Moyamoya Disease Associated With Polycystic Kidney Disease and Eosinophilic Granuloma

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Moyamoya disease has been associated with renal artery stenosis, cerebral hemorrhage, and multiple cranial traumas. We report a unique case of moyamoya disease associated with polycystic kidney disease and eosinophilic granuloma. Although the etiology of moyamoya disease is unknown, a familial pattern of occurrence has been documented. Of particular importance is its presentation with polycystic kidney disease, an autosomal dominant disease, suggesting a hereditary component to the etiology of this unusual vasculitic disease (Stroke 1989;20:1092-1094).

Moyamoya disease originally was described as a clinical and radiologic syndrome consisting of bilaterally occluded carotid arteries with stenosis in and around the circle of Willis. Consequently, a telangiectatic network of collateral vessels develops. Clinically, multiple stroke syndromes occur, producing variable neurologic deficits including mild dementia as a common feature.

We report a patient with a history of multiple cranial traumas, hypertension, polycystic kidney disease, and eosinophilic granuloma. Although several other associations have been reported with moyamoya disease, a presentation with polycystic kidney disease and eosinophilic granuloma is unique.

Case Report

A 30-year-old black woman incurred head trauma without loss of consciousness in a motor vehicle accident. Two days later she had blurred vision, decreased comprehension and memory, confusion, and slurred speech with difficulty expressing her thoughts. Examination showed receptive and expressive aphasia with a right hemiparesis. History revealed chronic hypertension and low baseline intellectual function although she was well-coordinated and socially active. At age 18, she had sustained left orbital trauma. She had a vague history of using diet aids (Dexatrim).

A head computed tomogram demonstrated cerebral atrophy, enlarged lateral ventricles, and four low-density infarcts within the left frontal, right parietal, right cerebellar, and pontine regions. An arteriogram showed occluded bilateral internal carotid arteries with "puff of smoke" collateralization arising from the circle of Willis (Figure 1). Collateral circulation was via lenticulostriate, thalamic perforating, meningeal, and transdural collateral arteries from branches fed by the external carotid artery.

An abnormal chest roentgenogram demonstrated diffuse, nodular pulmonary opacities. Lung biopsy histologically confirmed eosinophilic granuloma (Figure 2). Intravenous pyelogram was suspicious for left renal artery stenosis, but subsequent arteriogram demonstrated polycystic kidney disease.

The patient declined external–internal carotid artery bypass surgery. For 2 years she did well, until she presented to the emergency room with the sudden onset of headache, slurred speech, and left-sided weakness. A head computed tomogram demonstrated right putaminal hemorrhage with extension into the ventricle. Within hours, she progressed to bilateral decerebrate posturing. After a prolonged course in the intensive care unit, her neurologic status deteriorated to brain death. Autopsy was declined.

Discussion

Moyamoya disease is a poorly understood vasculitic disease characterized by compensatory collateralization that follows arterial occlusion at the base of the brain. Although predominantly a disease of children and females (female: male ratio 1.5:1), moyamoya disease can occur at any age and has a 7% familial occurrence. Originally, moyamoya disease was thought to develop chronically over months to years. However, it can occur rapidly, within 2 months, following subarachnoid hemorrhage.
Eosinophilic granuloma exerts its effects on the reticuloendothelial system through the proliferation of histiocytes; its etiology is unknown. All cases involve the lungs and, rarely, the central nervous system, with involvement of the frontoparietal region.7 Our patient demonstrated a hypodense area on computed tomograms in this region, which was presumed to be an infarct.

Adult polycystic kidney disease is an autosomal dominant disorder, with end-stage renal failure by the fourth or fifth decade. Its pathogenesis remains unknown. Renal and, rarely, liver parenchymal cysts occur. In addition, aortic root dilatation, aortic dissection, coarctation, valvular lesions, Marfan's syndrome, inguinal hernias, and colon diverticulosis have been reported.6,9 Congenital intracranial aneurysms are documented.10 Similar microaneurysms have been observed in moyamoya disease.11 Rupture of an aneurysm could account for the cerebral hemorrhage found in our patient. The concomitant occurrence of these two diseases, each associated with aneurysms, is intriguing.

Many nonprescription diet aids contain the amphetamine analogue phenylpropanolamine, which has been associated with cerebral vasculitis and hemorrhage.12,13 Although the exact etiologic factor remains unknown, phenylpropanolamine represents a risk factor for the development of intracranial hemorrhage. The relation of phenylpropanolamine to moyamoya disease in this patient is unclear.

Although moyamoya disease has been associated with neurofibromatosis,14 tuberculous meningitis,15 connective tissue diseases,16 irradiation,17 trisomy 21,18 cerebral hemorrhage,19 and cranial trauma,20 the triad of moyamoya disease, eosinophilic granuloma, and polycystic kidney disease is a precedent and suggests a genetic etiology to this vasculopathy.

References
FIGURE 2. Photomicrograph of pulmonary nodule of eosinophilic granuloma (×100). Edge of nodule with focal necrosis is on right, with normal lung parenchyma on left. Inset: Characteristic cell morphology of eosinophilic granuloma (×680).


Key Words • kidney, polycystic • eosinophilic granuloma • moyamoya disease
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