Long-segment thoracoabdominal aortic coarctation in a child with Down syndrome

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Midaortic syndrome is a rare vascular anomaly characterized by coarctation of the descending thoracic and abdominal aorta. Down syndrome is associated with multiple congenital cardiac malformations but is rarely associated with developmental vascular anomalies. Midaortic syndrome may result in severe renovascular hypertension that requires early intervention to prevent life-threatening complications. We report a child with Down syndrome who presented with occlusion of the aorta and was treated with aortic bypass. More than 4 years after the procedure, the patient's renal function remains normal, and there is no evidence of recurrent hypertension. Long-term follow-up is important to assess the benefits of surgical repair. (J Vasc Surg Cases 2015;1:171-3.)

In 1838, Pinel was the first to describe the physical and intellectual development of an individual with Down syndrome (DS).[1] DS affects one in 700 newborns[1,2] and is also associated with a number of major malformations. About half of individuals with DS may have congenital heart abnormalities,[3] including congenital stenosis in the proximal thoracic aorta (aortic coarctation), with an estimated prevalence of 1.2%.[4]

Schlessinger first described subisthmic narrowing of the aorta around the same time of the phenotypic description of DS, and the term midaortic syndrome (MAS) is currently used for obstructive lesions of the midaorta, regardless of etiology.[5,6] MAS is a rare anomaly, accounting for 0.5% to 2.0% of all aortic coarctations and has an incidence of one in 62,500 in autopsy studies.[5,7,8]

We describe a child with DS associated with MAS presenting as renovascular hypertension complicated by left ventricular hypertrophy and symptoms of heart failure. Consent was obtained from the patient and her parents to publish the images and clinical history.

CASE REPORT

A 7-year-old girl with a phenotype typical trisomy of the 21st chromosome and short stature was attended at the emergency ward with dyspnea, weakness, and no leg edema. She did not present with lower extremity fatigue. Arterial hypertension had been previously detected and treated with a combination of three different antihypertensive agents. Blood pressures measured in the upper extremities were 160/110 mm Hg, pulse was 130 beats/min, and a lack of the pulse was detected in the abdominal aorta and femoral arteries. She was hypoxic (saturated oxygen at rest was 70% on room air) and was intubated for respiratory distress.

Results of the complete blood count, serum electrolytes, urea, creatinine, liver function, and coagulation tests were all within normal reference ranges. There was evidence of left ventricular hypertrophy and mild diastolic dysfunction, and echocardiography showed a caliber reduction of the descending aorta. After 5 days, the patient was extubated and transferred to a regular floor. She was still hypertensive despite administration of furosemide (50 mg/d), milrinone (4.5 μg/d), amlodipine (5 mg three-times daily), carvedilol (3.125 mg once daily), and hydralazine (50 mg four-times daily).

These findings prompted a computed tomography angiography, which revealed an atresia of the descending aorta and its branches with robust collateral circulation (Fig 1). Distal to the atresia, the infrarenal aorta was 4.5 mm in diameter. After 3 weeks, the attempt to medically control her hypertension was not very successful.

The aorta was approached through a thoracophrenolaparotomy in the fifth intercostal space, followed by medial rotation of the viscera. After heparinization, the aorta was cross-clamped immediately above the diaphragm and immediately above the bifurcation. The aorta was opened for an end-to-side anastomosis. A retroperitoneal tunnel through which the graft was to be placed was created from the left thorax, through the left leaf of the diaphragm, and behind the left kidney to the infrarenal aorta. An oversized graft (Fig 2), compared with the initial distal aortic diameter, was used, anticipating growth of the child. In addition, a modest graft redundancy was allowed for later axial growth.

The patient was discharged on postoperative day 23 with normal plasmatic renal function (creatinine 0.43 mg/dL), and normotensive (124/79 mm Hg) on two antihypertensive agents. Four years after vascular surgery, computed tomography

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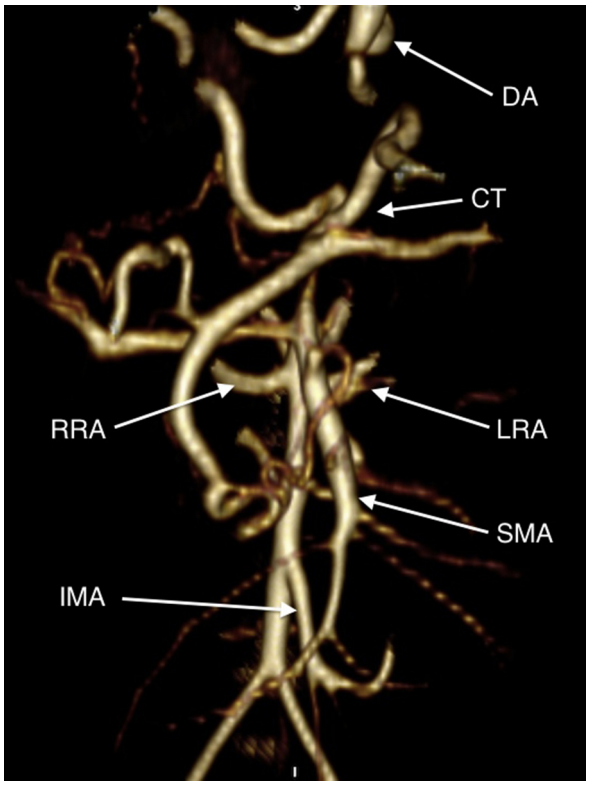

Fig 1. A preoperative a three-dimensional computed tomography angiography reconstruction demonstrates a segmental occlusion of the descending aorta to the midabdomen. A robust collateral circulation was seen for the infrarenal aorta and a retrograde flow into renal and mesenteric arteries. *CT,* Celiac trunk; *DA,* descending aorta; *IMA,* inferior mesenteric artery; *LRA,* left renal artery; *RRA,* right renal artery; *SMA,* superior mesenteric artery.

angiography showed a well-functioning aortoaortic bypass with a modest redundancy and no pseudoaneurysms (Fig 3).

At the time of this report, the child was 11 years old and did not need antihypertensive agents to control her normal blood pressure. Her renal function has remained normal throughout. She has an excellent quality of life, with normal growth and development.

DISCUSSION

Although uncommon in Western countries, lesions of the abdominal aorta, including MAS, inflammatory aortitis, and aneurysm, are well documented in children.[9] The incidence of renovascular hypertension in childhood is not known, but low blood pressure is reported in DS.[5,10] In addition, according to Greene et al,[11] patients with DS have a reduced risk of vascular anomalies compared with the general population.

MAS has multiple congenital and acquired etiologies. MAS is speculated to be a result of incomplete fusion or overfusion of the paired embryonic dorsal aortae during the fourth week of gestation.[12-14] Congenital causes include neurofibromatosis, William syndrome, Marfan syndrome, and mucopolysaccharidosis.[8] MAS may also be

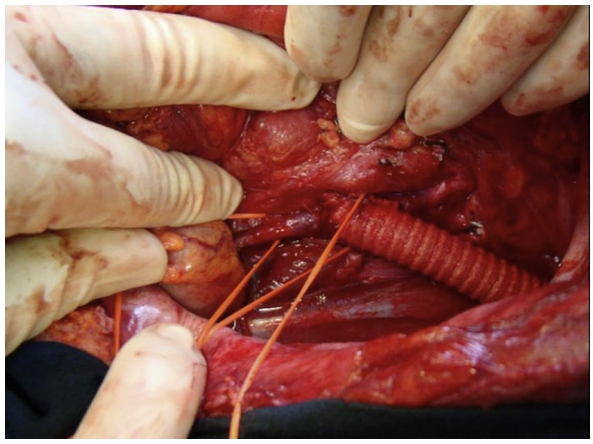

Fig 2. A synthetic graft was implanted in the distal end of the aortic segment. An aortoaortic bypass graft was performed with a 10-mm Dacron (DuPont, Wilmington, Del) tubular graft.

acquired, caused by Takayasu disease, arteritis, and rubella. The growth of a developmentally normal aorta may be disrupted by interference in cell growth by viruses or an inflammatory process.[15] Therefore, acquired lesions usually have evidence of an adventitial inflammatory reaction with fibrous proliferation.[5]

Depending on the location and extent of the coarctation, MAS may have many diverse clinical presentations. Patients with hemodynamically significant infrarenal coarctation may have exercise-related lower extremity fatigue, but true claudication is rare.[16] On one hand, these patients are more likely to be diagnosed at an older age due to superior-to-inferior mesenteric[8] and superior-to-inferior epigastric (Fig 1) collateralization. On the other hand, patients with severe renovascular hypertension because of suprarenal or inter-renal stenosis are diagnosed at an earlier age.[8]

Two classification systems have been developed to describe the variation of the disease and to help to define the appropriate operative strategy. An anatomic classification system divides MAS into hypoplastic and segmental groups.[15] The affected aorta in relation to the renal arteries was used to create subgroups: suprarenal, interrenal, and infrarenal.[15,17] Another classification system based on the on the relationship between the location of the abdominal coarctation to the renal arteries (suprarenal vs infrarenal) and whether renal artery stenosis was present or absent.[17]

The prognosis of aortic hypoplasia when associated with renovascular hypertension is generally poor in untreated patients, depending on the location with respect to the renal arteries and the severity of the secondary complications.[15] Initial standard MAS management includes treatment with antihypertensive medication and frequent monitoring by cardiology and nephrology teams until the patients are adolescents and can be treated definitively using an appropriate surgical procedure.[18] However, evidence of end-organ disease, such as congestive heart failure

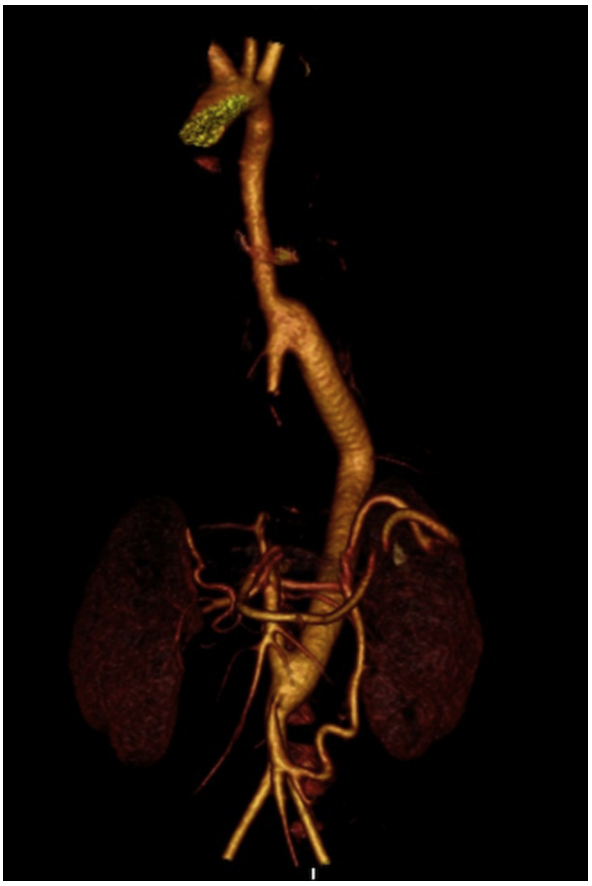

Fig 3. Posterior-anterior view of a three-dimensional computed tomography angiography reconstruction performed 4 years after surgery shows the graft with normal structure and function of the distal aorta and its branches.

or renal failure, is an indication for early operative intervention.

Aortic occlusive disease can be treated by bypass, aortoplasty, or percutaneous transluminal angioplasty (PTA). However, PTA is not likely to achieve a favorable outcome in patients with longer segments or hypoplasia.[17] Failures after PTA alone suggest that stent placement can be used to overcome the postdilatation recoil of aortic narrowing that is remote from the visceral arteries.[16] In addition, surgical repair is usually required because stents and stent grafts cannot grow with the child and restenosis is frequent after angioplasty.[9] Stanley et al[16] suggested using a conduit 60% to 70% of the size of the patient's adult aorta, using 8-mm to 12-mm grafts in young children.

CONCLUSIONS

The occurrence of aortic coarctation may be an incidental finding but may also represent a manifestation of the vascular malformation. Cumulative reviews of the literature are necessary to define the spectrum of their clinical presentation due to the infrequent occurrence. Because many similar cases in the literature are reported without long-term follow-up, the ultimate results of operative therapy remain unknown. In this patient, the diseased aortic segment did not involve the perirenal aorta, and an anastomosis to the infrarenal aorta improved renal artery blood flow via retrograde aortic flow. We report this case to add to the already broad range of the DS phenotype.

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