IMAGING TEACHING CASE

Midaortic Syndrome in Neurofibromatosis Type 1 Resulting in Bilateral Renal Artery Stenosis

Imran Saif, MD,[1] Dare Seriki, MD,[2] Roger Moore, MD,[3] and Alexander Woywodt, MD, FRCP[1]

We describe the case of a 23-year-old white woman with a long history of hypertension. She was referred to us 7 years after her initial diagnosis of hypertension when her blood pressure control worsened during pregnancy. Clinical examination showed an abdominal bruit and weak femoral pulses. Imaging showed midaortic syndrome with bilateral renal artery stenosis as the cause of her hypertension, and further investigations showed neurofibromatosis type 1 as the underlying disorder. Midaortic syndrome, a rare disorder of the abdominal aorta that is different from classic coarctation, typically is associated with neurofibromatosis. Renal artery stenosis is common, as are weak femoral pulses and impaired development of the lower limbs. Because of the rarity of this syndrome, only anectodal evidence exists with regard to treatment. Surgery and interventional treatment with stent placement in the abdominal aorta have been reported, as well as good outcomes with long-term medical management. Our patient continues to be healthy without intervention, with reasonable blood pressure control and normal kidney function on a 4-drug antihypertensive regimen. We discuss midaortic syndrome with a focus on diagnosis, differential diagnosis, associated conditions, and management. Nephrologists, radiologists, and ultrasonographers should be aware of this rare cause of renovascular hypertension.
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INDEX WORDS: Midaortic syndrome; renal artery stenosis; neurofibromatosis; hypertension.

INTRODUCTION

Renovascular disease is a well-described cause of secondary hypertension. Eighty percent of cases are caused by atherosclerotic renal artery stenosis, whereas the remaining 20% are caused by fibromuscular dysplasia and a number of other uncommon disorders (Box 1).[1] Midaortic syndrome, a congenital disease of the aorta and its branches, is a very rare cause of renovascular hypertension. The disease is different from classic coarctation of the aorta and involves diffuse and irregular narrowing of a segment of the abdominal aorta, often with concomitant stenosis of renal and/or mesenteric arteries. We describe a young woman who presented with severe hypertension and normal kidney function. Imaging showed midaortic syndrome with bilateral renal artery stenosis. Further investigation led to a diagnosis of neurofibromatosis type 1, a disease associated with midaortic syndrome. We describe the case with emphasis on imaging and provide a brief review of midaortic syndrome.

CASE REPORT

Clinical History and Initial Laboratory Data

A 23-year-old white woman was evaluated for a history of severe hypertension. She was first found to be hypertensive 7 years earlier when she started experiencing headaches. Blood pressure control had always been difficult despite 3-4 antihypertensive medications. During a recent successful pregnancy, her blood pressure had to be controlled with high doses of labetalol. She was now using amlodipine, 5 mg, and bisoprolol, 2.5 mg, once daily.

The patient had no other medical history of note. She worked as a waitress and did not report illicit drug intake. None of her family members had hypertension or kidney disease, but her mother had neurofibromatosis type 1.

On examination, she was slim and appeared healthy, with blood pressure of 176/112 mm Hg. There was no cardiac murmur and the chest was clear on auscultation. Femoral pulses were weak, and both dorsalis pedis and posterior tibial pulses were absent on Doppler examination. She had an abdominal bruit. There were two 1 × 1-cm pinkish elevated nodules on her anterior abdominal wall. There were no other skin lesions. Serum creatinine level was 0.79 mg/dL (70 μmol/L) with an estimated glomerular filtration rate of 90 mL/min/1.73 m^2 (1.5 mL/s/1.73 m^2), calculated using the 4-variable Modification of Diet in Renal Disease

From the Departments of [1]Nephrology, [2]Radiology, and [3]Cardiology, Lancashire Teaching Hospitals NHS Foundation Trust, Preston, UK.

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Address correspondence to Alexander Woywodt, MD, FRCP, Department of Renal Medicine, Lancashire Teaching Hospitals NHS Foundation Trust, Royal Preston Hospital, Preston PR2 9HT, UK. E-mail: alex.woywodt@lthtr.nhs.uk

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Box 1. Causes of Renovascular Hypertension

- Atherosclerotic renal artery stenosis
- Fibromuscular dysplasia
- Arteriovenous malformation
- Thrombosis or embolism of the renal artery
- Cholesterol emboli disease
- Renal artery trauma
- Renal artery aneurysm
- Takayasu arteritis
- Aortic dissection with ostial involvement of the renal arteries
- Polyarteritis nodosa
- Giant cell arteritis
- Midaortic syndrome
- External compression of the kidney (Page kidney)
- Aortic thrombosis (Leriche syndrome)

Source: Textor.[1]

(MDRD) Study equation, modified for isotope dilution mass spectrometry (IDMS). Serum potassium level was 4.3 mEq/L (4.3 mmol/L), and C-reactive protein level was normal.

Imaging Studies

On ultrasonographic examination, the right kidney was 103 mm and the left kidney was 97 mm, with preserved parenchymal thickness and smooth outlines bilaterally. There was no hydronephrosis. The resistive index in distal segmental arteries was low bilaterally (0.48 on the right and 0.52 on the left side). The abdominal aorta had a funnel-like narrowing distal to the origin of the renal arteries, with a decrease in aortic diameter from 12.5 to 7.2 mm (Fig 1). A concomitant increase in blood velocity in the aorta from 190 to 564 cm/s was noted. Aliasing in both proximal renal arteries was noted (maximum velocities in the right and left renal arteries were 453 and 740 cm/s, respectively). Magnetic resonance angiography confirmed tapering of the aorta at the level of the renal arteries, with a diffusely narrowed irregular infrarenal portion of aorta from just below the origin of renal arteries to the aortic bifurcation (Fig 2). Computed tomographic angiography (CTA) of the abdomen was performed, which excluded external compression of the abdominal aorta by neurofibroma. The origins of both renal arteries showed significant stenosis, more severe on the left side (Fig 3A). There also was stenosis at the origin of the superior mesenteric artery (Fig 3B; Movies S1 and S2, provided as online supplementary material associated with this article at www.ajkd.org).

Diagnosis

The imaging diagnosis was midaortic syndrome with stenosis of the bilateral proximal renal arteries and superior mesenteric artery. A clinical diagnosis of neurofibromatosis type 1 was made on the basis of 2 neurofibromas and a first-degree relative with the disease.[2]

Clinical Follow-up

The patient continues to be healthy, and her kidney function has remained normal (serum creatinine, 0.81 mg/dL [71 μmol/L]; estimated glomerular filtration rate, 86 mL/min/1.73 m^2 [1.4 mL/s/1.73 m^2]). She currently uses amlodipine, 10 mg; nebivolol, 5 mg; moxonidine, 400 μg; and doxazosin, 4 mg, daily. Home blood pressures are 130/85-140/90 mm Hg, and there are no side effects from these drugs. A human geneticist confirmed our clinical diagnosis of neurofibromatosis and believed that genetic testing was not required.

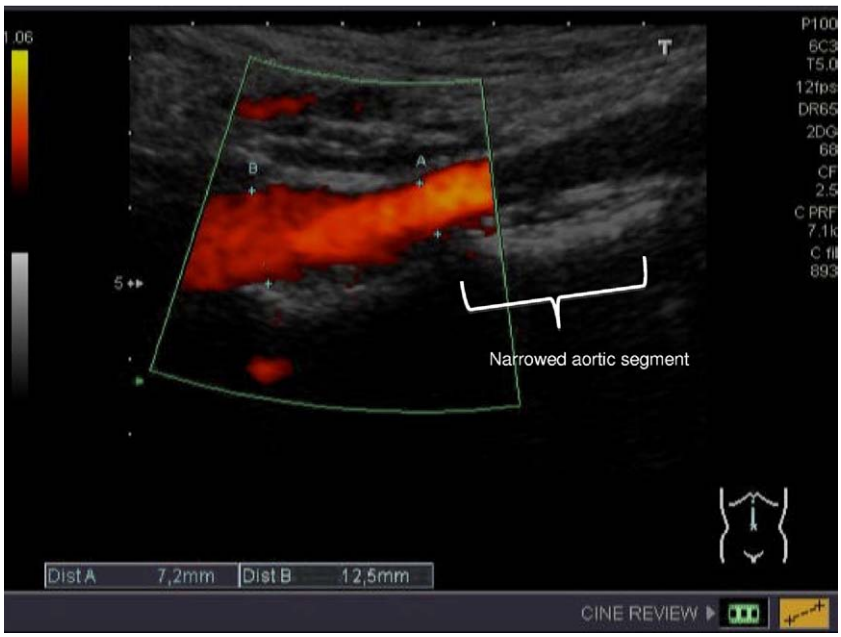

Figure 1. Power Doppler ultrasound of the abdominal aorta (longitudinal axis) shows funnel-like narrowing.