The Frequency of Cardiovascular Anomalies among the Families of the Patients with Bicuspid Aortic Valve or Other Left-sided Cardiovascular Anomalies

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Introduction:
Bicuspid aortic valve is the most frequent congenital heart disease (0.5 to 1%). It was observed that BAV and other LSCAs (valvular aortic disease apart from BAV (VAD), coarctation of the aorta (CoA), interrupted aortic arch, and hypoplastic left heart syndrome) could coexist in the same patient and also clustering of BAV in some families. We aimed to evaluate the frequency of the cardiovascular anomalies among the families of the patients with BAV or other LSCAs and the features of those cases. This was the first study from Turkey that performed echocardiographic screening of the relatives of the patients with LSCAs.

Methods:
The patients, evaluated with BAV or any LSCA in our clinic between October 2010 and August 2011 and their first degree relatives were enrolled into the study. The patients with any genetic abnormality were excluded. All the participants underwent an echocardiographic examination. The patients were assessed in three groups: BAV, BAV+CoA, and other LSCAs. 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. The males were more frequently affected from LSCAs (male/female:4.4/1)(Table 1). Seventeen of 261 relatives (6.5%) were diagnosed with LSCA. BAV incidence among relatives of the patients with BAV was found 2.4% (Table 2). It was observed that “aortic root ± ascending aorta dilatation” was associated with R-N morphology and valvular dysfunction (Table 4). The most of the patients with BAV (59.1%) had R-L valvular morphology (Table 5).

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. This was the first study from Turkey that performed echocardiographic screening of the relatives of the patients with LSCAs.