Scientific Research Publishing

World Journal of Cardiovascular Diseases, 2023, 13, 283-288
https://www.scirp.org/journal/wjcd
ISSN Online: 2164-5337
ISSN Print: 2164-5329

Unicuspid Aortic Valve, an Extremely Rare Congenital Anomaly in Adults: A Systemic Review

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How to cite this paper: Kumanayaka, D.D., Otto, A., Chauhan, M., Belinschi, V., Mirza, N., Rayad, M.N. and Suleiman, A. (2023) Unicuspid Aortic Valve, an Extremely Rare Congenital Anomaly in Adults: A Systemic Review. *World Journal of Cardiovascular Diseases*, **13**, 283-288.
https://doi.org/10.4236/wjcd.2023.136025

Received: May 26, 2023
Accepted: June 27, 2023
Published: June 30, 2023

Abstract

The most common congenital heart defect is the bicuspid aortic valve (BAV) occurring with fusion of one of the three-valve commissures. A rarer valvular phenomenon is the unicuspid aortic valve (UAV) which has an estimated prevalence of 0.02% in the adult population. Two morphologic subtypes of UAV exist: the earlier presenting, acommissural UAV and the later appearing unicommissural. To better characterize and manage patients with UAVs, a systematic review of this rare phenomenon was performed. The objective of our study was to conduct a systematic review of adequate case studies involving UAV patients to describe patient characteristics, clinical presentation, management, and common sequela featured there within. The mean age of presentation in this review was 35.6 years. When diagnosing UAV, TEE was the most utilized diagnostic modality (65%) followed by intraoperative diagnosis (17%) discovered with valve repair or incidentally during another procedure. TTE was utilized to diagnose only 7% of the cases reviewed likely owing to the difficulty and inaccuracies in identifying UAV with this method that were previously established by multiple series. Interventional options for UAV are balloon versus surgical valvotomy, aortic valve replacement, Ross procedure and aortic root replacement. They are performed on an individual basis and all cases ultimately require aortic valve replacement (AVR) or Ross procedure (to avoid anticoagulation). Aortic root replacement is additionally required if aortic root/ascending aorta diameter exceeds 4.5 cm. In this review, Aortic aneurysm (16%) cases resulted in surgical interventions. There may be a need to implant permanent pacemaker (in 3% - 8%) during AVR when calcified UAV has calcification extending into interventricular septum.

Keywords

Unicuspid Aortic Valve, Bicuspid Aortic Valve, Heart Defects, Unicommisural Valve

1. Introduction

With the increased utilization of echocardiogram for diagnosis, the prevalence of congenital heart disease is reported as high as 50.3 out of 1000 live births [1]. The most common congenital heart disease defect is the bicuspid aortic valve which occurs with the fusion of one of the three-valve commissures [2] [3]. Rarer valvular phenomena include the unicuspid aortic valve and the quadricuspid aortic valve which have an estimated prevalence of 0.02% and 0.005% in the adult population respectively [4] [5]. For unicuspid aortic valves (UAV), two morphologic subtypes exist: the earlier presenting, acommissural UAV and the later appearing unicommissural UAV [6] [7] [8]. UAVs are typically seen in association with aortic valve stenosis, regurgitation, and aortic root dilation [8] [9] [10]. Current literature for understanding UAV exists primarily in case studies and sporadic surgical reports and is therefore limited in a holistic sense.

Due to the earlier need for aortic valve replacement and the risk of sudden cardiac death in UAV patients compared to bicuspid aortic valves (BAV), it is imperative to have an accurate diagnosis of UAV and its type. To better characterize and manage patients with UAVs, a systematic review of this rare phenomenon was performed. The objective of our study was to conduct a systematic review of adequate case studies involving UAV patients to describe patient characteristics, clinical presentation, management, and common sequela featured there within.

2. Materials and Methods

In this review, we included case studies and series describing adult patients with UAV that had basic demographic, diagnostic, clinical presentation and outcome information. Articles were excluded if patients were less than 18 years old, were non-english or did not meet previously mentioned inclusion. PubMed® database (https://pubmed.ncbi.nlm.nih.gov) was searched from 1994 to 2020 for the following key phrases: unicuspid aortic valve, echocardiography, aortic stenosis. Two reviewers screened studies for inclusion/exclusion and a third reviewer was utilized in the event of a disagreement. 119 articles were screened and 85 case reports or case series were included for data extraction. Studies were screened at the abstract and study title level. Full texts for articles deemed eligible at this level were retrieved, and references describing the same study were matched and duplicates removed. Data on patient age, sex, and method of UAV diagnosis, presenting symptoms, complications of unicuspid AV, management, and prognosis was extracted by the review team. The most common presenting symptoms, radiographic or intraoperative findings, and method of diagnosis were presented

as percentages of patients yielded from our search. The patient age of diagnosis was presented as an average. The complaints of syncope, presyncope and dizziness were tallied together due to likelihood of similar etiology and challenge of eliciting differences in this historical information from patients based on article review. Not all studies reported published their imaging findings and verification of UAV based on imaging or gross pathology was not possible in all cases.

3. Results

A total of 92 patients (males n = 76, female n = 16) with UAV were identified from 85 case reports accessed through PubMed®. Mean patient age was 35.6 years. Of the presenting symptoms dyspnea was the most common (41%) followed by chest pain (17%), syncope (including dizziness 11%), palpitations (7%), and asymptomatic (7%). Most often, UAV presented with aortic stenosis (49%), aortic regurgitation (36%), aortic aneurysm (16%), left ventricular hypertrophy (10%). Transesophageal echocardiogram was the most utilized diagnostic modality (65%), followed by intraoperative finding (17%), 3-dimensional echocardiogram (7%), and computerized tomography scan (5%). Treatment of UAV was predominately operative with 2% of patients managed with medical therapy. The outcome of the management resulted in either complete resolution of patient symptoms or significant improvement (96%). Causes of death post treatment included cardiogenic shock (n = 1), Covid-19 complications (n = 1), and sudden cardiac death (n = 1).

4. Discussion

Unicuspid aortic valve is an extremely rare congenital anomaly that was first reported by Edwards in 1958 [11]. The annual incidence of UAV has been estimated at 0.02% in the adult population and at a rate of 4% to 5% for patients undergoing aortic stenosis repair. A normal trileaflet aortic valve consists of 3 cusps with 3 associated commissures, which develop from embryonic tubercles of the aortic trunk. Unicuspid aortic valve is thought to occur due to failure of the 3 aortic cusps to separate before birth [12].

The earlier appearing subtype of UAV is acommissural and described as having a pinhole shape. The age of presentation is usually in children and young adults and previous studies suggest this type is associated with more aggressive ascending aorta dilatation [6] [13]. Later appearing and less symptomatic is the unicommissural UAV which is slit-shaped. Like a previous review of UAV cases by Mookadam *et al.*, limited studies reviewed here distinguished unicommissural and acommisural morphologies making it difficult to understand true differences in their prevalence and presentation [13]. This group urges future providers to emphasize the specific morphology observed in their patients.

The mean age of presentation in this review was 35.6 years. This is lower than previous reviews and cohorts that consisted of mean ages of 42 years and 51 years [12] [13]. These differences may relate to varying compositions of UAV

morphologies in studies or a potential trend of earlier detection because of more sophisticated imaging modalities. The higher male prevalence (76 to 16, M:F) of UAV in this review mirrors the male bias that previous groups have identified for BAV. However, UAV seems to have an even larger male prevalence than BAV which has been describe as between 3:1 and 4:1 [2] [14]. Additionally, male patient prevalence of UAV observed in this review was slightly higher than that observed in previous bodies of work for UAV that observed a 4:1 male to female ratio [13].

The most common presenting symptoms of UAV were dyspnea (41%), chest pain (17%), and syncope (11%). These nonspecific symptoms align with the most common UAV sequela observed: aortic stenosis (49%) and aortic regurgitation (36%). A similar study observed nearly universal aortic stenosis in UAV patients [12]. The lower association between UAV and aortic stenosis observed in our review may be due to the differences in diagnosis of UAV which varied from diagnostic imaging to direct visual examination. When diagnosing UAV, Transesophageal echocardiogram (TEE) was the most utilized diagnostic modality (65%) followed by intraoperative diagnosis (17%) discovered with valve repair or incidentally during another procedure. Transthoracic echocardiogram was utilized to diagnose only 7% of the cases reviewed likely owing to the difficulty and inaccuracies in identifying UAV with this method that were previously established by multiple series [8] [15].

Interventional options for UAV are balloon versus surgical valvotomy, aortic valve replacement, Ross procedure and aortic root replacement. They are performed on an individual basis and all cases ultimately require aortic valve repair or Ross procedure (to avoid anticoagulation). Aortic root replacement is additionally required if aortic root/ascending aorta diameter exceeds 4.5 cm. In this review, aortic aneurysm (16%) cases resulted in surgical interventions. There may be a need to implant permanent pacemaker (in 3% - 8%) during aortic valve

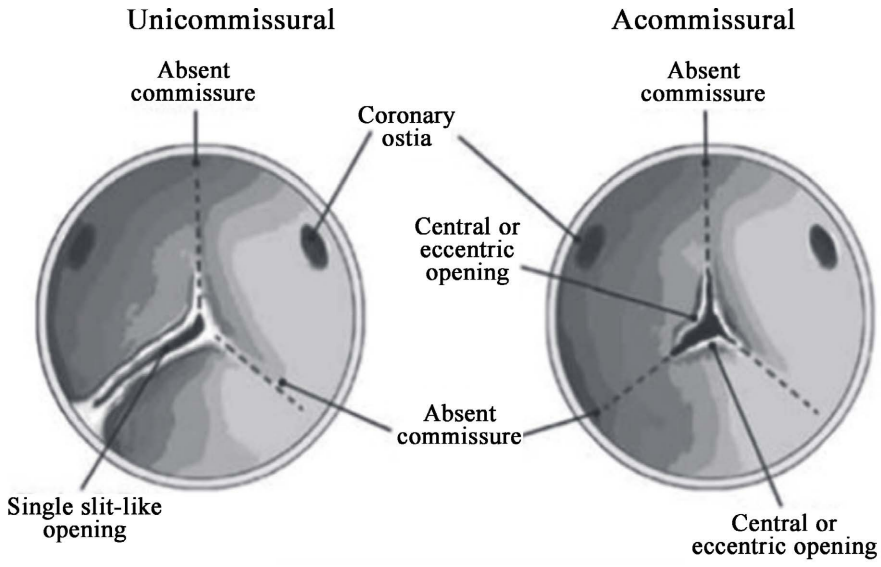

Figure 1. Unicommisural vs acommisural morphologies of aortic valve [16].

repair when calcified UAV has calcification extending into interventricular septum [16].

Limitations of this literature included only one database being included, although more than 100 articles were screened. The reviewers also noted significant heterogeneity between articles and possible publication bias as limitations.

Due to the similarities in patient presentation and characteristics between UAVs and BAVs, previous reports have viewed UAV as an extreme in the spectrum of BAV syndromes rather than thinking of UAV as its own pathology. We suggest that UAV can be considered as its own separate entity to highlight the importance of recognizing it as early as possible to prevent complications and reduce associated mortality/morbidity. Further studies are required to investigate the phenotype and pathology of UAV as a separate entity, with the understanding that aortic stenosis and aortic complications tend to occur earlier in those patients with UAV compared to BAV population (**Figure 1**).

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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