International Journal of Cardiology xxx (2017) xxx–xxx

Contents lists available at ScienceDirect

International Journal of Cardiology

journal homepage: www.elsevier.com/locate/ijcard

Short communication

Yield of family screening in patients with isolated bicuspid aortic valve in a general hospital

Luc Cozijnsen [a,*,1], Hester J. van der Zaag-Loonen [b,1], Richard L. Braam [a,1], Mirjam Bakker-de Boo [a,1], Jan G. Post [c,1], Berto J. Bouma [d,1], Barbara J.M. Mulder [d,1]

[a] Department of Cardiology, Gelre Hospital, Apeldoorn, Netherlands
[b] Department of Clinical Epidemiology, Gelre Hospital, Apeldoorn, Netherlands
[c] Department of Genetics, University Medical Centre, Utrecht, Netherlands
[d] Department of Cardiology, Academic Medical Centre, Amsterdam, Netherlands

ARTICLE INFO

Article history:
Received 27 June 2017
Received in revised form 9 November 2017
Accepted 13 December 2017
Available online xxxx

Keywords:
Bicuspid aortic valve
Family screening
Congenital heart disease

ABSTRACT

Aim: To determine the prevalence of unidentified bicuspid aortic valve (BAV) or aortic dilatation (>40 mm) in first degree relatives (FDR) of patients with isolated BAV in a general hospital.
Methods and results: Patients with isolated BAV received information advising cardiac screening of their FDR. Referred and screened were 134 FDR of 54 adult index patients with isolated BAV (median 2 per index patient). FDR's mean age was 49 years (range 16–83 years) and 41% were male. They comprised 5 parents (3.7%), 52 siblings (39%) and 77 offspring (57%). Among these FDR, the prevalence of BAV was 6.0% (8 patients). In FDR without BAV, 10 (7.5%) had aortic dilatation. 'Familial BAV' was present in 9/54 families (17%).
Conclusion: In a general hospital, screening of FDR of patients with isolated BAV resulted in a substantial yield of 13% new cases with BAV or aortic dilatation without BAV.

© 2017 Elsevier B.V. All rights reserved.

1. Introduction

Bicuspid aortic valve (BAV), the most common congenital heart defect (CHD), warrants timely diagnosis. It may lead to valvular dysfunction requiring surgery in up to 50% and is associated with progressive ascending aorta dilatation in over 40% [1–3]. However, patients are mainly asymptomatic until either significant hemodynamic changes or aortic dissection occurs. Although dissection risk is low (3 cases per 10,000 patient-years), it is still 8 times higher than in the general population [3], and it sometimes is the first manifestation of BAV. Timely diagnosis allows preventive surgery and supportive measures [4].

Familial clustering of BAV has been demonstrated, which allows family screening to serve as a diagnostic aid in the early detection of BAV [5–8]. Guidelines in 2014 stated that screening of first degree relatives (FDR) 'may be considered', especially if BAV is associated with either aortic dilatation [9,10] or a family history of valvular heart disease [10]. These recommendations were based on the so-far-reported prevalence of 8–10% of BAV in FDR of patients with BAV [5–8], an occurrence of 'familial BAV' (defined as families with >1 affected member) of 32–37% [5,6] and a reported prevalence of 32% of aortic dilatation in FDR without BAV [8]. However, these studies all suffered from potential sources of bias as they came from tertiary centres and were contaminated with BAV patients with other CHDs. Therefore, it is unknown whether this 2014 recommendation of FDR screening of BAV should be followed in general hospitals.

The aim of our study was to determine the prevalence of unidentified BAV in FDR of patients with isolated BAV in a general hospital. Additionally, we aimed to determine the prevalence of aortic dilatation in FDR without BAV and the total yield of newly diagnosed cases with BAV or aortic dilatation. Finally, we aimed to assess the occurrence of 'familial BAV', including both newly diagnosed and already known BAV.

2. Methods

Our study was conducted in a general non-academic teaching hospital without onsite cardiothoracic surgery and started in 2012. Patients with BAV, including those with isolated BAV who attended the outpatient clinic, received a letter that cardiac screening was advised for their FDR. Our electronic medical record system contained 285 patients with isolated BAV [11].

BAV-morphology (index patients or FDR) was determined by surgical inspection [11] or echocardiography, according to the orientation of the commissures [12]. In case of poor image quality, MRI was performed additionally. Aortic dimensions were measured end-diastolic in the parasternal long axis view, leading edge to leading edge [13]. Sinus of Valsalva and tubular ascending aorta were defined as dilated if the diameter exceeded 40 mm [9].

* Corresponding author at: Gelre Hospital, Albert Schweitzerlaan 31, 7334 DZ Apeldoorn, Netherlands.
E-mail address: l.cozijnsen@gelre.nl (L. Cozijnsen).
[1] This author takes responsibility for all aspects of the reliability and freedom from bias of the data and their discussed interpretation.

https://doi.org/10.1016/j.ijcard.2017.12.046
0167-5273/© 2017 Elsevier B.V. All rights reserved.

Please cite this article as: L. Cozijnsen, et al., Yield of family screening in patients with isolated bicuspid aortic valve in a general hospital, Int J Cardiol (2017), https://doi.org/10.1016/j.ijcard.2017.12.046

The study was judged by the hospital scientific committee as an observational study not within the scope of Dutch law of medical-scientific research with humans (WMO).

3. Results

Referred were 134 FDR of 54/285 (19%) patients with isolated BAV (Fig. 1A). Mean age of the index patients was 62 years (standard deviation [SD] ± 13, range 27–81 years), 33 (61%) were male and 36 (67%) had undergone aortic valve replacement (AVR), including concomitant ascending aorta replacement in 12 (22%). Among 18 patients who had not been operated, the mean diameter of sinus of Valsalva and tubular ascending aorta was 38 mm (SD ± 6.9 mm) and 39 mm (SD ± 8.7 mm), respectively; 11 patients (20%) had an aortic dilatation. Altogether, 43% of the index patients had an aortic dilatation (present or repaired).

We screened 134 FDR (median 2 per index patient) who comprised 5 parents (3.7%), 52 siblings (39%) and 77 offspring (57%). Mean age was 49 years (SD ± 15, range 16–83 years) and 55 (41%) were male. The diagnostic imaging modality was echocardiography in 129 subjects (96%) and MRI in 5.

BAV was diagnosed in 8 FDR (6.0%, 95% confidence interval [CI] 2.6–11.4%) (Fig. 1A). Their mean age was 40 years (SD ± 16), five were male (63%) and 3 also had aortic dilatation. The 8 newly diagnosed BAV were relatives of 4 index patients (Table 1). Medical treatment for aortic dilation was initiated in 2 (beta-blocker in 1, losartan in 1) and additional family screening was performed in 1 (cascade screening).

In the screened FDR with TAV, 10 had an aortic dilatation (7.5%; 95%CI 3.6–13%), 8 at level of the sinus of Valsalva and 2 at level of the tubular ascending aorta (Fig. 1A); measurement of the tubular ascending aorta was not possible in 14 patients (11%). Mean age of these 10 FDR was 58 years (SD ± 12), 6 were male, 4 had hypertension and 1 had a parent with an aortic dilatation (49 mm). Medical treatment for aortic dilatation was initiated in 4 of them (beta-blocker in 1, losartan in 3). Overall, 18 new cases (13%) of BAV or aortic dilatation with TAV were diagnosed.

In the 54 index patients, we could trace 22 FDR with already known aortic valve phenotype of whom 6 had a BAV (diagnosed at AVR) (Fig. 1B). They were related to 6 index patients. In one family (D in Table 1), one BAV was newly diagnosed and one already known. 'Familial BAV' (including the already known and the newly diagnosed) was present in 9 out of the 54 families (17%, 95%CI:7.9–19%).

4. Discussion

Our study is the first to investigate the yield of cardiac screening in FDR of patients with isolated BAV in a general hospital. In this population, we found newly diagnosed BAV in 6.0%, aortic dilatation without BAV in 7.5%, summing up to 13% new cases with BAV or aortic dilatation.

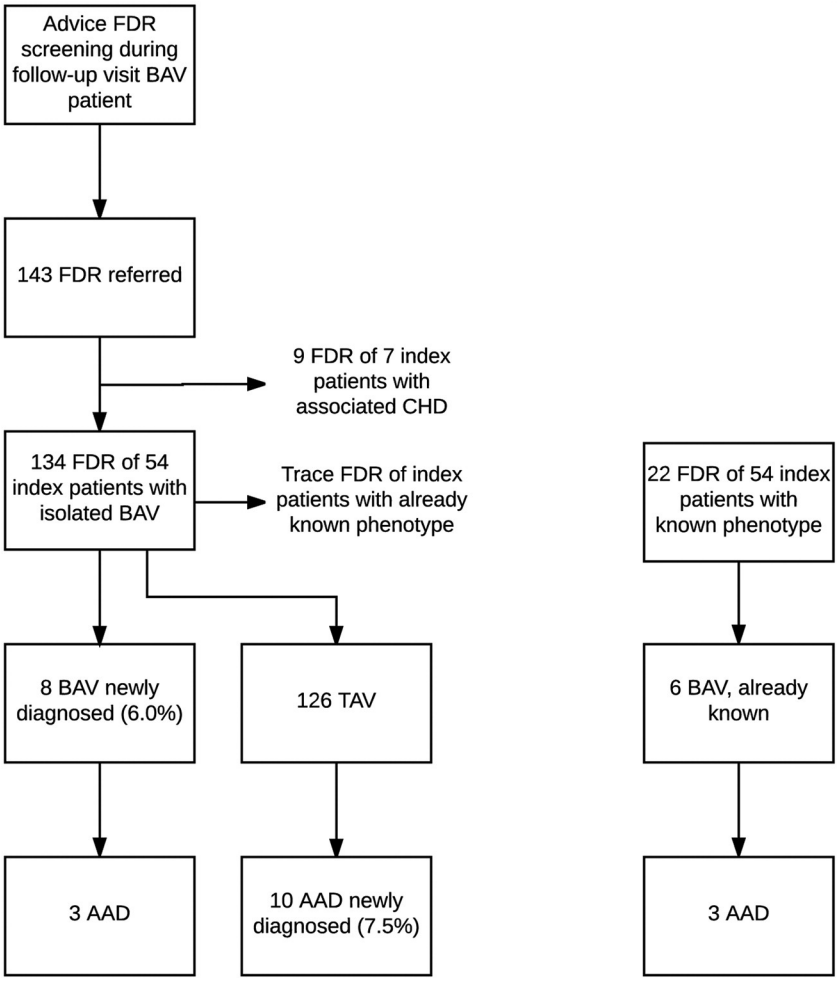

Fig. 1. Flowchart of patient selection and results. Fig. 1A Screening of unidentified BAV and AAD; Fig. 1B Tracing of already known BAV. AAD, ascending aorta dilatation; BAV, bicuspid aortic valve; FDR, first degree relatives; TAV, tricuspid aortic valve.

Please cite this article as: L. Cozijnsen, et al., Yield of family screening in patients with isolated bicuspid aortic valve in a general hospital, Int J Cardiol (2017), https://doi.org/10.1016/j.ijcard.2017.12.046

Download English Version:

https://daneshyari.com/en/article/8662440

Download Persian Version:

https://daneshyari.com/article/8662440

Daneshyari.com