

## Agenesis of the internal carotid artery: a family pathology?

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**SUMMARY:** Agenesis of the internal carotid artery: a family pathology?

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*The internal carotid artery agenesis is a rare malformation disorder. We report the case of a 12-year-old boy suffering migraine, who had presented an episode featuring amaurosis fugax, spontaneously re-*

*gressed. CT angiography images show hypoplasia of the left common carotid artery with loss of opacification of the left internal carotid artery consistent to agenesis. Moreover CT scans through the skull base demonstrate absence of left petrous carotid canal and an hypertrophic left middle cerebral artery originating from an aberrant artery arising from the right cavernous carotid. All diagnostic examinations confirmed the presence of the internal carotid artery agenesis, as Lie's type IV. We started an annual follow up that over the next 7 years did not reveal any change in magnetic resonance angiography images.*

**KEY WORDS:** Vascular abnormalities - Internal carotid artery agenesis - Magnetic Resonance Imaging - Magnetic Resonance Angiography.

## Introduction

The internal carotid artery (ICA) agenesis is a rare malformation disorder. This disease is estimated to be less than 0.01% of the population (1). From a clinical point of view its severity can be very variable. In most cases, the malformation can remain asymptomatic for a long time. Diagnosis is often made in adulthood after a color doppler ultrasound of neck vessels. Sometimes the onset can be characterized by mild headache or amaurosis fugax, but sometimes the symptoms are severe and begin with acute crises of headache and cerebral hemorrhage (2).

The embryology of the internal carotid artery is very complex (3) and this leads to the possibility of malfor-

mation disorders during its development. Lie described six anatomical variants in his classification (4) and in some cases the presence of aneurysms along the course of the primary vessel exacerbates the complexity of the disease (5). In most cases reported in literature abnormalities in the ICA course are found (agenesis or hypoplasia associated with deformed accessory vessels). In other cases saccular dilatation, or real aneurysms with clearly defined wall and collar, may be present along the malformed vessels.

We describe the case of a male teenager, suffering from migraine who presented an episode of amaurosis fugax.

## Case report

A 12-year-old Caucasian boy, was admitted in November 2009 at the Headache Center of "Policlinico Umberto I" in Rome, suffering from headache episodes in the last three years, characterized by constrictive pain localized in frontal region, accompanied by nausea, pallor, photophobia and rarely vomiting. The boy had also presented an episode featured by amaurosis fugax, lasting a few seconds and associated with fatigue, but not accompanied by headache, and regressed spontaneously.

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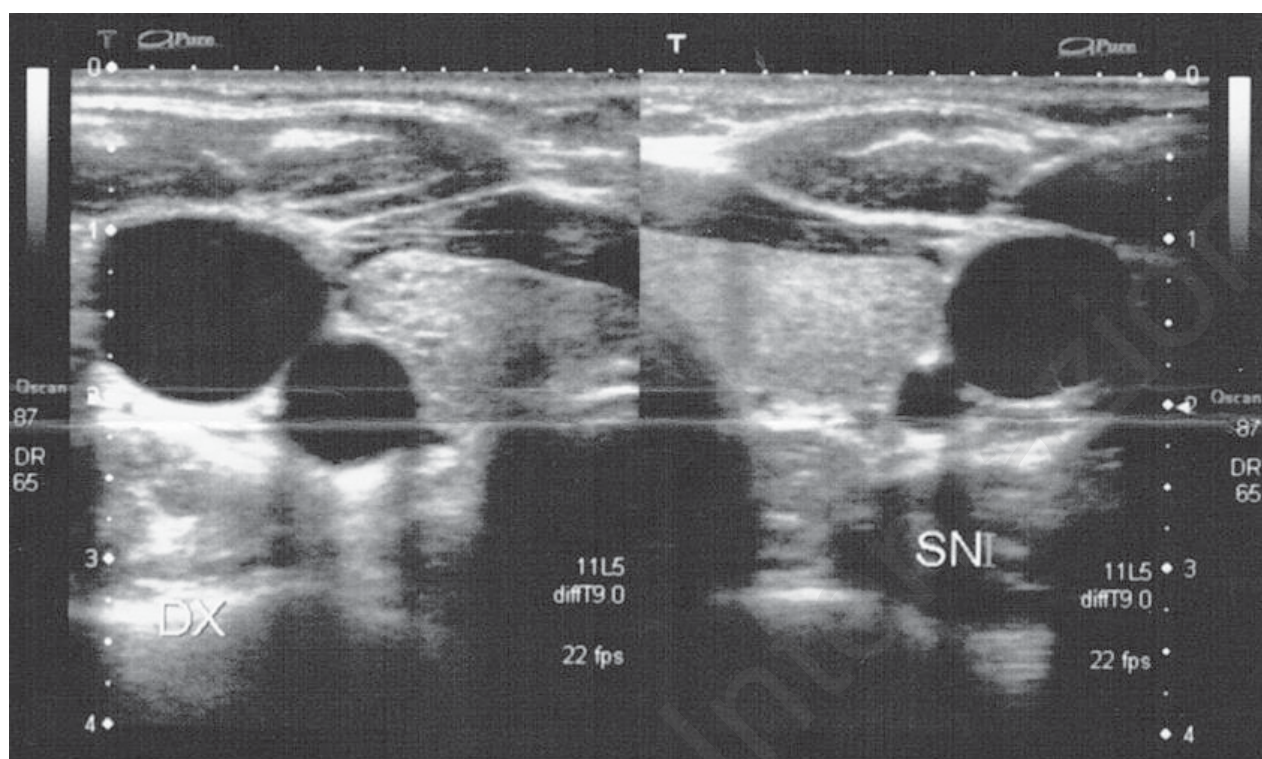


Fig. 1 - Carotid ultrasound: note the distinct difference in size between the right common carotid artery and the left one.

In the family history it was found an episode of acute cerebral ischemia in his mother, afflicted with patent foramen ovale, subsequently corrected. Moreover, his 23 years old sister was affected by an aneurysm, of about 3 mm in diameter, of the medial wall of the right ICA, with 1.8 mm collar and no signs of fissures.

The patient has been subjected, with parental consent, to emocoagulative surveys for the detection of thrombophilic risk factors. Results was in the normal range. An electroencephalographic (EEG) examination and a color doppler echocardiography were carried out; those were negative for abnormalities. Doppler ultrasound examination of the vessels of the neck (Fig. 1) and a magnetic resonance angiography (MRA) showed instead a sharp reduction of the left carotid artery diameter, compared to the contralateral vessels (right common carotid artery: 7.8 mm; left common carotid artery: 4.5 mm) (Fig. 2).

CT angiography images (Fig. 3A) show hypoplasia of the left common carotid artery with loss of opacification of the left internal carotid artery, without any identifiable remnant, consistent to agenesis. Moreover CT scans through the skull base demonstrate absence of left petrous carotid canal and an hypertrophic left middle cerebral artery originating from an aberrant artery arising from the right cavernous carotid. Such aberrant artery is also connected to the basilar artery via other anasto-

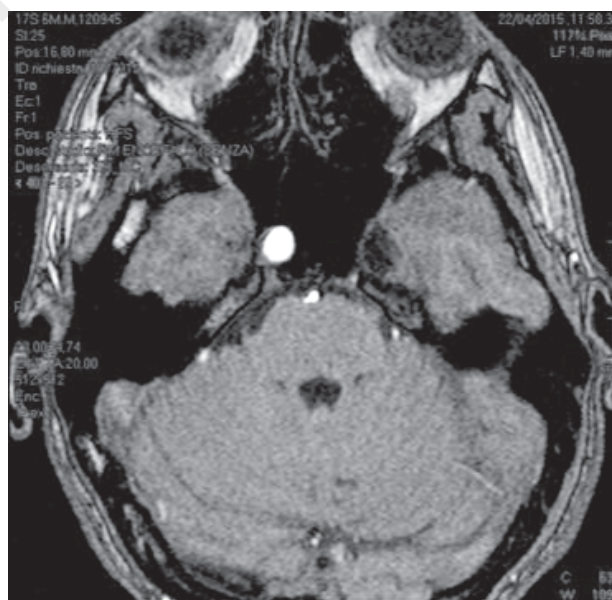


Fig. 2 - MRA: no evidence of the left ICA at the skull base.

motoc aberrant vessels (Fig. 3B) and also fills both posterior cerebral arteries. Loss opacification, for agenesis, of left A1 tract of the anterior cerebral artery with shared origin of both A2 tracts from the right A1 tract is also found. Basilar artery shows reduced diameter.

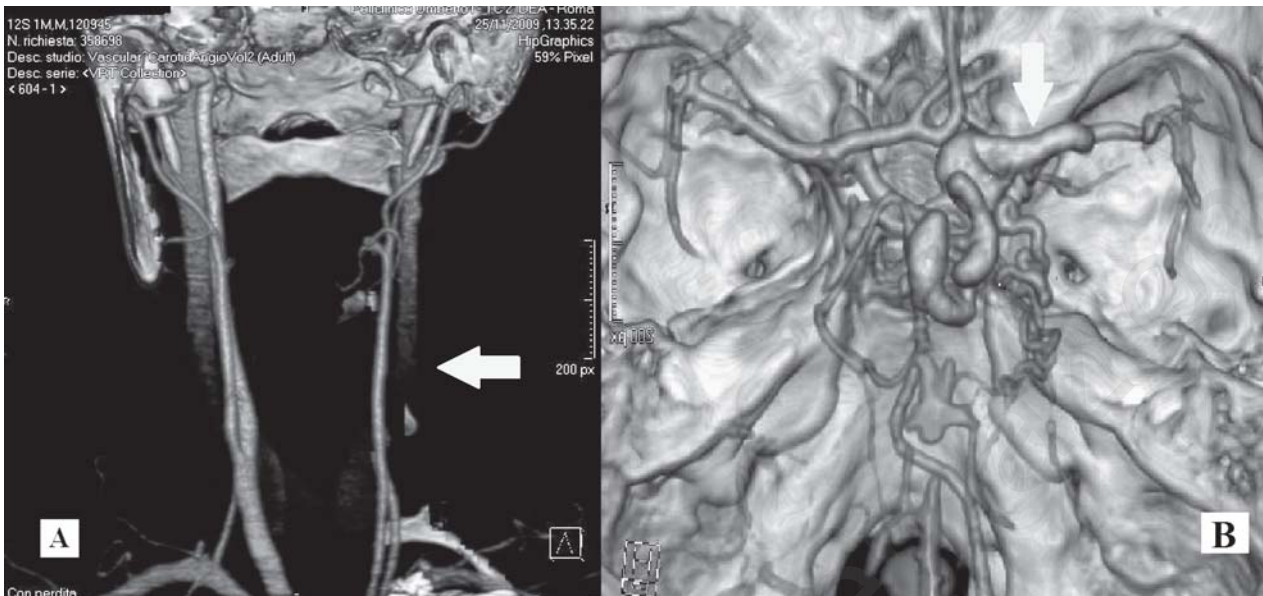


Fig. 3 - CT angiography scan. (A) Images show hypoplasia of the left common carotid artery with loss of opacification of the left internal carotid artery (arrow), consistent to agenesis. (B) Hypertrophic left middle cerebral artery (arrow) originating from an aberrant artery arising from the right cavernous carotid artery. Such aberrant vessel is also connected to the basilar artery via other anastomotic aberrant vessels and also fills both posterior cerebral arteries. Loss opacification, for agenesis, of left A1 tract of the anterior cerebral artery with shared origin of both A2 tracts from the right A1 tract is also found.

Vertebral arteries, common and internal right carotid arteries and right middle cerebral artery were normal. The examination was consistent with the presence of the internal carotid artery agenesis, as Lie's type IV.

In accordance with neurosurgeons, given the young age of the patient and not serious symptoms, we decided to give up any invasive treatment and we started an annual follow up with color Doppler ultrasound, CT scan and MRA of the supra-aortic trunks, that over the next 7 years remained unchanged compared to the first tests. We also ran repeated echocardiographic controls and monitoring of cardiovascular risk factors (Smoking, Hypertension, Diabetes, Hypercholesterolemia, and Hypertriglyceridemia) that were always normal.

## Discussion

In 1664 Thomas Willis already demonstrated the presence of an anastomotic circle at the base of the brain. It is a system that connects the anterior circulation with the rear one, allowing the adaptation of pressure and vascular and arteriolar resistances to changes in systemic blood pressure. It is well known that there are correlations between the variants of the circle of Willis and the development of aneurysms (6) and the incidence of variants are significantly higher in circles with aneurysms than in those without aneurysms.

In light of these results, as already demonstrated for malformations of the venous circulation associated to neu-

rological and other diseases of the carotid vascular malformations (7, 8), it is likely to have consequences on the hemodynamic changes of Willis circle, and that it plays some role in the development of cerebral aneurysms.

The complexity of this system makes vascular malformations in a particular risk. These are the result of complicated processes of growth, rotation and migration that are carried out during the phases of embryonic and fetal development. The most frequent congenital anomalies of the ICA, can be classified as agenesis, aplasia and hypoplasia, and may be unilateral or bilateral. In the literature it is reported the increased frequency of anomalies of the ICA to the left (left/right ratio 3:1) (9), without a significant gender difference. The collateral vessels that develop due to the presence of congenital anomalies of the ICA, are not only compensating vascular mechanisms, but may be suggestive of congenital abnormalities.

As part of ICA agenesis, Lie described six different anatomical variants (4). In our case, according to this classification, the anatomical variation was type IV (Fig. 3) and was found unilateral hypoplasia of the cervical portion of the left ICA, associated with transcavernous communications. A tortuous collateral vessel originated from the right cavernous carotid artery and it was in connection with the basilar artery.

In literature it is reported that in 24% - 34% of cases the anomaly is associated with aneurysms along the course of the anomalous vessel (9). In our case, with regard to the symptomatology onset, our patient presented with symptoms characteristic of this pathology, as



reported by Lee (2). The patient did not appear to be suffering from aneurysm at the diagnosis time or during follow-up, but an aneurysm of the right internal carotid artery had been found in his sister and his mother had an episode of acute cerebral ischemia, we don't know if it was in correlation to the presence of a cerebral aneurysm. This picture may be suggestive of genetic predisposition or at least of a family history of vascular malformations. During follow-up the symptoms became more nuanced and less frequent, comforting us with the choice made about abstaining from surgery.

We believe that in agreement with what reported in the literature (10), every single case must be evaluated depending on the set of the anomaly to determine any possible surgical therapies, presenting in any case a potentially fatal risk. Above all it is necessary a close MRA follow-up for the possible development of aneurysms, and the evaluation of risk factors for cardiovascular disease. Also we suggest in all

cases to carry out a family screening given the often familiar nature of the disease.

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#### **Disclosure statement**

All authors declare that they have no conflicts of interest.

#### **Ethical approval**

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

#### **Informed consent**

Informed consent was obtained from all individual participants included in the study.

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