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The Evaluation of the Frequency and Echocardiographic Features of Cardiovascular Anomalies Among the Families of the Patients with Bicuspid Aortic Valve or Other Left-sided Cardiovascular Anomalies

Demir F., Karadeniz C., Atalay S., Tekin M., Tutar E.
Ankara University, Medical School, Department of Pediatric Cardiology and Pediatric Genetics, Ankara, Turkey

Objective: To evaluate the frequency of the cardiovascular anomalies among the families of the patients with bicuspid aortic valve (BAV) or other left-sided cardiovascular anomalies (LSCAs) and the echocardiographic features of these anomalies.

Methods: The patients, evaluated with BAV or any LSCA (aortic stenosis without BAV, coarctation of the aorta (CoA), interrupted aortic arch, hypoplastic left heart syndrome, and isolated dilatation of ascending aorta) in our clinic between October 2010 and August 2011 and their first degree relatives were enrolled into the study. The patients with any known genetic abnormality were excluded. All the participants underwent an echocardiographic examination. The relatives of every individual with newly diagnosed cardiovascular anomaly were also sequentially included. The patients were assessed in three groups: BAV, BAV+CoA, and other LSCAs. The relatives were grouped and evaluated according to the diagnosis of the proband in their families. The echocardiographic measurements of the patients and their siblings were standardized by computing Z-scores.

Results: The numbers of the patients in BAV, BAV+CoA, and other LSCAs group were 52, 14, and 20 respectively. Any LSCA was determined in 19 of the 263 relatives (7.2%) of the patients with BAV or LSCAs. Fourteen of those (5.3%) had aortic dilatation and five (1.9%) had BAV. A second individual with a left-sided cardiovascular anomaly was observed in 11 of the 86 (12.8%) families investigated. Most of the 66 patients with BAV (81.8%) were male and 14 out of these 66 patients (21.2%) had CoA as an associated anomaly. The frequencies of aortic stenosis (AS), aortic regurgitation (AR), AS+AR, and aortic dilatation in the patients with BAV were found 37.9%, 53%, 25.8% and 48.5% respectively. The aortic measurements of the patients with BAV were larger than the healthy siblings' values. The valvular dysfunction and aortic dilatation were more commonly observed in the patients who had right coronary and noncoronary leaflet fusion.

Conclusion: Almost all the complications of BAV and other LSCAs are preventable. Because the clustering of LSCAs in some families is observed, we recommend echocardiographic screening of those relatives. If not possible, at least it should be achieved for BAV.