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Aortopathy Is Prevalent in Relatives of Bicuspid Aortic Valve Patients

Biner, S[1]; Rafique, A[2]; Siegel, R[2]; Tolstrup, K[2]
[1]Tel Aviv Sourasky medical Center, Tel Aviv, Israel; [2]Cedars Sinai Medical Center, Los Angeles, USA

Objective :
This study aimed to determine the prevalence of dilation and abnormal elastic properties of aortic root in firstdegree relatives (FDRs) of bicuspid aortic valve (BAV) patients.
Background:
Evidence indicates that BAV is a genetic disorder. Although FDRs of affected individuals have increased prevalence of BAV, their risk of aortic root abnormalities is unknown. Methods We studied dimensions as well as the elastic properties of the ascending aorta in 48 FDRs with morphologically normal tricuspid aortic valves, 54 BAV patients, and 45 control subjects using 2-dimensional echocardiography.
Results:
The prevalence of aortic root dilation was 32% in FDRs and 53% in BAV patients, whereas all control subjects showed normal aortic dimensions ($p < 0.001$). The FDRs and BAVs had significantly lower aortic distensibility (1.7 ± 1.4X10-3 mm Hg and 1.4± 2.0 X10-3 mm Hg vs. 2.5 ± 1.6 X10-3 mm Hg, $p < 0.001$) and greater aortic stiffness
index (26.7 ± 25.8 and 55.92 ± 76.8 vs. 18.7 ± 40.1, $p < 0.001$) compared with control subjects. This difference remained significant in subjects without aortic root dilation or hypertension ($p = 0.002$ and $p = 0.004$, respectively).
Conclusions:
The aortic root is functionally abnormal and dilation is common (32%) in first-degree relatives of patients with BAV. Screening of FDRs by transthoracic 2-dimensional echocardiography should be considered for detection of aortic valve malformation and dilated ascending aorta.