AJKD
Imaging Teaching Case

Anuric Acute Kidney Injury in Neurofibromatosis 1

Usman Khan, MD,[1,2] Rishi Razdan, MD,[3] and Mark A. Perazella, MD[1]

Neurofibromatosis is one of several genetic disorders characterized by hyperpigmented skin spots (café au lait spots) and multiple neurofibromas. It was first reported by Friedrich Daniel von Recklinghausen in 1882. There are 2 types of neurofibromatosis described in the literature: von Recklinghausen disease, or neurofibromatosis 1 that involves peripheral nerve sheaths, and central, or neurofibromatosis 2. Neurofibromatosis 1 very commonly involves renal arteries, and hypertension commonly is present in these patients due to stenotic ostial lesions resulting in bilateral renal artery stenosis. However, renal artery aneurysms, renal artery thrombosis, or external compression by neurofibromas also may cause hypertension. We present a unique case of a man with neurofibromatosis 1 presenting with anuric acute kidney injury, followed by a brief discussion of neurofibromatosis 1 and its association with renovascular disease.
Am J Kidney Dis. 61(3):519-522.

INDEX WORDS: Neurofibromatosis-1; renovascular disease; renal artery obstruction; acute kidney injury; midaortic stenosis.

INTRODUCTION

Neurofibromatosis is one of several genetic diseases that involve skin and nerves and collectively are called neurocutaneous disorders. It is one of the most common single-gene disorders of the central nervous system, with a prevalence of 1 event/3,000 population.[1] It was reported first by Friedrich Daniel von Recklinghausen in 1882.[1] Neurofibromatosis is characterized by hyperpigmented skin spots (café au lait spots) and multiple neurofibromas. However, neurofibromatosis 1 can involve many organs, such as the skeleton and vascular system, including the renal arteries (causing stenosis). We present a case of a man with neurofibromatosis 1 who presented with anuric acute kidney injury (AKI) due to an acute renal artery thrombus with underlying renal artery stenosis.

CASE REPORT

Clinical History and Initial Laboratory Data

A 43-year-old man with a long-standing history of neurofibromatosis 1 presented with shortness of breath and weakness. He was in good health until 4 days prior when he developed progressive dyspnea, as well as fatigue, cough, mild anorexia, and a decrease in urine output. Review of systems otherwise was negative. Medical history was pertinent for hypertension and neurofibrosarcoma of the spine status post resection and radiation therapy, followed by metastases to the right upper lung lobe treated with wedge resection. He also had a right nephrectomy in 1972 for unclear reasons. He reported no use of tobacco, alcohol, or illicit drugs. Medications included metoprolol, amlodipine, benazepril, levothyroxine, quetiapine, trazodone, and oxycodone.

On evaluation, the patient was afebrile, with blood pressure of 190/110 mm Hg, heart rate of 108 beats/min, respiratory rate of 20 breaths/min, and oxygen saturation of 96% on 4 L of oxygen by nasal cannula. There were multiple café au lait spots on his abdomen; neurofibromas on his forehead, chest, and abdomen; kyphosis; and scoliosis.

Laboratory tests showed serum creatinine level of 9.7 mg/dL (estimated glomerular filtration rate [eGFR] of 6 mL/min/1.73 m^2 calculated using the 6-variable MDRD [Modification of Diet in Renal Disease] Study equation),[2] up from a baseline creatinine level 6 months earlier of 0.6 mg/dL (eGFR >120 mL/min/1.73 m^2). Potassium level was 6 mEq/L and bicarbonate level was 17.6 mEq/L. Peaked T waves were noted on a 12-lead electrocardiogram. Urinary sediment examination showed few leukocytes and isomorphic red blood cells, but no cellular or granular casts. Urine output was <100 mL for 24 hours.

Imaging Studies

Retroperitoneal ultrasonography showed a 13.3-cm solitary left kidney with no evidence of calculus or hydronephrosis, whereas Doppler study of the renal arteries was nondiagnostic. A noncontrast computed tomographic scan showed abdominal aorta narrowing, suggesting midaortic syndrome (Fig 1). Magnetic resonance imaging/angiography also revealed midaortic stenosis and a diminutive left renal artery (Fig 2).

Diagnosis

A diagnosis of renal artery stenosis with thrombosis was made and then confirmed on left renal artery angiogram. The patient underwent arteriography that showed severe ostial stenosis with poststenotic dilatation of the mid–renal artery that contained a thrombus (Fig 3B). There was collateral flow from the left testicular artery reconstituting the renal artery near the

From the [1]Department of Medicine, Section of Nephrology; [2]Program of Applied Translational Research, Department of Medicine, Section of Nephrology; and [3]Section of Vascular and Interventional Radiology, Yale University School of Medicine, New Haven, CT.

Received May 4, 2012. Accepted in revised form August 28, 2012. Originally published online November 22, 2012.

Address correspondence to Usman Khan, MD, Yale University, Section of Nephrology, PO Box 208029, New Haven, CT 06520-8029. E-mail: usman.khan@yale.edu

0272-6386/$36.00
http://dx.doi.org/10.1053/j.ajkd.2012.08.044

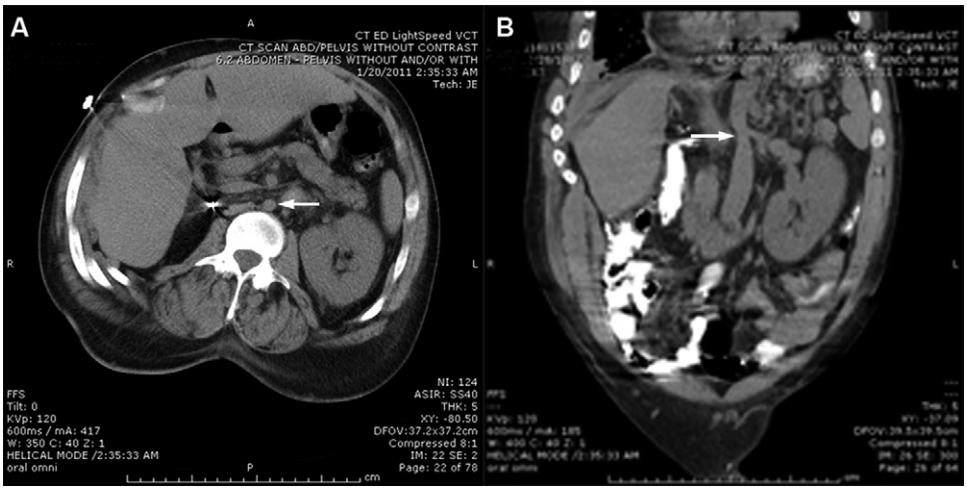

Figure 1. Computed tomographic images show narrowing of the abdominal aorta on (A) a transverse section (arrow) and (B) again on a coronal section (arrow), consistent with midaortic syndrome.

hilum (Fig 3A). Emboli from the main thrombus were noted in renal branches at midpole. The main left renal artery was revascularized with a combination of mechanical and pharmacological thrombolytic methods and stenting of severe ostial stenosis (Fig 3C). This resulted in perfusion of approximately half the kidney parenchyma (Fig 3D).

Clinical Follow-up

Tissue plasminogen activator infusion into the left renal artery followed by heparin to dissolve the remaining clot was undertaken. Kidney function improved, with increasing urine output and a gradual decrease in serum creatinine concentration at discharge to

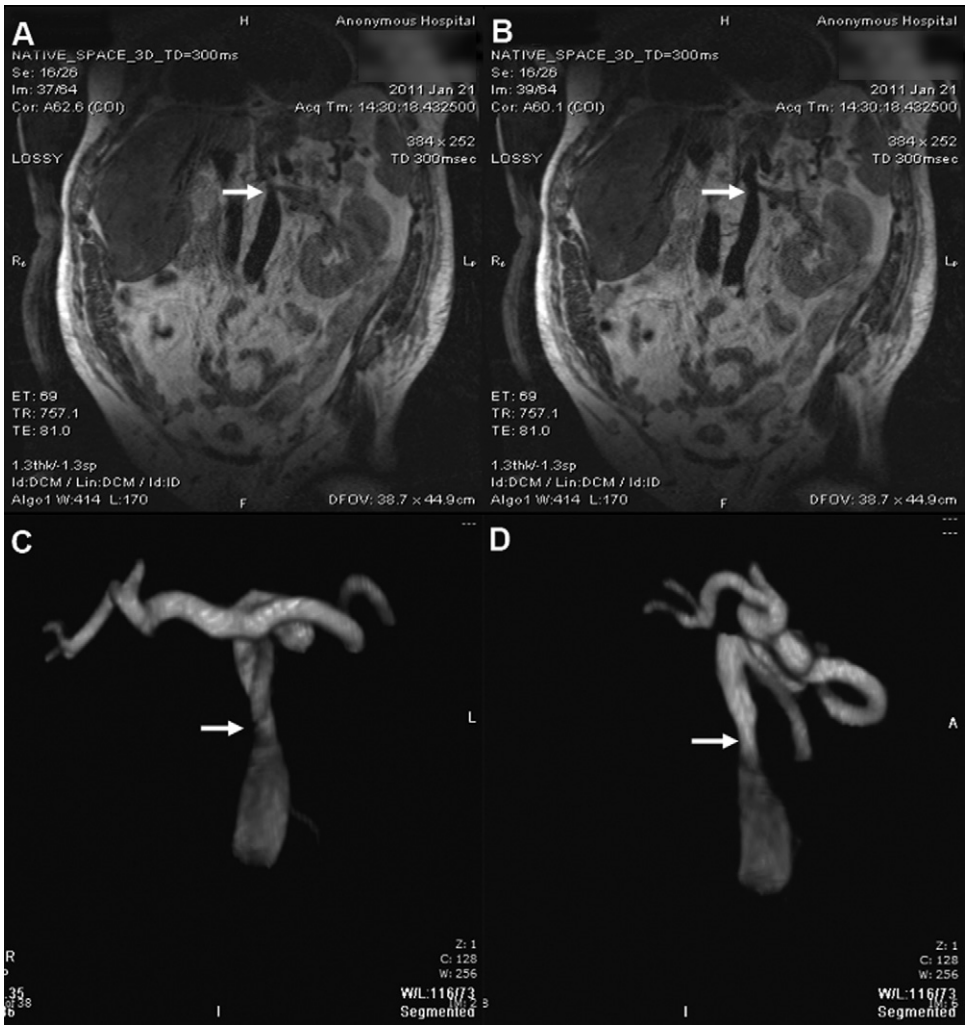

Figure 2. Magnetic resonance images show (A, B) narrowing of the abdominal aorta (arrows). (C, D) Also note the narrowing of the abdominal aorta (arrows), consistent with midaortic syndrome.