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SUPRAVALVULAR AORTIC STENOSIS*

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Supravalvular aortic stenosis in infancy, childhood and adolescence is a rare condition, presumably congenital in origin[1-3]. The pathognomonic lesion is an hourglass-shaped deformity or a localized fibrous diaphragm of the ascending aorta above the valve or tubular narrowing of the entire aorta[1-5]. The occurrence of the disorder may be familial, associated with characteristic facies and mental retardation, sporadic or, rarely, the result of congenital rubella[6-10]. A rare type of congenital supravalvular aortic stenosis with normal facies and intelligence, with hypercalcemic attacks, encountered and treated in our clinic, is the subject of this paper.

Case Report

N.C., a 17-year-old Caucasian female university student was referred to the Hacettepe University Cardiology Department for evaluation. A cardiac murmur had been diagnosed at the age of 6; however she gave no history of rheumatic fever.

She appeared to have normal facies and growth. Physical examination revealed an arterial blood pressure of 100/80 mmHg. No palpable differences in the pulsus and blood pressure were present in the upper and lower extremities. No precordial pulsation was observed. The first heart sound was normal and the second sound was split normally. There was a prominent systolic thrill over both carotid arteries and at the suprasternal fossa. A IV/V° harsh ejection type systolic murmur was heard at the same region maximally. This murmur radiated over the entire precordium and especially along the right upper sternal border with lower intensity. There was no systolic ejection click and no diastolic murmur.

Laboratory examination results were as follows: hemoglobin 14.20 g/dl, leukocytes 7000/mm^3 and serum calcium levels were 10-12.3 mg/dl (normal level is 9-11 mg/dl). A telecardiogram revealed a left ventricular enlargement without postste-

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notic dilatation of the ascending aorta (Figure 1). The ECG showed left ventricular enlargement (Figure 2). The data shown in the table below were obtained by cardiac catheterization.

TABLE I

Results of Cardiac Catheterization

Place of catheter	Pressures
Pulmonary capillary	10
Pulmonary artery	25/10
Right ventricle	25/7
Right atrium	7
Left ventricle	210/10
Aorta, proximal, distal	210/65-100/65

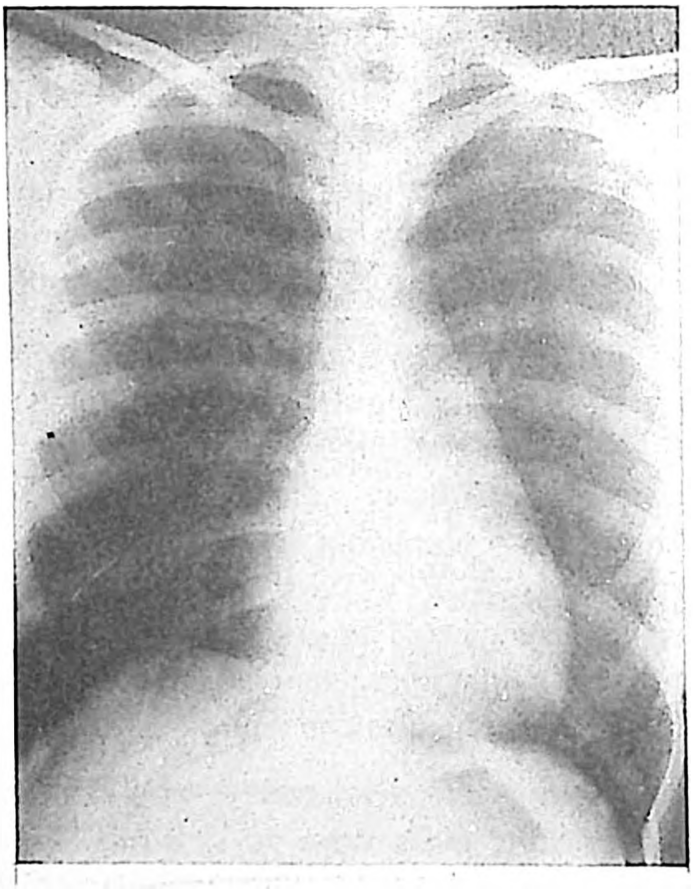

Figure 1

Chest roentgenogram.

There was no gradient over the aortic and pulmonic valves. A pull-back tracing from the left ventricle to the aorta showed a narrowed area 1.5-2 cm in length, just above the valves distal to the coronary artery orifices, with a systolic gradient of 110 mmHg (Figure 3). The left ventricular angiography showed ventricular hypertrophy,

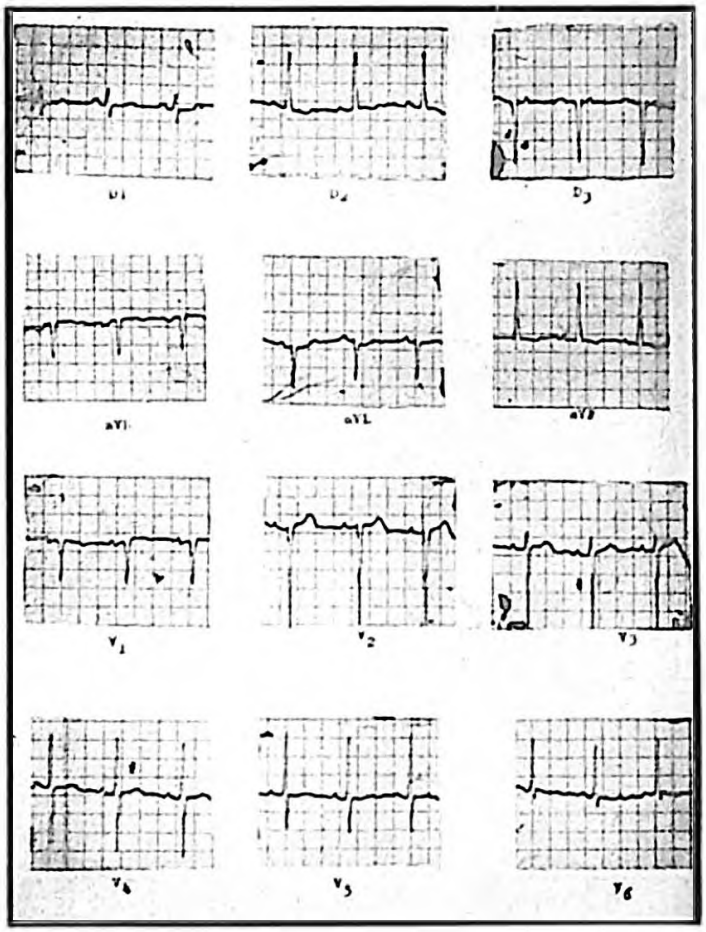

Figure 2

Electrocardiogram.

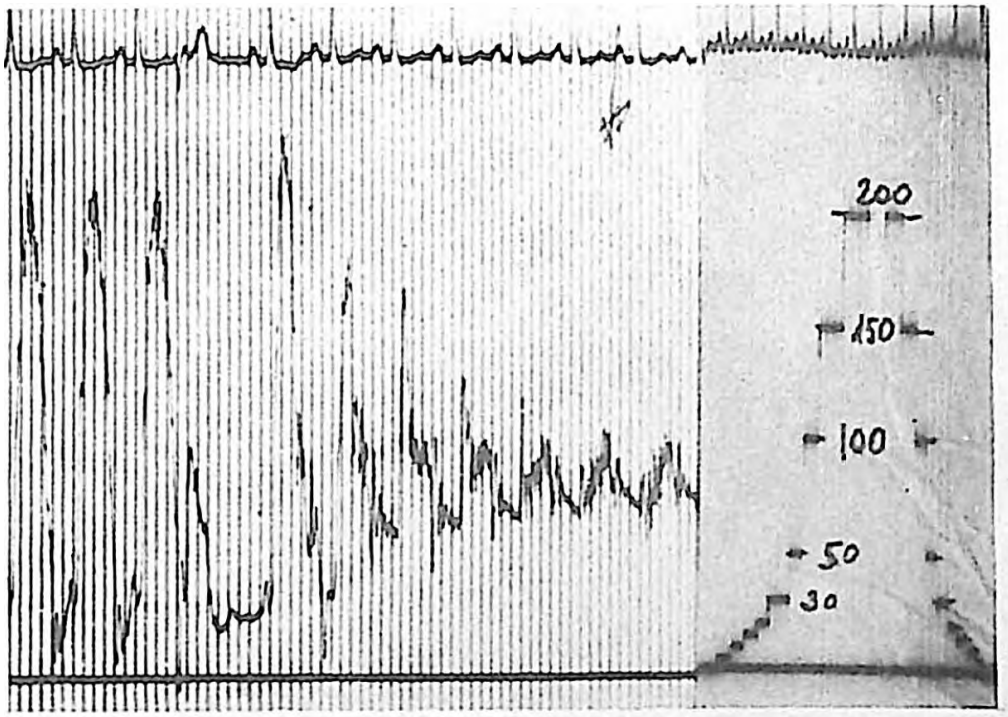

Figure 3

A pull-back tracing from the left ventricle to the aorta.

however with good contractions, and there was no sign of subvalvular narrowing (Figure 4). Supraaortic angiography demonstrated that the aortic lumen narrowed with an hourglass obstruction just above the valves, distal to the coronary artery orifices. The lumen was approximately less than 1 cm for a distance of about 2 cm (Figure 5). The coronary arteries looked larger than normal. There was neither systemic arterial nor pulmonary arterial stenosis.

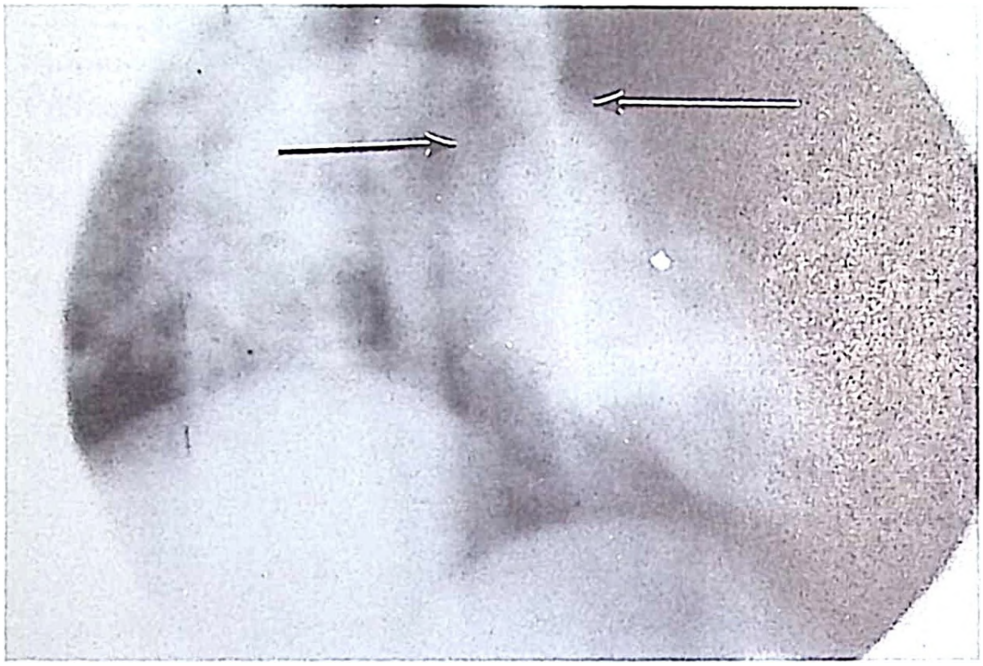

Figure 4

Left ventricular angiography shows annular stenosis of the ascending aorta.

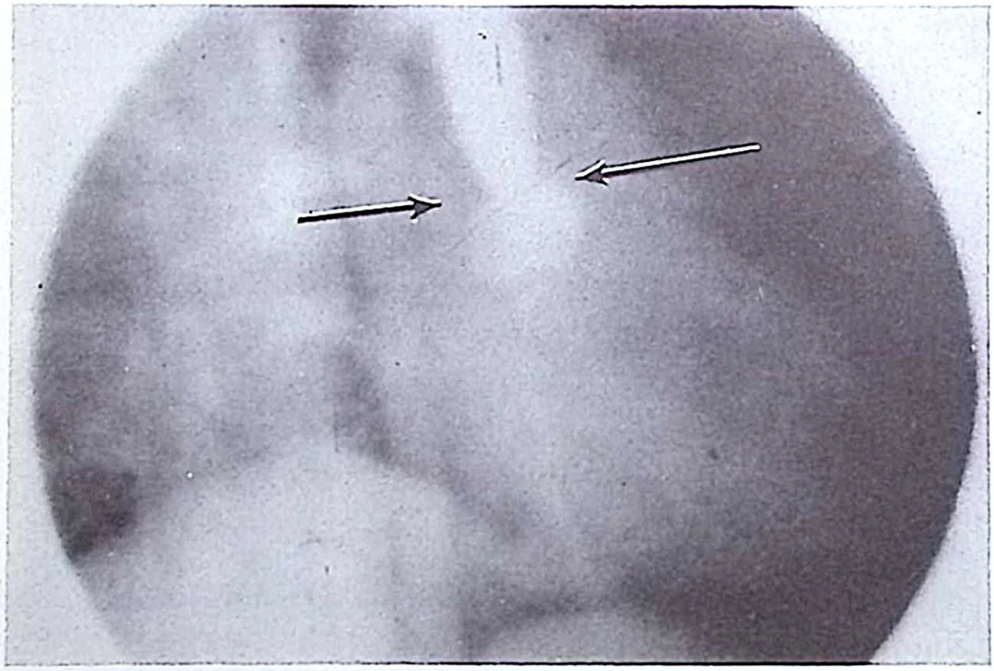

Figure 5

Aortic root angiography shows an hourglass obstruction above the valves.

After diagnosis the patient underwent open heart surgery. A median sternotomy was performed and left femoral artery cannulation was utilized for the cardiopulmonary bypass with general hypothermia (30°C). The aorta looked quite small. An aortotomy was performed very close to the valve. One cm distal from the valve an annular stenosis was observed; the aortic wall and the cusps looked rather thickened. This portion was resected and a teflon patch 4.5 × 2.5 cm in size was applied with teflon-supported pledgets to the wall. The postoperative course of the patient was uneventful and no aortic insufficiency was observed.

Discussion

Since its first description by Archer in 1878, more than 250 patients with supravalvular aortic stenosis have been reported[1-15]. In 1961 Williams et al[15] described four cases with the triad "characteristic facies, mental retardation and supravalvular aortic stenosis". Characteristic facies consist of a high prominent forehead with protruding lips, epicanthal folds, low set ears and strabismus. Therefore, the terms elfin-face syndrome or Williams syndrome are used to designate this entity[1,7]. This syndrome may also present itself in different clinical and pathological forms. The typical elfin facies may be absent, as in our case.

Although the majority of cases are sporadic in occurrence, the syndrome has been observed in monozygotic twins, in siblings, and in second cousins[2,3,6,14]. Transmission from father to son and daughter has suggested an autosomal dominant inheritance. Under this syndrome three subgroups are observed: normal facies appearance and intelligence with no familial inheritance, normal facies with mental retardation and familial inheritance, and abnormal facies with mental retardation[10]. Our patient is in the first subgroup.

The etiology of the syndrome remains obscure. It has been associated with idiopathic infantile hypercalcemia and, rarely, as a result of congenital rubella[8-10]. Attention has been drawn to hypercalcemia resulting from vitamin D intoxication due to high dose vitamin D prophylaxis during pregnancy and infancy[12]. There is also the possibility of an inborn error of metabolism or a sensitivity to vitamin D excess. Hypercalcemic episodes may sometimes result in renal damage, vascular lesions and mental deficiency[8,9]. As in our case, hypercalcemic periods may result in vascular damage such as supravalvular aortic stenosis.

The degree of supravalvular stenosis may vary from mild to severe. Three types of supravalvular stenosis have been described[1,11]: a diffuse hypoplasia of the ascending aorta, a membranous obstruction, and an hourglass stenosis[5]. We observed an hourglass type obstruction. In 1962 Beuren et al[3,4] demonstrated two additional features, namely peripheral systemic and peripheral pulmonary arterial stenosis. However, none of these entities was present in our case. Prolonged local hyperten-

sion is blamed for the dilatation in the coronary arteries observed in the majority of cases, as in ours[9,11]. Additionally, mitral insufficiency and mitral valve prolapsus may be seen with this syndrome[6,15]. None of these findings was present in our case.

After the diagnosis of supravalvular aortic stenosis is made, surgical correction should be advised as soon as possible. As in our case, resection of the obstruction and patch-plasty at the narrowed site of the aorta may be performed successfully. This procedure relieves the high pressure on the coronary arteries and improves the left ventricular function.

Summary

In this paper a rare hourglass type of supravalvular aortic stenosis with normal facies and intelligence is presented. The patient was operated on utilizing a cardiopulmonary bypass and resection of the obstruction. Patch-plasty at the narrowed site of the aorta was successfully performed. This procedure relieves the high pressure on the coronary arteries and improves the left ventricular function. Therefore, the operation should be advised as soon as possible after the diagnosis is made.

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