CLINICAL IMAGE

A 21-year-old woman with neurofibromatosis type 1, mid-aortic syndrome, and hypertension

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Neurofibromatosis type 1 (NF-1) is an autosomal dominant multisystemic neurocutaneous disorder with predominant features of café au lait macules, bone dysplasia, central nervous system gliomas, and neurofibromas. NF-1 and fibromuscular dysplasia are the most common causes of pediatric renal artery stenosis (RAS). In patients with NF--1, the renal arteries and aorta are the most frequently affected arteries, with the reported frequency of RAS ranging from 7% to 58%.

A 21-year-old female patient with NF-1 was referred to the Department of Hypertension for a detailed clinical reevaluation and consideration of surgical treatment of abnormalities in the aorta, as well as surgical revascularization of the right RAS. At the age of 10, the patient was first diagnosed with mid-aortic syndrome (MAS), right RAS, and hypertension. At that time, a decision was made to place a stent into the narrowed abdominal aorta. This procedure was complicated by the dissection of the aortic wall and by the necessity of stent implantation in the previously narrowed renal artery ostium. The early diagnosis of NF-1, based on clinical criteria, was then confirmed by genetic testing (heterozygous deletion in exon 39 of the *NF1* gene).

Physical examination on admission revealed previous findings. Ambulatory blood pressure monitoring showed normal daytime and nighttime blood pressure (mean, 126/73 mm Hg and 105/58 mm Hg, respectively) on 4 antihypertensive drugs (nebivolol, 5 mg/d; indapamide, 1.5 mg/d; amlodipine, 10 mg/d; and doxazosin, 2 mg/d). The patient had normal renal function at baseline, with serum creatinine levels of 80 μmol/l and an estimated glomerular filtration rate of 90 ml/min/1.73 m^2. However, renal scintigraphy showed decreased function of the right kidney with RAS (18%).

Plasma-free metanephrines were within the reference range. Considering the presentation of MAS on computed tomography angiography (FIGURE 1), a tentative decision to treat the patient surgically was made, together with a team of vascular surgeons and interventional radiologists.

NF-1 is the leading cause of symptomatic RAS in childhood and is associated with other vascular anomalies, including MAS and narrowing of the mesenteric arteries. Doppler duplex ultrasound and computed tomography angiography or magnetic resonance angiography are typically used for imaging of vascular lesions.[1] NF-1 should be differentiated from fibromuscular dysplasia, but both entities share common angiographic features with regard to the extent and distribution of arterial anomalies.[2] However, RAS in NF-1 is characterized by a higher frequency of ostial RAS and mid-aortic narrowing. Early detection of RAS or MAS can reduce serious cardiovascular manifestations in adulthood, and the treatment of aorto-renal disease in patients with NF-1 involves a combination of antihypertensive therapy, percutaneous angioplasty, and surgery. All methods, used in carefully selected patients, yield durable results.[3,4]

In summary, we report the case of an adult female patient with NF-1 and MAS, highlighting the clinical presentation and diagnostic workup of this rare disorder. Although rare, a high index of clinical suspicion for RAS should be raised in patients with hypertension, particularly when resistant to multidrug therapy, or the presence of other features of NF-1 syndrome.

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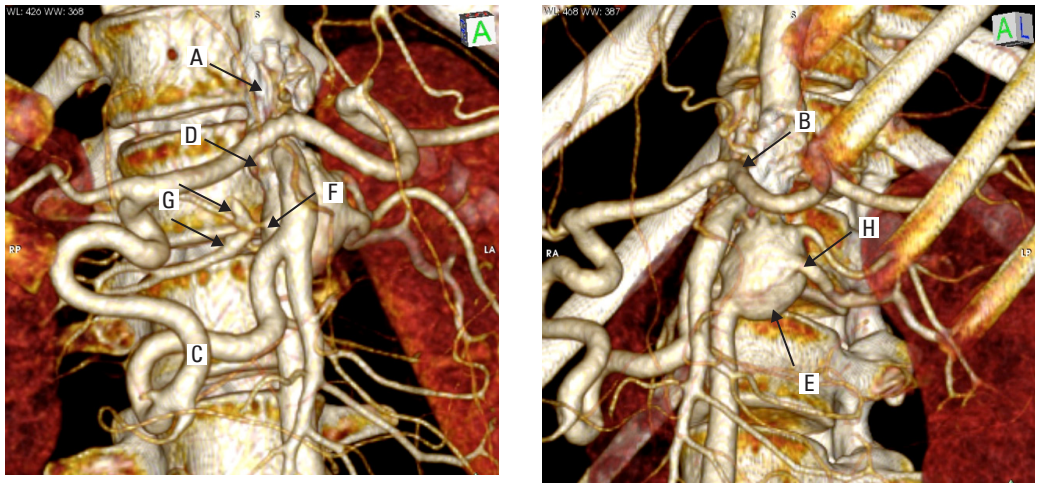

FIGURE 1 Computed tomography angiography of the renal arteries (volume rendering reconstruction); mid-aortic syndrome. Examination 11 years after aortic stenting. **A** – a stent visible at the level of the celiac trunk and upper mesenteric artery; **B** – critical stenosis of the celiac trunk origin; **C** – collateral circulation from the superior mesenteric artery to the celiac trunk; **D** – a small dissection channel present behind the stent (right-side presentation); **E** – poststenotic aortic dilation below the stent, at the level of the renal artery origins; **F** – the right renal artery with critical stenosis in the first segment; **G** – 2 small aneurysms of the right renal artery branches; **H** – 2 left renal arteries (patent, nonstenotic)

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