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Authors: Małgorzata Niemiec, Bartosz Gruchlik, Jacek Zarzecki, Romuald Wojnicz, Katarzyna Mizia-Stec

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Unicuspid aortic valve: More data and more doubts in the light of six years follow-up observation

Short tittle: Unusual presentation of congenital aortic valve defect

Małgorzata Niemiec[1], Bartosz Gruchlik[1], Jacek Zarzecki[1], Romuald Wojnicz[2], Katarzyna Mizia-Stec[1]

[1]1st Department of Cardiology, Medical University of Silesia, Katowice, Poland

[2]Department of Histology and Cell Pathology, Medical University of Silesia with the Division of Dentistry in Zabrze, Medical University of Silesia, Katowice, Poland

Correspondence to:

Prof. Katarzyna Mizia-Stec, MD, PhD,

1st Department of Cardiology, Medical University of Silesia,

Ziołowa 45/47, 40–635 Katowice, Poland,

phone: +48 32 359 88 90,

e-mail: kmiziastec@gmail.com

A unicuspid aortic valve is a rare congenital heart defect with an incidence is 0.02% [1, 2]. Optimal time for cardiosurgical treatment in young adults with congenital aortic valve disease may be a matter of controversy [3, 4]. In this population left ventricle (LV) remodeling is an ongoing process since organogenesis [5], and its degree may not match the severity of the defect.

We report a case of a 26-year old male patient with unicuspid aortic valve and consecutive diagnostic dilemmas in the interpretation of discrepancy between LVH and degree of unicuspid valve pathology during 6-year follow-up.

The unicuspid aortic valve was functionally incompetent- in 2016 both aortic regurgitation (AR) and stenosis (AS) were observed in transthoracic (TTE) and transesophageal echocardiography (TEE) (AR jet, 9 mm; PHT, 360 ms; V_{max}, 3.8m/s; P_{mean}, 38 mm Hg; P_{max}, 57 mm Hg; AVA, 2.1 cm^2; bulb, 37 mm; AoAsc, 35 mm; AoDesc, 20 mm with normal flow and no signs of coarctation). Moreover, a significant concentric LVH was found (interventricular septum [IVS] up to 16 mm, posterior wall 18 mm) with normal systolic and diastolic diameters and a preserved LV ejection fraction (LVEF, 67%) (Figure 1A–C). Additional clinical findings involved: a negative family history of hypertrophic cardiomyopathy (HCM); normal blood pressure (120/80 mm Hg) and normal kidney function (GFR above 90 ml/min/1.73 m^2). At discharge further observation was indicated.

During the next hospitalization (2020), the patient did not present any limitations in physical activity (New York Heart Association [NYHA] class I, N-terminal pro-B-type natriuretic peptide [NT-proBNP], 117 pg/ml) and complained about pain and paresthesia in the lower extremities. TTE / TEE showed mild progression of the aortic valve disease (AR jet, 10 mm; PHT, 310 ms; V_{max}, 4.2m/s; P_{mean}, 46 mm Hg; P_{max}, 67 mm Hg; AVA, 1.36 cm^2; bulb, 40 mm; AoAcs, 45 mm) and more advanced LVH (IVS, 21 mm; posterior wall, 19 mm) with LVEF of 65%. LV global longitudinal strain (GLS) was 15% with a typical pattern for amyloidosis. An cardiac magnetic resonance (CMR) confirmed the LVH (Figure 1D) and LV hyperkinesis and multifocal intramuscular regions of late gadolinium enhancement. Taking into regard the progression of LVH, symptoms and TTE results other potent etiology of LVH were verified — both endomyocardial biopsy (hypertrophy and mild degree atypical reactive inflammation —Figure 1E) and biochemical/genetic tests were negative in regards to Anderson-Fabry disease, amyloidosis or HCM. The patient was discharged with the recommendation of clinical and TTE control once a year.

In 2022 the still asymptomatic patient presented increased NT-proBNP level (370 pg/ml), as well as more advanced signs of LV and aortic remodeling. Echocardiography revealed: LVH up to 20 mm, normal LV diameters, LVEF, 60%; LV GLS, 8.7% (Figure 1F) and the presence of an ascending aortic dilatation (bulb 40mm, AoAsc 49mm). The patient was qualified by Heart Team for surgical aortic valve replacement and ascending aortic surgery.

To conclude, the presented case shows that unicuspid aortic valve may provide to the complex form of valve structural and functional incompetence. The advanced LV remodeling may pose some diagnostic problems. Moreover, a young patient’s age, atypical symptoms, potent concomitant diseases make the decision about further treatment more complicated.

Article information

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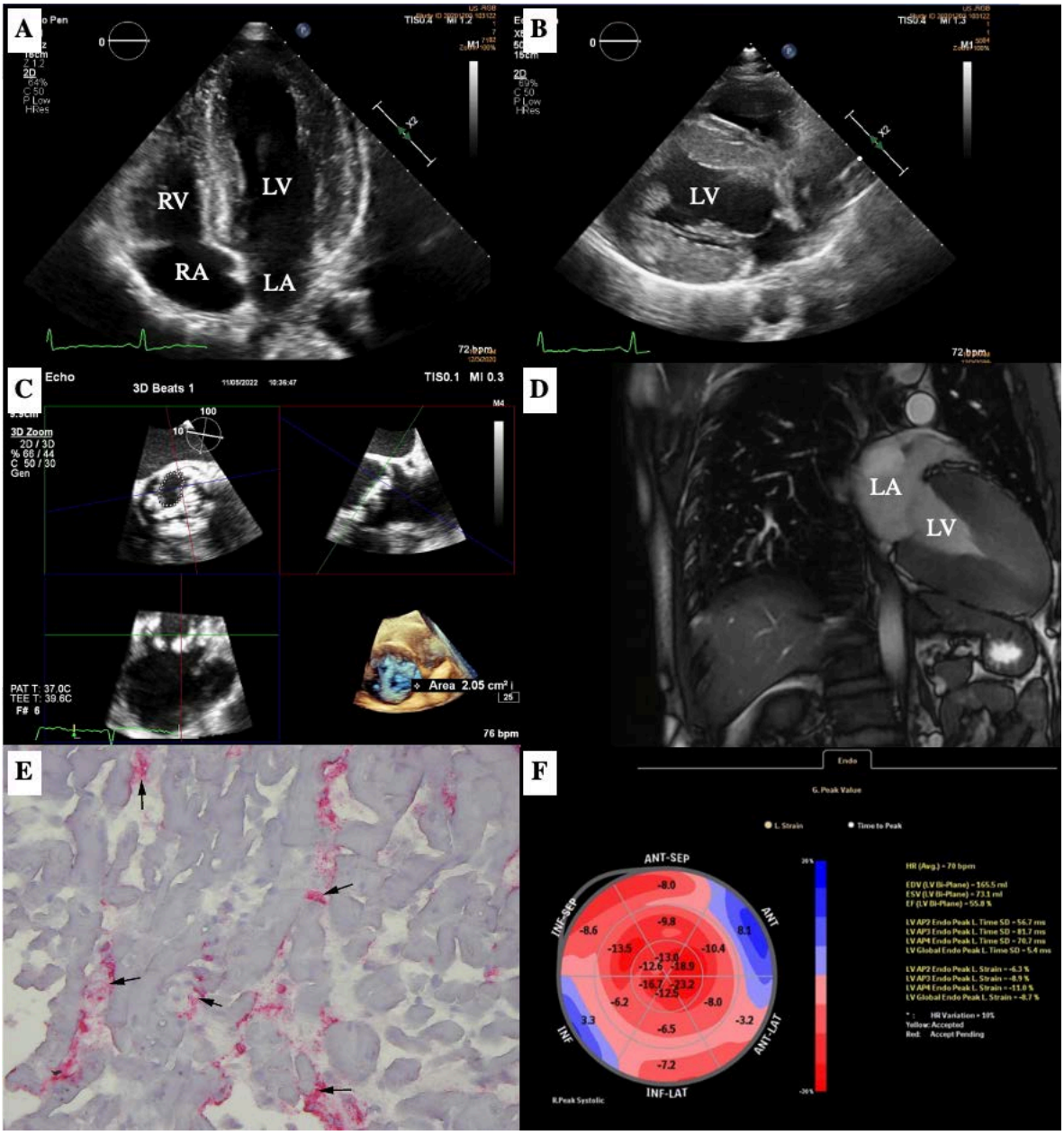

Figure 1. A. TTE, four-chamber view. **B.** TTE, parasternal long axis view. **C.** TEE, 3D acquisition and multislice assessment of aortic valve area. **D.** CMR with contrast. **E.** Endomyocardial biopsy demonstrated patchy distributed of CD68(+) macrophages (red color) with concomitant myocyte injury (the arrows) suggesting reactive inflammation; **F.** LV GLS, 8.7%

Abbreviations: CMR, cardiac magnetic resonance; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; TEE, transesophageal echocardiography; TTE, transthoracic echocardiography