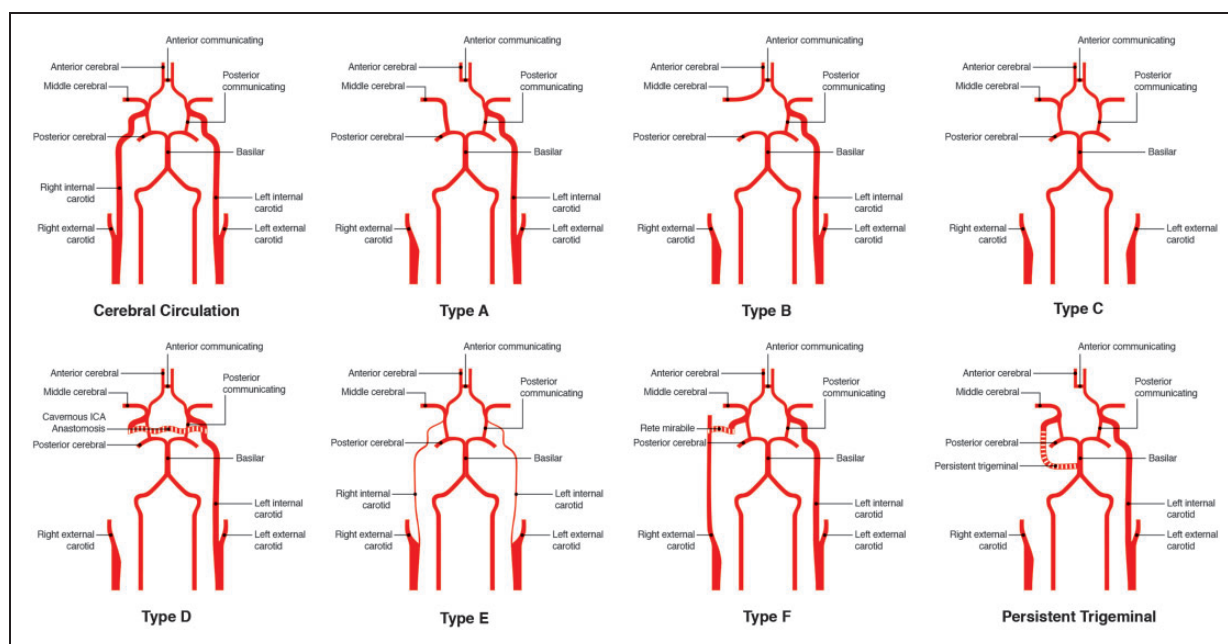


Figure 6. Diagram illustrating the different types of anastomosis in the setting of internal carotid artery (ICA) agenesis. Basic anatomy is illustrated in the first panel, with type A through type F following. An additional type of anastomosis, the persistent trigeminal artery, is also depicted here in the last panel.

tinnitus, ischemic stroke/transient ischemic attack (TIA) symptoms, migraine, and Horner's syndrome.⁹

Rare syndromes, such as Posterior fossa brain malformations, Hemangiomas, Arterial lesions, Cardiac abnormalities/aortic coarctation and Eye (PHACE) abnormalities, Goldenhar syndrome, coarctation of aorta, Klippel-Feil syndrome, have also been reported in the setting of carotid agenesis.^{24–27} Other endocrinologic deficiencies have also been reported in carotid agenesis, such as hypopituitarism and growth hormone deficiency.^{28–30} These reports, however, remain anecdotal, and no overarching or unifying theory exists to tie carotid agenesis with these reported abnormalities.

Treatment options and clinical implications

Given the asymptomatic and congenital nature of carotid agenesis, no treatment is necessary or possible to re-establish the ICA. However, with the high prevalence of associated cerebral aneurysms, it is our opinion that non-ionizing imaging methods, such as MRA can be useful for screening of cerebral aneurysms in patients with known carotid agenesis. No guidelines currently exist pertaining to the management of carotid stenosis with contralateral agenesis, and therefore indications for treatment are the same as in the general population.³¹

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Conflict of interest

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