An Abdominal Aortic Aneurysm in an 8-Month-Old Girl with Tuberous Sclerosis

S.-B. Moon, W.-Y. Shin, Y.-J. Park, S.-J. Kim*

Department of Surgery, Seoul National University College of Medicine, Seoul, Republic of Korea

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Tuberous sclerosis; Abdominal aortic aneurysm

Abstract
The association between an abdominal aortic aneurysm (AAA) and tuberous sclerosis (TS) is rare. An 8-month-old girl presented with a seizure, and the clinical evaluation revealed TS. An abdominal evaluation showed a 3-cm infrarenal AAA. A normal diameter of infrarenal aorta for an 8-month-old girl is about 6 mm. The patient underwent an open repair with a polytetrafluoroethylene (PTFE) prosthesis. The pathology showed a loss of elastin fibres in the media of the aorta. The graft was patent on computed tomography (CT) angiography, performed 4 months after the operation. However, the patient died of complications related to seizures 5 years after the surgery. The graft remained patent until the time of death.

Introduction
An abdominal aortic aneurysm (AAA) is usually found in the elderly; it is rarely diagnosed in children. Most cases of childhood AAAs are associated with systemic diseases, such as Takayasu’s arteritis and Kawasaki disease, and connective tissue disorders, such as Ehlers–Danlos syndrome.1 Tuberous sclerosis (TS) is an autosomal-dominant genetic disease characterised by seizures, mental retardation and facial adenoma sebaceum. Here we report a case of an 8-month-old girl with TS and an AAA.

Case Report
An 8-month-old girl was admitted to the hospital with seizures and loss of consciousness. There was neither a significant medical history nor a familial history of TS. On physical examination, there were multiple hypopigmented macules on the extremities and the back, along with adenomata sebacea on the face. Physical examination revealed that the abdomen was normal. The blood pressure was within the normal range. Bilateral femoral pulsations could be easily palpated. The brain magnetic resonance imaging (MRI) showed typical calcified nodules associated with TS. Cardiac echocardiography showed a 6-mm mass in the papillary muscle of the right ventricle suggestive of a rhabdomyoma. Abdominal ultrasound revealed an AAA, and computed tomography (CT) scanning demonstrated...
a large, infrarenal saccular aneurysm measuring 30.6 mm in width and 37.8 mm in length (Fig. 1A). The aortic bifurcation was not involved.

Laparotomy revealed an aneurysm measuring $3 \times 3.5 \times 3$ cm. Endo-aneurysmal graft replacement, with a PTFE prosthesis (Hemashield${}^{\circ}$) of 10 mm diameter, was performed. The inferior mesenteric artery (IMA) was ligated, and the graft was sutured in an interrupted manner. Calcification was absent in the aortic wall. Pathological examination revealed the loss of elastin fibres in the media of the aorta (Fig. 1B). The postoperative recovery was uneventful. A CT scan confirmed that the graft was patent for 4 months after the operation (Fig. 2). However, the patient died of complications secondary to uncontrolled seizures 5 years after the operation. There were no further symptoms associated with graft occlusion or recurrence of the aneurysm.

Figure 2 Three-dimensional volume-rendered image acquired 4 months after the operation. The graft is patent and the distal flow is good.

Discussion

TS is a genetic disorder with autosomal-dominant inheritance; it is common in children. It affects a variety of organs, including the brain, heart and kidneys. Although previously recognised, the association of TS with an aortic aneurysm is rare. In a review of 15 TS patients with an aortic aneurysm reported by Jost et al., the aneurysm associated with TS was usually large at presentation and rapidly progressive; in addition, in one-third of the cases the aneurysm had ruptured. Therefore, an aortic aneurysm should routinely be ruled out in patients with TS. If an AAA is identified, it should be managed by elective repair rather than by observation. Early detection of the aneurysm, soon after the diagnosis of TS, and prompt repair could prevent fatal complications.

In an 8-month-old girl, the diameter of the infrarenal aorta is about 6 mm and grows to 12 mm at 15–18 years of age according to the nomogram by Munk et al. We used a 10-mm prosthesis because we had planned a second operation, had the patient survived, to provide a larger graft during the adolescent period.

Figure 1 (A) Three-dimensional volume-rendered image demonstrates the infrarenal saccular aneurysm (measurements in millimeters) and (B) histological image of the aortic wall (H&E; $\times$ 100).

The pathogenesis of aortic aneurysms associated with TS is unknown; however, abnormality of connective tissue has been suggested as a possible cause. Loss of elastin fibres has been noted in aortic aneurysms associated with TS, as in cases with Marfan’s syndrome; our patient had a similar histopathology. The occurrence of a new aneurysm, superior or inferior to the graft after aneurysm repair, might be explained by an innate weakness of the aortic wall associated with TS.

We treated AAA diagnosed in an 8-month-old girl with TS; the short-term outcomes were good. However, the patient died from complications unrelated to AAA. Therefore, regular surveillance for postoperative AAA is important for such cases.

Conflict of Interest

The authors do not have any conflict of interest.
References


