Echocardiographic Detection of Supravalvular Aortic Stenosis

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SUMMARY

The echocardiographic features of supravalvular aortic stenosis of the segmental or hourglass variety is described for the first time. Utilizing a strip recorder and an M mode or sector scan the diameter of the aortic lumen is demonstrated to narrow at the stenotic area just distal to the aortic valve. As the transducer sweeps further cephalad the aortic root lumen widens to a normal diameter.

Additional Indexing Words:
Left ventricular outflow obstruction Aortic root

SINCE THE DEVELOPMENT AND POPULARIZATION OF the technique for examining the heart by pulsed ultrasound,1-4 echocardiography has become a valuable and proven noninvasive method for the diagnosis and evaluation of congenital and acquired cardiac abnormalities.5-8 The echocardiographic features of valvular,9-10 subvalvular aortic stenosis of the discrete membranous11 as well as hypertrophic muscular12-14 variety have been reported. We report the echocardiographic features of the segmental or hourglass variety of supravalvular aortic stenosis.

Report of Case

K.V., a 9-year-old Caucasian male product of a normal pregnancy and delivery, was noted to have a systolic murmur at birth. He was followed with the diagnosis of probable ventricular septal defect and valvular pulmonic stenosis until age 3 when the clinical diagnosis of supravalvular aortic stenosis was first entertained on the basis of his facies, apparent mental retardation, and the systolic ejection murmur. At age 6 psychological testing revealed an IQ of 61 on the Stanford-Binet Form L-M. He was first evaluated in the Cardiology Clinic at Brooke Army Medical Center at age 8.

Physical examination revealed a pleasant but hyperactive acyanotic male with a narrow face, high forehead, slightly low-set ears and wide gaps between his teeth. The indirect blood pressure was 100/80 mm Hg in both arms. A thrill was palpable in the suprasternal notch and over the right carotid artery. The apical impulse was at the midclavicular line. S1 was normal and S2 was narrowly but physiologically split. There were no clicks or gallops. A grade III/VI harsh systolic ejection murmur was best heard along the right sternal border in the second intercostal space but was also heard at the left sternal border and the apex where it had a more holosystolic quality. The rest of the examination was normal. The ECG was normal and the cardiac series did not demonstrate cardiomegaly.

Left and right cardiac catheterization included continuous pressure recording during pullback of the arterial catheter from the left ventricle to the ascending and transverse aorta. Damping of the pressure curve occurred as the catheter was withdrawn from the aortic valve area to the ascending aorta and a 15 mm Hg peak systolic gradient was recorded across the stenotic area (fig. 1). The stenotic supravalvular area was confirmed by left ventricular cineangiography in the 60° LAO projection (fig. 2).

The echocardiogram was obtained using a Smith Kline Ekoline 20 while the patient was in the recumbent position. The complete echocardiographic tracing was recorded on an Electronics for Medicine recorder. A continuous recording was obtained while the transducer was directed sequentially from the left ventricle to the mitral valve and aortic root. As the echoes of the aortic valve cusp disappeared, narrowing of the aortic root occurred. The diameter of the aorta at the valve level was 2.4 cm compared with a 1.3 cm diameter at the supravalvular stenotic area. As the sweep of the transducer was continued cephalad the aortic root widened to 2.1 cm (fig. 3). The echographic pattern was quite reproducible.

Discussion

Supravalvular aortic stenosis is a congenital deformity of the ascending aorta which originates just distal to the level of the origins of the coronary arteries. There are three types of supravalvular aortic stenosis.15 The localized, segmental, or hourglass variety is the most common16 and is characterized by extreme thickening of the medial layer of the ascend-
ing aorta associated with an hourglass deformity of the external aspect of the involved segment.\(^{15}\) The hypoplastic or diffuse type is the second most common type and is characterized by hypoplasia of the entire ascending aorta.\(^{15}\) The least common is the membranous type which consists of a single diaphragm composed of fibrous tissue and containing a single perforation.\(^{15}\)

It was not until 1958 that clinical attention was focused on the congenital malformation of supravalvular aortic stenosis.\(^{16}\) In 1961 Williams and co-workers\(^{17}\) reported the association of mental retardation and elfin facies in patients with supravalvular aortic stenosis. In 1963 Black and Bonham Carter\(^{18}\) focused attention on the association of infantile hypercalcemia and elfin facies. Other reports\(^{19-23}\) have confirmed the association of elfin facies, infantile hypercalcemia, mental retardation and supravalvular aortic stenosis. When present, the triad of left ventricular outflow murmur, mental retardation and the characteristic elfin facies should suggest the diagnosis of supravalvular aortic stenosis.

In patients with normal mentation and facies the differential diagnosis of valvular, subvalvular and supravalvular aortic stenosis is a more formidable task. In recent reviews\(^{24-26}\) emphasis has been placed on the presence or absence of an ejection click, the carotid pulse contour, location of the murmur, and the presence or absence of aortic insufficiency. The diagnosis of discrete membranous and hypertrophic muscular subvalvular aortic stenosis can be confirmed by echocardiography.\(^{9,11-14}\) Although the echocardiographic criteria for the diagnosis of valvular aortic stenosis have been described there has been little

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**Figure 1**

Continuous pressure recording during pullback of the catheter from the left ventricle across the aortic valve and into the supravalvular stenotic area. The pressure curve dampens as the catheter is withdrawn into the supravalvular stenotic zone, and there is a 15 mm Hg gradient recorded.

**Figure 2**

Frame of left ventricular cineangiogram in 60° LAO projection demonstrating a narrowed area (hourglass appearance) just distal to the aortic valve and a normal sized aorta distal to the stenosis.

**Figure 3**

Continuous strip chart recording of echocardiogram as the transducer sweeps cephalad from the mitral valve demonstrating the narrowing of the aorta from 2.4 cm to 1.3 cm at the stenotic area and subsequent diameter of 2.1 cm in aorta distal to the stenosis. There are increased echoes at the site of the stenosis.
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clinical application of the method because of the inconsistency with which the aortic valve cusps can be recorded. Consequently confirmation of the diagnosis of valvular or supravalvular aortic stenosis has necessitated cardiac catheterization of the left ventricle and angiography. By using a strip chart recorder and an M mode sector scan the diameter of the lumen of a major portion of the ascending aorta is easily recorded by echocardiography. As in the patient presented, the diagnosis of supravalvular aortic stenosis of the segmental or hourglass type can be made when the lumen of the aorta narrows just above the aortic valve and then returns to a normal diameter more distally. It is anticipated that in the diffuse or hypoplastic variety of supravalvular aortic stenosis there would be a persistent narrowing of the aortic root lumen as has been demonstrated in two patients with aortic atresia and hypoplastic left ventricle.27 Further observations will be required to confirm this postulate.

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