Aplasia of the cervical internal carotid artery and malformation of the circle of Willis associated with Klippel-Trenaunay syndrome

Case report

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The authors describe the radiographic findings in a patient with Klippel-Trenaunay syndrome who also had aplasia of the left internal carotid artery and a very unusual malformation of the circle of Willis. This constellation of clinical and radiographic findings is unique and has not been previously reported in the medical literature.

KEY WORDS - internal carotid artery agenesis - Klippel-Trenaunay syndrome - circle of Willis - dermal telangiectasis

The triad of soft-tissue and skeletal hypertrophy, segmentally distributed cutaneous nevi, and vascular anomalies was initially described by Klippel and Trenaunay in 1900. The vascular malformations associated with Klippel-Trenaunay syndrome generally involve the deep venous systems of the extremities; however, arteriovenous malformations (AVM's), hemangiomas, and lymphangiomas have also been associated with the syndrome.

We describe the case of a patient with Klippel-Trenaunay syndrome who also had aplasia of the left cervical internal carotid artery. She also had an unusual ectatic tortuous channel coursing through the suprasellar cistern to provide collateral flow to the left middle cerebral artery area. The radiographic findings are presented and their implications discussed.

Case Report

This 24-year-old white woman presented in November, 1981, with severe left-sided occipital headaches which woke her in the middle of the night and lasted for 4 days. The headaches were associated with nausea and photophobia. She had experienced similar isolated episodes since 1977 when she was initially admitted to the hospital. At that time, the diagnoses of Klippel-Trenaunay syndrome and migraine headaches were established. Her medical history was remarkable for frequent nosebleeds and blindness in the left eye since birth. At no time had she ever had a documented subarachnoid hemorrhage.

Examination. Her physical findings included exotropia of the left eye with marked decrease in visual acuity. The funduscopic examination suggested either retinal detachment or a possible coloboma. Multiple dermal telangiectatic nevi were noted on the left side of her face, left neck, left breast, and left shoulder. Her left ear was somewhat larger than its counterpart on the right, and the left arm was also larger than the right arm. There were multiple superficial dilated veins involving the proximal aspect of the left arm and shoulder.

Radiographs of the skull revealed floccular calcifications lying slightly superior to and just left of a normal-appearing sella turcica. An unenhanced cranial computerized tomography (CT) scan demonstrated calcifications in the suprasellar cistern extending into the medial portion of the left Sylvian cistern (Fig. 1 left). There was no evidence of blood in the subarachnoid...
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Fig. 1. Left: Unenhanced computerized tomography (CT) scan showing calcifications just lateral to the tip of the dorsum sellae in the suprasellar cistern which extend into the proximal portion of the left middle cerebral artery cistern. Note the lack of blood in the subarachnoid cisterns. No evidence of left cerebral hemiatrophy or infarction was seen on higher scans. Right: Contrast-enhanced CT scans showing a modest amount of enhancement surrounding the calcified mass. This appearance is most suggestive of a vascular malformation.

cisterns or ventricles. A CT scan following intravenous contrast administration revealed enhancement both central and peripheral to the suprasellar calcifications (Fig. 1 right). The CT findings were thought to be suggestive of either an AVM or, possibly, an aneurysm involving the left internal carotid artery. Cerebral angiography following the CT scan demonstrated agenesis of the cervical portion of the left internal carotid artery. The petrous and cavernous portions of the carotid artery on the left filled from the ophthalmic artery but made no significant contribution to the cerebral circulation (Fig. 2). The right carotid injection demonstrated normal anatomy with filling of the right middle cerebral artery and both anterior cerebral arteries. Right vertebral angiography revealed a very tortuous and dilated ectatic vessel which served as a collateral channel to the left middle cerebral artery system from the posterior circulation. This dysplastic vessel, which appeared to involve the P₁ segment of the left posterior cerebral artery and the left posterior communicating artery, coursed through the lateral aspect of the suprasellar cistern and terminated in the horizontal portion of the ipsilateral middle cerebral artery. The submentovertex view of the right vertebral angiogram showed multiple aneurysmal dilatations with calcified walls, which likely accounted for the abnormal densities seen on the unenhanced CT (Fig. 3).

Course. Due to the location and extent of the arterial malformation, surgical resection was deemed inadvisable. The patient was discharged the day after angiography and has remained neurologically stable during the ensuing 1½ years.

Fig. 2. Left common carotid angiograms. Left: Early arterial phase, lateral view. There is no filling of the cervical internal carotid artery. The two small branching arteries posterior to the internal maxillary artery are tributaries of the external carotid system. Center: Later arterial phase, lateral view. There is no evidence of delayed filling of the cervical internal carotid artery, confirming aplasia of this vessel. Right: Anteroposterior view. The distal internal carotid artery is present, filling from the ophthalmic artery (arrow). Note the lack of intracerebral vascular opacification.
Discussion

Although hypoplasia of the internal carotid artery has been identified in neurofibromatosis, dysplastic abnormalities of the carotid arteries have not previously been associated with the less common phakomatosis of Klippel-Trenaunay syndrome. This is not remarkable, in that the vascular anomalies found in these patients almost always involve the deep venous systems of the extremities; fewer than 10% of patients with Klippel-Trenaunay syndrome have involvement of the head or neck.

In the majority of the reported cases of unilateral partial agenesis of the internal carotid artery, the ipsilateral anterior cerebral and middle cerebral vascular territories are supplied by collateral flow from the contralateral internal carotid artery. Rarely demonstrated is an anomalous transsellar communication between the cavernous portions of the internal carotid arteries, which serves as a collateral channel to the affected cerebral hemisphere. In a recent review of the literature, Elefante, et al., identified nine such anomalous intercarotid anastomoses in addition to their own case. The origin of a middle cerebral artery ipsilateral to an aplastic carotid artery from the posterior cerebral artery, as was demonstrated in our patient, was first described by Hyrtl in a handbook of human anatomy in 1846. Since then, only four other cases of this anomaly have been reported. Ours is the first such case in which the pathology has been demonstrated both by pancerebral angiography and cranial CT scanning.

The tortuosity, dilatation, and ectasia of the collateral channel supplying the middle cerebral artery territory from the posterior cerebral artery is strikingly unique, not having been described in any of the five previous cases. The appearance of this vessel suggests an underlying deficiency of the arterial wall. Vascular dysplasias have been associated with a variety of congenital disorders besides Klippel-Trenaunay syndrome, including Marfan's syndrome, Ehlers-Danlos syndrome and Menkes' kinky-hair syndrome, and pseudoxanthoma elasticum. A pathological examination of the abnormal arterial segments in these entities generally reveals marked thickening of the medial vessel layers, hyaline degeneration of the lamina muscularis, and patchy sclerosis of the intima. Loss of the internal elastic membrane is a relatively common finding as well. It is not unreasonable to assume that the ectatic posterior cerebral to middle cerebral artery collateral channel seen in our patient is similarly deficient in its medial and intimal layers.

Orbital involvement in Klippel-Trenaunay syndrome has been well documented in the ophthalmological literature. Enophthalmos, heterochromia iridis, coloboma, and choroidal angiomas are the most common ocular findings. Blindness, if present, is most often secondary to either hydrophthalmos or chiasmatic compression by a distended third ventricle. Orbital abnormalities in Klippel-Trenaunay syndrome are not uniformly associated with facial nevi, as was found in our patient.

The etiology of our patient's headache remains in question. The sporadic nature of the unilateral headaches along with the associated nausea and photophobia is consistent with the diagnosis of either migraine headaches or recurrent subarachnoid hemorrhage. The neg-
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ative spinal tap, lack of cisternal blood on the scans, and the rarity of a subarachnoid hemorrhage in patients with cerebral arterial ectasia favors the diagnosis of migraine or vascular headaches. Furthermore, the association of the migraine headaches rather than subarachnoid hemorrhage in patients with Klippel-Trenaunay syndrome is well documented in the medical literature.12

References

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