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

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An unicuspid aortic valve is a rare congenital heart defect with an incidence is 0.02% [1, 2]. The 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 from organogenesis onwards [5], and its degree may not match the severity of the defect.

We report a case of a 26-year-old male patient with a unicuspid aortic valve and consecutive diagnostic dilemmas in the interpretation of a discrepancy between left ventricular hypertrophy (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 on transthoracic (TTE) and transesophageal echocardiography (TEE) (AR jet, 9 mm; PHT, 360 ms; V_{max}, 3.8 m/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 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 reported 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. Cardiac magnetic resonance (CMR) confirmed LVH (Figure 1D) and LV hyperkinesis and multifocal intramuscular regions of late gadolinium enhancement. Given the progression of LVH symptoms and TTE results, other potent etiology etiologies of LVH were verified — both endomyocardial biopsy (hypertrophy and mild degree atypical reactive inflammation —Figure 1E) and biochemical/genetic tests were negative with regard 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 an increased NT-proBNP level (370 pg/ml), and more advanced signs of LV and aortic remodeling. Echocardiography showed 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 40 mm, AoAsc 49 mm). The patient was qualified by Heart Team for surgical aortic valve replacement and ascending aortic surgery.

To conclude, the presented case shows that a unicuspid aortic valve may provide a complex form of valve structural and functional incompetence. Advanced LV remode-

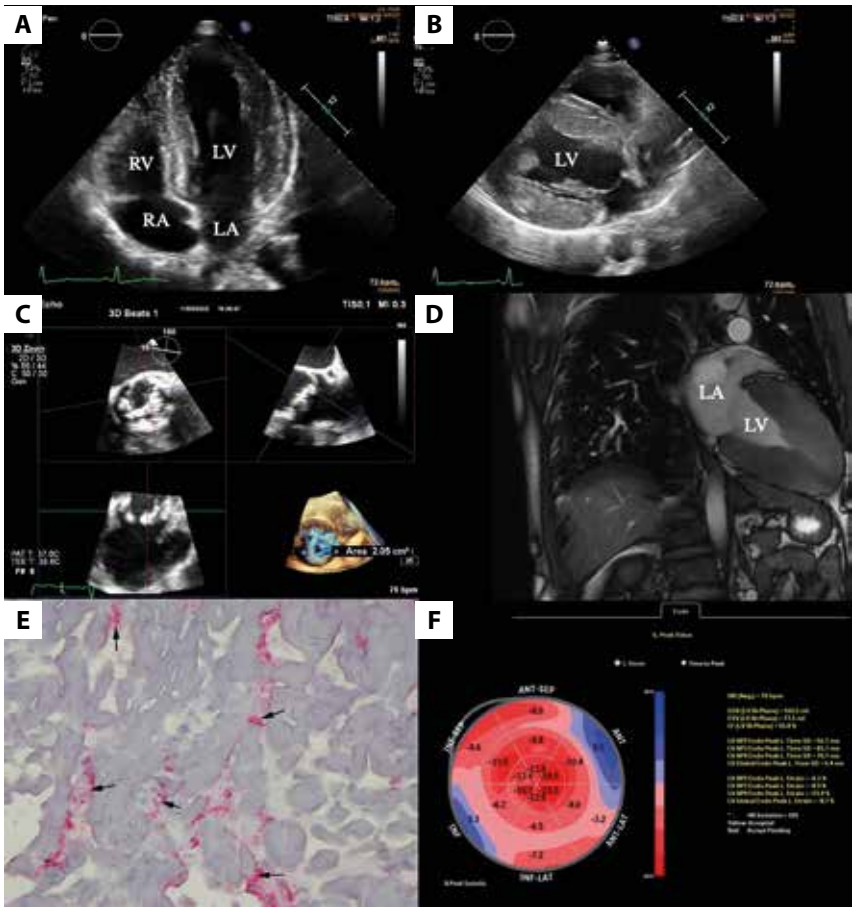

Figure 1. A. TTE, four-chamber view. **B.** TTE, parasternal long-axis view. **C.** TEE, 3D acquisition and multislice assessment of the aortic valve area. **D.** CMR with contrast. **E.** Endomyocardial biopsy demonstrated patchy distribution of CD68(+) macrophages (red color) with concomitant myocyte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

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

ling may pose some diagnostic problems. Moreover, the young age of the patient, atypical symptoms, and potent concomitant diseases make the decision about further treatment more complicated.

Article information

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