CLINICAL VIGNETTE

Leber's Hereditary Optic Neuropathy and Renal Infarct

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Case Report

A 43-year-old male with history of Leber's hereditary optic neuropathy (LHON) who is legally blind but otherwise in good health, developed acute onset of left upper quadrant abdominal pain lasting 45 minutes. The pain resolved spontaneously and returned a few days later at the same location, rated 10/10 intensity, sharp and radiating down his left flank. He had to fly from LA to San Antonio despite the pain and was admitted shortly after landing. In the ER, abdominal CT scan showed left renal infarct with an embolus in the artery to the lower pole of left kidney. Subsequent CT angiography re-demonstrated a left renal infract with embolus/thrombus in a lower pole branch of left renal artery with no evidence of AAA or dissection. He reports monthly flights between the two cities. There is no personal or family history of blood clots, TIA or stroke. He does not smoke or use recreational drugs. Initial resting for hypercoagulable state was negative and he was in normal sinus rhythm. Transesophageal echocardiogram did not show vegetations or clots. Carotid duplex revealed no atherosclerotic plaque in both carotid arteries. He was placed on IV heparin and was transitioned to warfarin. The pain subsided rapidly after initiation of IV heparin. After returning to LA, repeat hypercoagulable work up returned negative. MR angiogram revealed a distal left renal artery aneurysm at the level of the hilum measuring 10 mm, with a 5 mm neck and confirmed the left lower pole renal infarct. No evidence of intra-arterial thrombus was seen.

Discussion

Renal infarction typically results from cardioembolic disease, renal artery injury such as dissection, and hypercoagulable states. Hypercoagulability is associated with about 6% of renal infarcts and about one third of cases are idiopathic.[1]

LHON is a rare, inherited mitochondrial metabolic disease that leads to progressive vision loss in childhood due to the accumulation of reactive oxygen species. It involves 4 primary mitochondrial mutations and affects males more than females with a ratio of 2.5:1 for the G11778A and G3460A mutation, and 6:1 ratio for the T14484C mutation.[2] The metabolic injury of LHON appears to target primarily optic nerves. It may also concentrate in vascular tissues leading to aortic stiffening and microvascular ectasia involving unknown mechanism. Mitochondrial disorders other than LHON have been associated with carotid artery dissection and aortic rupture.[3] There is one prior case report of renal artery aneurysm in a patient with LHON.[4]

Common causes of renal artery aneurysm include congenital malformation of the kidneys or associated vessels, atherosclerosis, fibromuscular dysplasia, pregnancy, trauma, etc. Aneurysms can occur in main renal artery or its main branches, and can result in renal infarction due to thrombosis. This case illustrates that LHON can present with significant vascular complication and the patient developed renal infarct in the setting of renal artery aneurysm seen on MR angiogram. Clinicians should strongly consider vascular imaging for LHON patients who present with signs or symptoms concerning for vascular complications. Early diagnosis and timely intervention of renal infarction can prevent or minimize the loss of renal function. This patient will be followed with serial MR angiograms to monitor for progression of the aneurysm so timely endovascular repair can be planned if there is progressive growth to the conventional size criterion of 2 cm.

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