Bicuspid Aortic Valve Prevalence In A Large Series Of Echocardiograms In The Area Of Frequent Consanguineous Marriage

Sık Akraba Evliliği Yapılan Bir Bölgede Büyük Bir Ekokardiyografi Serisinde Bikuspit Aortik Kapak Prevalansı

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Özet

Biküspit aort kapak (BAK) kalbin en sık konjenital anomalilerinden olmakla beraber prevalansı tam olarak belli değildir. Akraba evliliğinin yaygın olduğu (Güneydoğu Anadolu Bölgesi) bir bölgeden tek merkeze ait, büyük bir ekokardiyografik veri tabanından, BAK'ın prevalansını tespit etmeyi amaçladık ve BAK ile ilişkili aort stenozu, yetersizliği ve çıkan aorta dilatasyonu gibi komlikasyonların sıklığını sunduk. Üniversite hastanemizde 2004 ve 2007 yılları arasında çeşitli endikasyonlar için yapılan ve ekokardiyografik veri tabanında kayıtlı bulunan 31.265 raporu retrospektif olarak inceledik. BAK sıklıkla aort stenozu, yetersizliği ve çıkan aortanın genişlemesi ile birliktedir. BAK infektif endokardit için bağımsız bir risk faktörüdür. BAK'un prevelansı hesaplandı ve BAK olanlarda aort darlığı, aort yetersizliği ve çıkan aortanın genişleme sıklığı araştırıldı. Veri tabanımız cinsiyet dağılımını 11.939 erkek (%47) 12.926 kadın(%53) olarak göstermiştir BAK prevalansı %0.57'ydi (n: 180). Tüm Türkiye de akraba evliliği oranı %20.9 dur. Bölgemizde akraba evliliği oranı %40.4. Cinsiyete özgü BAK prevalansı erkeklerde %1.08, kadınlarda %0.44'tü. BAK'lıların % 58.3'ünde aort yetersizliği (%28.9 hafif, %29.4 ileri), %40'ında aort darlığı (%17.8 hafif, %22.2 şiddetli) ve %33.3'ünde çıkan aortta genişleme gözlemlendi. Yüksek akraba evliliği oranına rağmen çalışmamızda BAK prevelansı daha önce yapılan ekokardiyografi ve otopsi çalışmaları ile benzer ve tahmin edilenden az bulundu. Veri tabanımıza göre aort yetersizliği en sık eşlik eden komplikasyondu.

Anahtar kelimeler: Ekokardiografi, Biküspit Aort Kapak

Abstract

Bicuspid aortic valve (BAV) is one of the most common congenital anomalies of the heart, though its definitive prevalence is not clear. We sought to determine the prevalence of BAV in a large database of echocardiographic measurements from a single institution in the area of frequent consanguineous marriage (South-east Anatolia region) and we present the frequencies of BAV-related complications such as stenosis, regurgitation, and assendan aorta dilatation. We retrospectively analyzed 31.265 echocardiograms performed at our academic institution between 2004 and 2007 for various indications. BAV is often associated with aortic stenosis, regurgitation, and ascending aorta dilatation and BAV is an independent risk factor for infective endocarditis. The prevalence of BAV was calculated, and the frequency of aortic stenosis, regurgitation, and ascending aorta dilatation among BAV cases was determined. Our database revealed a gender distribution of 11.339 males (47%) and 12.926 (53%) females.The total prevalence of BAV was 0.57% (n = 180). The rate of consanguineous marriage is 20.9% in overeall Turkey. The rate of consanguineous marriage is 40.4% in our region The gender-specific prevalence was 1.08% in males and 0.44% in females. Regurgitation was observed in 58.3% (28.9% mild, 29.4% severe) of BAV cases, stenosis in 40% (17.8% mild, 22.2% severe), and dilatation in 33.3%. The prevalence determined in our study is similar to the prevalence determined in other recent echocardiography and autopsy studies, suggesting a lower incidence of BAV than previously thought despite high rate of consanguineous marriage. Regurgitation was the most common valvular complication in our database.

Key words: Echocardiography, Bicuspid Aortic Valve

Introduction

Bicuspid aortic valve (BAV) is one of the most common congenital heart defects, characterized by presence of 2 leaflets instead of the regular 3 leaflet valve structure (1,2). The leaflets of a bicuspid aortic valve are usually of unequal size with a raphe or false commissure apparent in the large of the leaflets. BAV is often associated with aortic stenosis (3), regurgitation (4), and ascending aorta dilation over time (5). In addition, BAV is an independent risk factor for infective endocarditis (6). There is controversy concerning the prevalence of BAV due to its rare presence and the difficulty of its diagnosis. BAV is thought to occur in 1% to 2% of the population (1), though this figure is mostly based on earlier autopsy studies. Development of noninvasive techniques such as echocardiography has allowed physicians to diagnose this condition in vivo. Here, we report the prevalence of BAV in a large series of patients referred for echocardiography for various indications at a single institution in the area of high rate of consanguineous marriage. In addition, we present the frequencies of BAV-related complications such as stenosis, regurgitation, and dilatation.

Methods

We retrospectively analyzed a large database of 31.265 echocardiograms performed at our institution between 2004 and 2007 for various indications. All patient were living in South-East Anatolia region. The rate of consanguineous marriages in this specific region is established according to the data provided by the Turkish Statistical Institute. The echocardiograms were interpreted by different cardiologists at our institution. A Vivid 3 ultrasound system was used, with BAV defined by transthoracic echocardiography findings of 2 clear leaflets on the short axis parasternal view, with or without a raphe.

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The severity of aortic stenosis was graded on the basis of transvalvular gradients; the degree of aortic regurgitation was defined by means of standard color Doppler criteria as absent, mild, or severe. Measurements of the aorta were performed in 2-dimensional parasternal (or transesophageal) long axis views.

All measurements were taken at hemodynamically stable conditions; results were averaged from 3 subsequent measurements in sinus rhythm, according to the leading-edge method.

Results

Our database revealed a gender distribution of 11,339 males (47%) and 12,926 (53%) females. BAV was identified in 180 patients, resulting in a total prevalence of 0.57%. The rate of consanguineous marriages is established according to the data provided by the Turkish Statistical Institute, the rate of consanguineous marriage is 20.9% in overeall Turkey but the rate is as high as 40.4% in our region. Among the BAV patients, the male-female ratio was approximately 2.5:1, with 123 males and 57 females, and a gender-specific prevalence of 1.08% for males and 0.44% for females. Functional alterations of the aortic valve were assessed in BAV patients. Aortic regurgitation was identified in a total of 105 patients (58.3%), in mild (n = 52, 28.9%) or severe (n = 53, 29.4%) form. Severe stenosis was present in 40 (22.2%), while mild stenosis was present in 32 (17.8%) patients. Aortic dilatation was determined in 60 (33.3%) patients.

Discussion

In this study we report a BAV prevalence of 0.57% from the largest database of echocardiograms published to date. Interestingly, despite high rate of consanguineous marriage (40.4%). BAV prevalance was not affected significantly in this region compared with other reports.

Earlier assessments of BAV prevalence had relied on autopsy results, which were effectively summarized by Basso et al. (7) Although, some of these studies revealed a prevalence of 0.5% to 0.6% (8-11). BAV was generally accepted to be present in 1% to 2% of the population, mainly based on 2 studies. One of these included a consecutive series of 21,417 autopsies revealing a 1.37% prevalence of BAV (12), while the other determined a prevalence of 0.9% but argued that it can be as high as 2%, if patients with cardiac disease were included (13). These studies may be biased by male preponderance since BAV appears to occur more often in males than females.

The development of echocardiography allowed for in vivo assessment of BAV and there have been a number of studies on children and adults. Some of these studies were investigating congenital heart defects in children and revealed a very low prevalence of BAV, on the order of 0.1% (14-16).

Since BAV is often asymptomatic in early years, and these studies on children included prescreen for symptoms of cardiac defect, it is very likely that the majority of BAV cases were missed in these studies. A more accurate assessment of BAV in the young necessitates screening of all children, without regard to symptoms.

Echocardiographic assessment of 817 apparently healthy 10-year-old children yielded a prevalence of 0.5% (7). Similarly, 2 children were detected to have BAV in a group of 357 adolescents (0.56%) (17). In an echocardiographic screen of 1075 consecutive live births, BAV was detected in 0.46% of the neonates (18).

It would not be feasible to perform echocardiography on a very large database such as ours without clinical symptomatology indicating further examination. However, we believe we have overcome this bias by including patients of all ages, since BAV becomes symptomatic in the course of adult life. Indeed, our calculation of 0.57% is in line with the results from screens of healthy children and neonates. Finally, in a very similar study setting, Movahed et al. have analyzed a database of 23.957 echocardiograms taken for various reasons and a screen of 1742 teenage athletes; they reported prevalence rates of 0.6% and 0.5%, respectively (19).

Aortic regurgitation was the most common valvular dysfunction detected in our study. Close to 60% of the patients had either the mild or severe forms of regurgitation. Other studies investigating the outcome of BAV and age have also found a large proportion of adult patients with regurgitation (20-28). Alegret at al. indicated that patients younger than 50 years of age were more likely to have regurgitation, while those older than 50 years of age mostly had a combination of regurgitation and age-related stenosis (20).

Aortic dilatation is a common finding in patients with BAV. In necropsy and surgical series, dilatation or aneurysm of the proximal thoracic aorta was found in 10-35% of patients (23,24).

Results of the present study was in accordance with the previous ones revealing a frequency of aorta dilatation (33%) among patients with BAV. As stated in a review by Cecconi et al. BAV and aorta dilation are often found together, suggesting a common underlying vascular defect (25).

The high likelihood for genetic inheritance of BAV was rigorously assessed by Cripe and colleagues utilizing variance component methodology, to estimate the role of genetic factors in complex human diseases (26). However, our findings do not support Cripe and colleagues.

It has been accepted that there was the important relation between congenital heart disease and some environmental factors such as viruses, drugs and genetic factors. These findings suggest that congenital heart disease may be polygenic or multifactoriel (27).

According to the data provided by the Turkish Statistical Institute, the rate of consanguineous marriage is 20.9% in overeall Turkey but the rate is as high as 40.4% in our region.

Despite the high rate of consanguineous marriages, 0.5% prevelance of BAV in our study was similar with the data from previous studies. Thus our results provide interesting data that the prevelance of BAV was not high despite high rate of consanguineous marriage.

In conclusion, other recent studies on adults, children, and neonates and our current study suggest a 0.5% to 0.6% prevalence of bicuspid aortic valve. This figure is much lower than the previously estimated 1% to 2% prevalence despite high rate of consanguineous marriages, nevertheless, it continues to account for a considerable portion of all congenital heart defects. BAV should not be regarded as a benign condition, since several complications arise in adulthood, the most common being aortic regurgitation in our patient population.

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