Supravalvular aortic stenosis with sudden cardiac death

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ABSTRACT

Sudden cardiac death (SCD) most commonly results from previously undiagnosed congenital, acquired, or hereditary cardiac diseases. Congenital aortic valvular, subvalvular, and supravalvular disease with left ventricular outflow tract obstruction is an important preventable cause of sudden death. This report documents sudden death presumably due to acute myocardial ischemia in a young male with an undiagnosed supravalvular aortic stenosis (SVAS) due to a rare association of isolation of coronary sinuses of Valsalva. Congenital supravalvular pulmonary stenosis and mitral valvular dysplasia were also present.

Keywords: Congenital aortic stenosis, elastin arteriopathy, supravalvular aortic stenosis, sudden cardiac death, sinus of Valsalva isolation

INTRODUCTION

Each year; infants, children, and young adults die suddenly and unexpectedly, which are unfortunate and distressing events for their families and communities. In this context, sudden cardiac death (SCD) accounts for a significant proportion.[1] SCD is defined as sudden, unexpected death either within 1h of symptomonset (event witnessed) or within 24 h of having been observed alive and symptom-free (unwitnessed event) in a person with known or unknown cardiac disease, and it most commonly results from a previously undiagnosed congenital or hereditary conditions such as coronary arterial anomalies, inherited cardiomyopathies, or primary cardiac rhythm disorders.[2] Congenital aortic valvular, subvalvular, and supravalvular disease with left ventricular outflow tract obstruction is an important preventable cause of SCD. This report documents sudden death in a young male with supravalvular aortic stenosis (SVAS) with a rare association of isolation of coronary sinuses of Valsalva.

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CLINICAL SUMMARY

A 21-year-old male was admitted with sudden-onset severe chest pain and acute shortness of breath to the Emergency Services of our tertiary care hospital. He apparently had a "valvular abnormality", for which surgery had been advised. However, there were no details available and therapeutic option had not been pursued due to financial constraints. He expired within 20 min of hospital admission; no investigations could be performed.

A complete autopsy was performed. On external examination, he was of average build. Remarkable findings were seen in the heart. The heart was moderately enlarged and weighed 480 g. There was marked biventricular enlargement because of myocardial hypertrophy [Figure 1a]. A cross-section through the midportion of the ventricles revealed circumferential subendocardial congestion of the left ventricular myocardium. The right coronary artery had a dominant distribution and all the arteries were patent. The interatrial and interventricular septums were intact. The os infundibulum was narrow and measured 1.1 cm in diameter, while the infundibular chamber was small in size (2.6 × 1.1 cm) and lined by pearly white endocardium. The pulmonary valve was tricuspid; stenosis (diameter of 0.6 cm) was produced by a circumferential membrane growing over the cusps [Figure 1b]. The mitral valvular leaflets were dysplastic with flattening of papillary muscles, especially towards their apices. The chords attached to the posteromedial commissure were shortened and completely fused with

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each other [Figure 1c]. The circumference at the site of attachment of the semilunar cusps of the aortic valve was 3.7 cm, while at the sinotubular junction, it was 2.2 cm. At this area, there was a circumferential firm, grey-white ridge with a height of 0.6 cm [Figure 2a]. The noncoronary cusp (1.8 × 0.9 cm) was markedly thickened with rolled margins and prolapsed into left ventricular cavity. A jet lesion of aortic regurgitation was present below this. The thickened left coronary cusp (1 × 1 cm) was totally fused with supravalvular ridge, thus isolating its sinus with left main coronary arterial ostium [Figure 2]. The right coronary cusp (1.1 × 1 cm) was also similarly fused, leaving a central probe-patent opening, which was the only communication between aortic lumen and right sinus of Valsalva/right coronary arterial ostium [Figure 2]. Sudden death presumably occurred due to myocardial ischemia, seen as circumferential congestion in the subendocardial half of the left ventricular myocardium.

DISCUSSION

Congenital AS occurs due to narrowing of the outflow tract at, below, or above the aortic valve constituting 5-10% of all congenital defects. Among these, SVAS is the rarest subtype, accounting for about 0.5% of congenital heart diseases and 5% of congenital AS, and this undetected anomaly had produced SCD in our young patient. Most of the cases of SVAS are associated with Williams-Beuren syndrome with cognitive, behavioral, and phenotypic abnormalities.[3,4] Our patient did not have any such features, and hence would be an example of nonsyndromic SVAS, seen as a familial autosomal dominant form or sporadic isolated occurrence.[5] The underlying mechanisms common to all the three settings is mutation in the elastin gene (ELN) located on 7q11.23 region, with or without disruption of the neighboring genes.[6] This results in "elastin arteriopathy", characterized by paucity of elastic fibers with pathological malalignment, smooth muscle hypertrophy, and increased collagen content in the media.[7] Predictably but disproportionately, the ascending aorta is affected, though other aortic segments, and even the visceral arteries can be involved. Our patient did not have involvement of other arteries; also, genetic analysis could not be done. The severity of SVAS varies and manifests morphologically as an hour-glass deformity of the ascending aorta, discrete ring-like thickening at the sinotubular junction, or variable hypoplasia of the aortic segments. The discrete membranous obstruction shows stellate mesenchymal cells and collagen and elastic fibers in a background of mucopolysaccharides, as seen in the case presented.

Majority of the patients with nonsyndromic SVAS remain asymptomatic unless associated with other cardiac malformations.[5,8] They become symptomatic before the age of 20 years with signs and symptoms similar to valvular AS, largely due to progression of the stenotic lesion and consequent sequelae.[8] However, such patients are also at risk of sudden death, particularly during diagnostic or surgical procedures due to ischemia of the already compromised hypertrophied myocardium with or without arrhythmias.[9] Combination of coronary arterial abnormalities and biventricular outflow tract obstruction carry the highest risk.[9] Decreased coronary arterial blood flow may result from abnormalities of

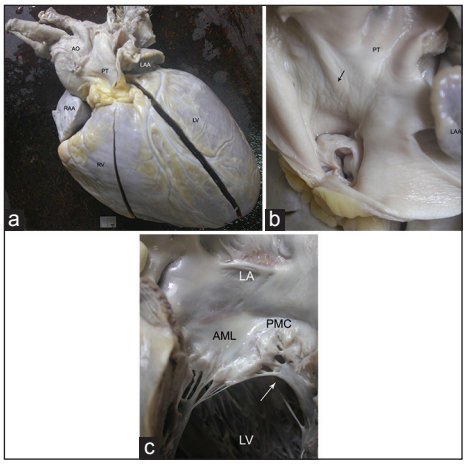

Figure 1: (a) Biventricular hypertrophy leading to moderate cardiomegaly. (b) The three semilunar cusps of the pulmonary valve are obscured by a nearly circumferential membrane. It has grown over the cusps leading to severe stenosis. There is poststenotic dilatation of the pulmonary trunk (PT) with intimal irregularity (arrow); jet lesion of pulmonary stenosis. (c) Dysplastic mitral valve with chordal abnormality (arrow) towards the posteromedial commissure PMC. Ao = Ascending aorta, AML = Anterior mitral leaflet, LA = Left atrium, LAA = Left atrial appendage, LV = Left ventricle, RAA = Right atrial appendage, RV = Right ventricle

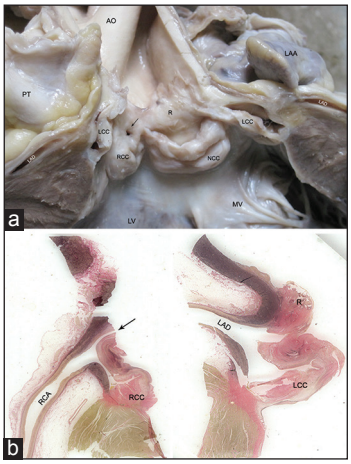

Figure 2: (a) Firm, grey white ridge at the sinotubular junction has produced severe stenosis. All the semilunar cusps are dysplastic. Of interest is the fusion of the coronary cusps to the ridge producing near-complete and complete isolation of right and left sinuses of Valsalva, respectively. This is well brought out in the (b) histological preparations (scanned slide of sections stained by elastic Van Gieson)

the arteries or their ostia. Exposure of the arteries to prestenotic high pressure leads to arterial dilatation and tortuosity, development of premature atherosclerosis, and in the long run, even aneurysms.[9] The coronary ostia may also be narrowed due to thickening of the sinus or aortic walls, especially when they are located close to the sinotubular junction.[5] Furthermore, the coronary ostia can get secondarily obstructed due to adhesion of the coronary cusps to the supravalvular ridge; the left coronary cusp seems to be affected more frequently than the right, and even total isolation of the sinus can occur.[5,9] In our patient, there was total and near-total, isolation of the left and right sinuses of Valsalva, respectively; a phenomenon seldom reported. To the best of our knowledge, this is the first histopathological documentation. All the cusps were thick and leathery with marked collagenization, which is probably due to suboptimal coaptation.[9] Often, peripheral and occasionally central pulmonary arterial stenosis occurs in conjunction with SVAS; valvular stenosis is rarely reported.[5] The heart in our patient revealed a supravalvular membrane that grew over the two anterior cusps, which has not been described so far. Mitral valvular abnormalities, as seen in this case, have also been described with SVAS, though their relation with elastin deficiency is still nebulous.[5,9] Our patient had all risk factors for SCD, that is, severe degree of stenosis, coronary ostial isolation, severe pulmonary valvular stenosis, and biventricular hypertrophy. A critical perfusion mismatch presumably resulted in subendocardial ischemia. This case highlights the need for heightened clinical suspicion of rare but treatable anomalies for prevention of SCD. All manifestations of this disorder, including the polyvalvular involvement and isolation of coronary ostia should be carefully assessed for optimal surgical management and prevention of SCD.

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