Severe bicuspid aortic valve regurgitation with ascending aortic aneurysm in a 53-year-old female patient with Turner syndrome

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Turner syndrome is a genetic disorder caused by missing or abnormal structure of the X chromosome. The incidence is estimated at 1:2500–1:3000 female births. The women, apart from typical phenotypic features and delayed mental development, present several cardiovascular disorders, which are coarctation, dilatation of the arch of the aorta, aortic aneurysms, and a bicuspid aortic valve. The average age at diagnosis is 9–10 years. The prognosis is usually good if regular monitoring and treatment of concomitant diseases are provided [1–3].

We present the case of a 53-year-old female patient admitted to the Cardiology Department for increasing shortness of breath and significant deterioration in exercise tolerance. The patient showed signs of mild intellectual disability. According to her documentation, the woman was diagnosed with Turner syndrome in her teenage years. For almost 40 years of her life, she remained without specialist monitoring and never had cardiological diagnostics for social reasons.

In the laboratory tests, elevated concentrations of N-terminal pro-B-type natriuretic peptide and troponin were noted. Sinus tachycardia with features of left ventricular hypertrophy and overload were noticed on the electrocardiogram. Transthoracic echocardiography showed good systolic function of the left ventricle and no signs of right ventricular overload or segmental contractility disorders. No pericardial effusion was also detected. However, we detected severe insufficiency (Figure 1A) of the bicuspid aortic valve (Sievers type 1a, left coronary cusp and right coronary cusp fusion) (Figure 1B) with an ascending aortic aneurysm measuring 57 mm and dilation of the main artery arch to 41 mm (Figure 1C). Additionally, below the origin of the left subclavian artery, aortic stenosis of 12 mm with a maximum gradient of 28 mm Hg was noticed (Figure 1D). These disorders had not been previously diagnosed. Computed tomography angiography excluded signs of dissection, estimated the dilation dimension at 63 × 60 mm (Figure 1E), and the narrowing of the main artery isthmus to 13 mm, with subsequent bending of the vessel at a right angle (Figure 1F). A horseshoe kidney was also found. Based on these findings, prognostic indicators were calculated: the aortic height index (AHI — 38 mm/m) and the aortic size index (ASI — 35.6 mm/m^2). The patient was classified as high-risk and had a consultation with the Heart Team, which resulted in qualification for a surgical procedure. The last element of the diagnostic process was a coronary angiography, in which no significant stenosis was found.

During her hospitalization in the Cardiac Surgery Center, the patient was implanted with a biological aortic valve and ascending aortic prosthesis. The stenosis of the aortic isthmus qualified for conservative treatment.

After the procedure, the patient was transferred to the Cardiac Rehabilitation Department. During her hospitalization, she remained in good condition. Control echocardiography showed normal function of the prosthesis and a decrease in the gradient at the site of the stenosis of the descending aorta, which was associated with a decrease

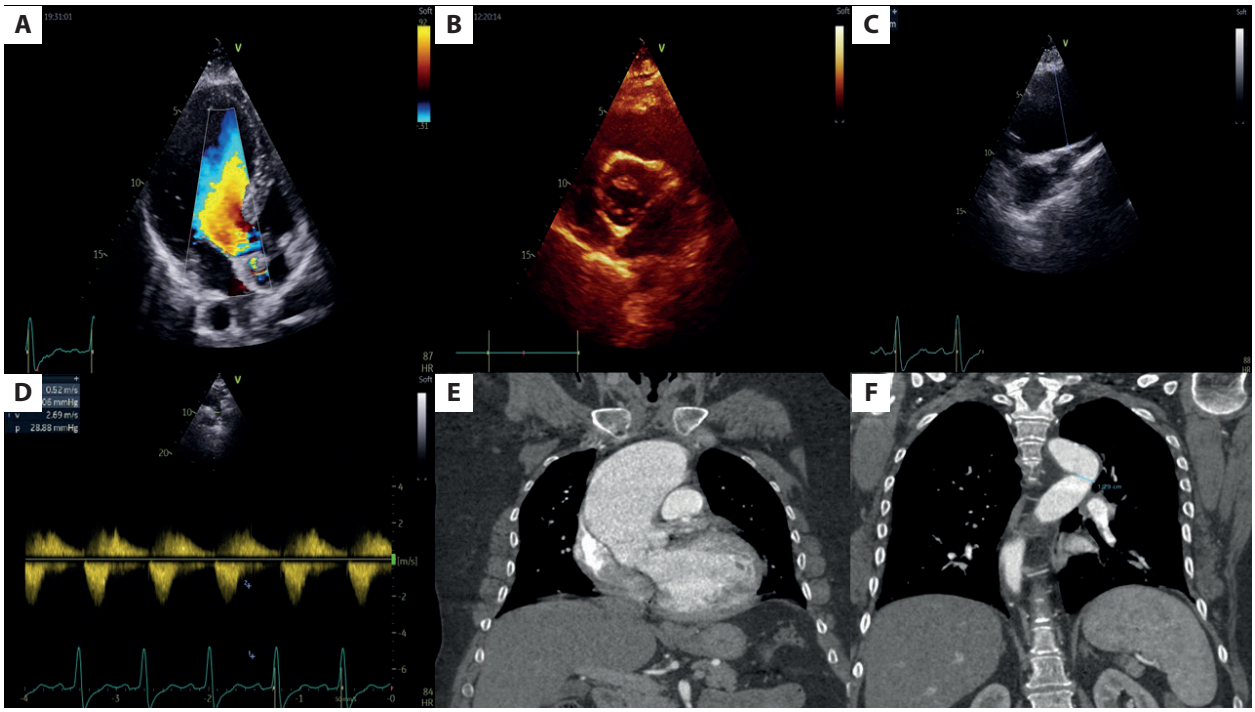

Figure 1. A. Severe aortic occlusion regurgitation. **B.** Bicuspid aortic valve. **C.** Ascending aortic aneurysm. **D.** Gradient at the site of narrowing of the descending aorta. **E.** Aortic aneurysm on computed tomography. **F.** Aortic isthmus stenosis on computed tomography

in cardiac output resulting from the correction of aortic regurgitation. A complication of the surgical treatment was pericarditis, which was treated with colchicine.

The presented case is a rare example of a congenital heart defect diagnosed at a relatively late age. It also illustrates the key role of echocardiography in the diagnosis and control of patients with Turner syndrome. These patients also require regular follow-up [4, 5].

Article information

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