Development of Aortic Aneurysms in Familial Supravalvar Aortic Stenosis

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SUMMARY. In a male patient with supravalvar aortic stenosis (SAS) and peripheral pulmonary arterial stenoses, aortic aneurysms developed between his first and fourth years of life. He died five days after correction of SAS and resection of aneurysms. Histologic examination revealed disarrangement as well as severe degeneration of elastic fibers in the aortic wall. This tissue defect is probably inherited through an autosomal dominant mechanism. It may lead to aneurysm formation. Only one case of SAS with aortic aneurysm has been previously reported.

KEY WORDS: Supravalvar aortic stenosis — Aortic aneurysms — Defect of elastic fibers — Autosomal dominant transmission

Supravalvar aortic stenosis (SAS) may occur sporadically or with familial aggregation [1, 4, 5, 12]. Three different anatomic types have been described [1–3, 11]. Degenerative changes of elastic lamellae of the aorta have been reported in localized SAS [7] as well as in the nonaffected aortic wall [9, 10]. We report the development of aortic aneurysms that probably was due to an inherited defect of the aortic wall structure in a young patient with SAS.

Case Report

Family History

The boy was a member of a family with known congenital heart disease through three generations. His grandfather had been operated on for supravalvar aortic stenosis at the age of 34 years and had a second operation for mitral valve replacement at the age of 47. He died at 52. His two sons were unaffected, but both his daughters had been operated on for supravalvar aortic stenosis at the ages of 14 and 15 years, respectively. Both also have peripheral pulmonary arterial stenoses. The brother of our patient died unexpectedly at the age of three months. A postmortem examination showed supravalvar aortic stenosis with diffuse hypoplasia of the aorta and pulmonary artery with extreme thickening of the wall of these vessels. All members of the family have normal facial appearance and normal intelligence.

Clinical Course

Our patient was investigated for the first time at the age of 11 months. Cardiac catheterization revealed the diagnosis of supravalvar aortic stenosis with diffuse hypoplasia of the aorta (Fig. 1a and b) and peripheral pulmonary arterial stenoses. His right and left ventricular pressures were 90 and 150 mmHg, respectively, and he had a 70-mmHg gradient across the aortic stenosis. No surgery was attempted and he was followed at regular intervals. He developed normally and was without complaints to his fourth year of life when fatigue on effort was noted. The ECG at that age showed left ventricular hypertrophy with a strain pattern and real-time echocardiography showed obstructive cardiomyopathy of the left ventricle with asymmetric septal hypertrophy, but without systolic anterior movement of the anterior mitral leaflet.

A second catheterization was performed at the age of four years. The patient still had a 70-mmHg gradient across the supravalvar stenosis. The left ventricular apex was not entered. Right ventricular systolic pressure was 80 mmHg. Left ventricular angiography confirmed the echocardiographic diagnosis of midventricular systolic obstruction due to asymmetric septal hypertrophy, but also showed three large aneurysms of the ascending aorta situated between supravalvar stenosis and origin of the innominate artery (Fig. 1c and d).

At operation, the supravalvar stenosis was relieved by patching. All three aneurysms were resected together with parts of the ascending aorta that was partly replaced by a Dacron graft. On examination, the resected material was markedly thickened and had a yellowish atherosclerotic appearance. Histology of the stenotic ring and the aneurysm wall revealed marked degeneration of the median layer. Elastic fibers were broken and conglom erated in some areas, while in others they were nearly missing and replaced by fibrous tissue (Fig. 2a). There were gross thickening and fibrosis of the intima. Resected tissue between the
Fig. 1. Left ventricular angiocardiogram, anteroposterior and lateral views. (a and b) At age 11 months. SAS with diffuse hypoplasia of thoracic aorta. (b and c) At age four years. Hypertrophic obstructive cardiomyopathy, SAS, and three saccular aneurysms of the ascending aorta.

aneurysms showed fraying and disarrangement of the elastic fibers (Fig. 2b) together with fibrosis. The patient died five days after operation of low-output cardiac failure. Postmortem histology of the left ventricular myocardium demonstrated secondary hypertrophy without gross abnormalities. Histology of the descending aorta was normal.

Discussion
There are three forms of clinical presentation in SAS. Nonfamilial sporadic cases with normal facies and intelligence, a nonfamilial syndrome with abnormal facial appearance and mental retardation [1,