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Double-chambered right ventricle in a 16-year-old patient with Williams syndrome

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Abstract

We present a case of double-chambered right ventricle diagnosed during preparation for colonoscopy due to gastrointestinal bleeding in a 16-year-old, mentally disabled boy with Williams syndrome. The patient was previously diagnosed with ventricular septal defect and mild pulmonary stenosis. Echocardiography performed under general anesthesia revealed hypertrophied muscular bundles in the right ventricle with the maximum gradient of 100 mmHg, causing severe outflow obstruction. This type of defect is extremely rare in patients with Williams syndrome, with only one case, which was diagnosed during invasive angiocardiography, described in world literature. A successful total surgical correction was performed based on echocardiography data.

Introduction

Williams syndrome (WS) is a genetic disorder caused by submicroscopic deletion in the q11.23 region of chromosome 7. It is characterized by dysmorphy ("elfin face"), mental disability and cardiovascular diseases caused by elastin deficiency. The following abnormalities are typical of WS: 1) arterial narrowings located in the supravalvular aortic segment, the pulmonary trunk and its branches, and, less commonly, along the aortic arch and isthmus; 2) pulmonary valve and aortic valve stenoses[1–7]; 3) structural and functional anomalies of the mitral valve; and 4) ventricular septal defects. Other congenial heart defects seem to be unrelated to elastin deficiency[1–7].

Williams syndrome is characterized by its diversity and by alteration of clinical manifestations, especially altered evolutions of arterial stenoses: aortic usually increase, while pulmonary often regress[2,4–7]. As a result, Williams syndrome is often diagnosed in different periods of patient's life, with the full-blown syndrome usually diagnosed during infancy, and partial presentation identified much later[2,4–7].

Cardiological diagnosis based on ECHO may be hindered by the tendency of clinicians to focus on typical anomalies, especially in uncooperative, mentally disabled children. Usually, it is possible to evaluate the proximal sections of main arteries, where most lesions are located; however, a detailed picture of the heart and peripheral sections of the vessels is difficult to obtain.

It should be emphasized that the dynamics of morphological changes in WS requires frequent re-evaluation. In order to illustrate these difficulties, we present a case of double-chambered right ventricle, which is untypical of Williams syndrome.

Case report

A 16-year-old boy with genetically confirmed WS was admitted to Gastroenterology Department due to body mass deficiency and gastrointestinal bleeding. Eventually, Crohn's disease and gastroesophageal reflux disease were diagnosed. Patient' medical history revealed ventricular septal defect that closed spontaneously and mild pulmonary valve stenosis. Only some of the medical records were available due to frequent changes of place of residence.

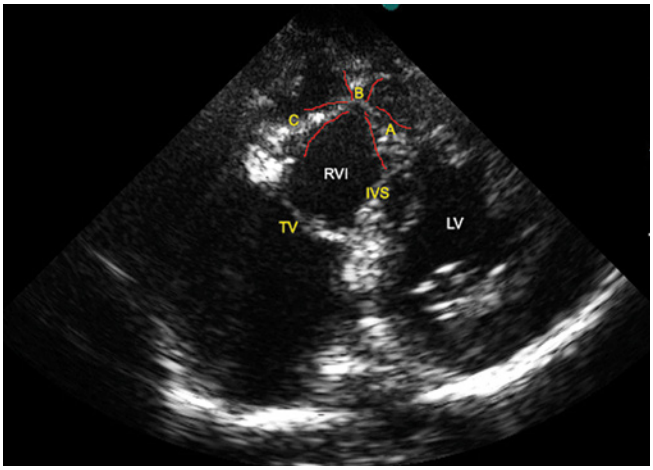

Fig. 1. *Modified parasternal cross-sectional view displaying the left ventricle at the level of the papillary muscles, the right ventricle inflow tract, and muscle structures inside the right ventricle, dividing it into a high-pressure inflow chamber (RVI) and a low-pressure outflow chamber. Three abnormal groups of interconnected muscles are displayed: A: moderator band (hypertrophic, displaced anterior and upward; B: hypertrophic muscle bundles arising from the anterior wall of the RV, connected with the base of the anterior papillary muscle; and C: an additional, thick muscle bundle extending from the tricuspid ring – from the bottom part of the anterior leaflet – to the moderator band, to which it connects above the base of the medial papillary muscle (Lancisi). The borders of abnormal muscles are highlighted with red lines. LV: Left Ventricle, IVS: Interventricular Septum, TV: Tricuspid Valve, RVI: low-pressure inlet of the Right Ventricle*

The patient showed typical phenotypic features of Williams syndrome and severe mental retardation. A loud (5/6), harsh systolic heart murmur with thrill and intensive pulsation of the precordium were detected. Echocardiography performed in an awake patient showed a turbulent flow within the right ventricle, with V_{max} of 4.2 m/s. The examination was repeated under general anesthesia due to lack of patient cooperation and revealed extensive fibromuscular intraventricular narrowing of the right ventricle, formed by three abnormal groups of muscles: 1) hypertrophic moderator band, displaced anteriorly and upward; 2) thick muscle bundles arising from the anterior wall of the RV, fused with the base of the anterior papillary muscle; and 3) anomalous, thick muscle bundle extending from the tricuspid ring – from the lowest part of the anterior leaflet – to the moderator band, above the base of the medial papillary muscle (Fig. 1). We found an opening of not more than 4 mm in the diameter between the inflow and the outflow chambers (Fig. 2 and 3) with a maximum gradient of 120 mmHg (Fig. 4). Tricuspid regurgitation grade III with V_{max} of 4.85 m/s was detected (Fig. 5). The pulmonary valve was normal. Additionally, mitral valve prolapse with mild regurgitation and mild aortic regurgitation were observed. The ascending aorta was hyperechogenic, with a mild localized stenosis (11 mm) 1 cm distal to the sinotubular junction (Fig. 6) (aortic sinus 19.7 mm, distal ascending aorta 14–15 mm; max. flow velocity 1.75 m/s). The interventricular septum showed deformation suggesting spontaneous closure of ventricular septal defect: displaced conal septum and the

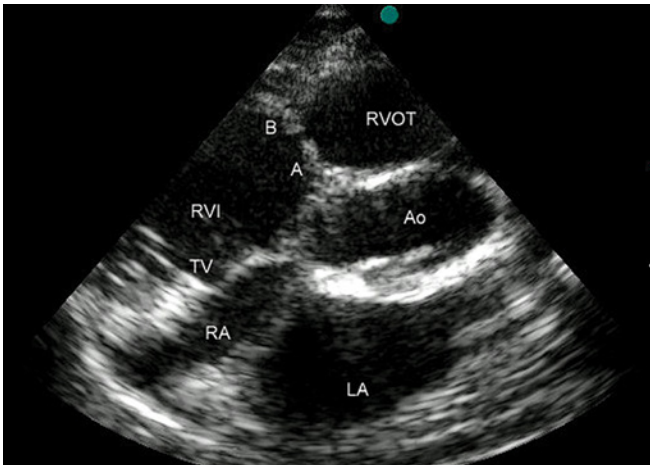

Fig. 2. *Modified high vascular short-axis view, oriented more sagittally than usual. A fibro muscular diaphragm formed by the above described structures, dividing the right ventricle into a high-pressure inflow chamber (RVI) and a low-pressure outflow chamber (RVOT), is visible. The flow through the diaphragm dividing the RV is visualized using color Doppler. Only a 3–4 mm communication between the two chambers of the right ventricle is visible. Ao – Aortic valve, LA Left Atrium, RA Right Atrium, TV – Tricuspid Vlave:* **A.** *moderator band;* **B.** *Hypertrophied band on the anterior wall of RV*

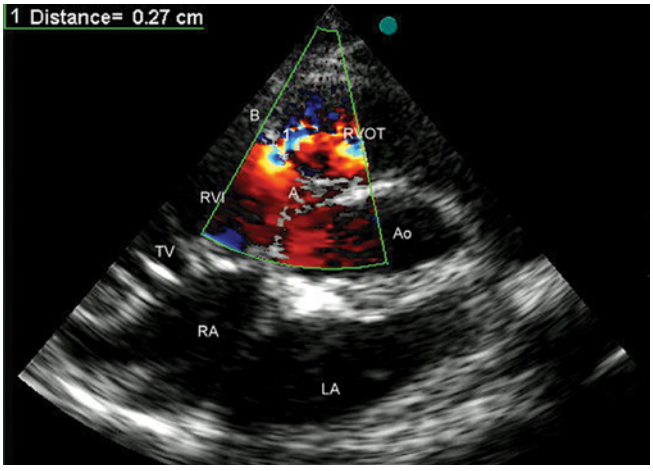

Fig. 3. *Visualizations with color Doppler of the flow through the diaphragm dividing the RV; An orifice of only 3–4mm diameter is visible. Designations as in Fig. 2*

main body of the septum connected with thin hyperechoic tissue (Fig. 7). Doppler examination showed no intraventricular shunt, however the final diagnosis was left to the discretion of a surgeon due to high pressure in the inflow portion of the RV.

The patient was diagnosed with double chambered right ventricle based solely on echocardiography and was qualified for open heart surgery.

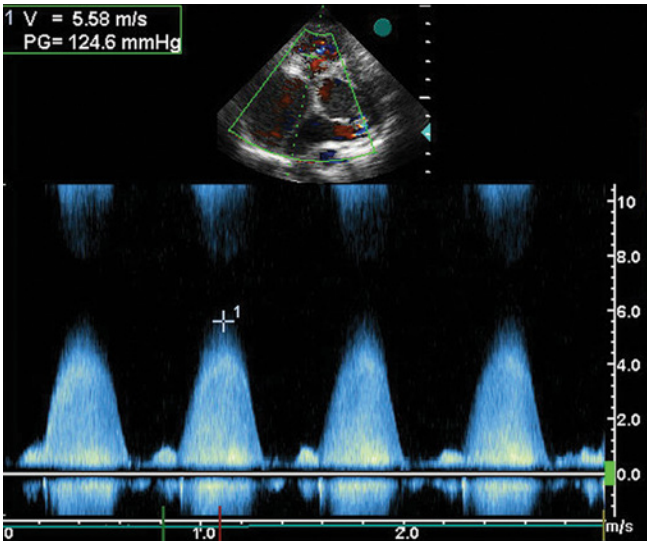

Fig. 4. *The measurement of the flow velocity through the RV narrowing. Calculated maximum pressure gradient of 124 mmHg*

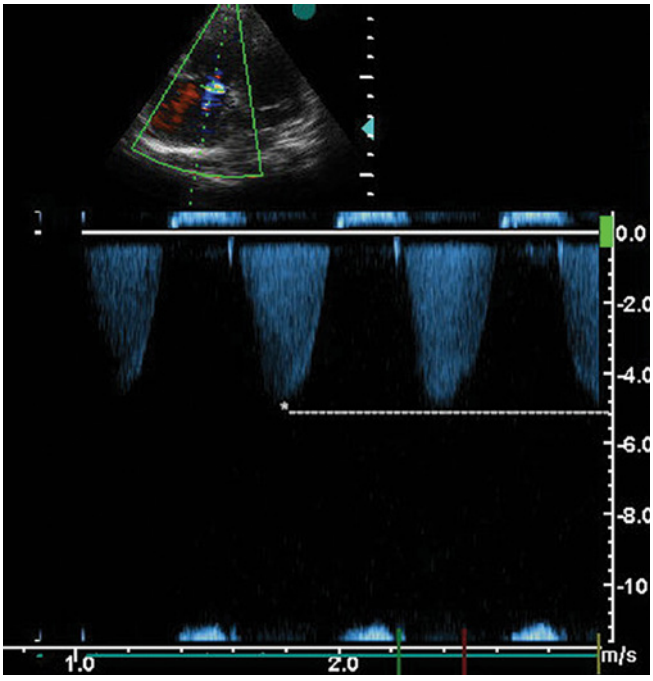

Fig. 5. *The measurement of the velocity of tricuspid regurgitation – calculated maximum gradient RV-RA of 120 mmHg*

The surgery was performed using the right atrial approach. No macroscopic abnormalities of the ascending aorta were found. Pronounced hypertrophied muscle bundles covered with fibrotic tissue narrowed the right ventricular outflow tract. We found that a fibromembranous structure arising from the site of the closed ventricular septal defect was an additional important component of the right ventricular outflow obstruction. The entire abnormal fibromuscular structure was resected and a wide outflow tract was restored. The postoperative period was uneventful.

Ten months after surgery, mild flow turbulence inside the right ventricle (Fig. 8), an increase in the flow velocity within RVOT up to 1.7 m/s (Fig. 9), and mild tricuspid valve regurgitation with velocity of 2.4 m/s were recorded. Mitral and aortic valve regurgitations remained unchanged.

Discussion

We found only one published case of double chambered right ventricle in a 25-year-old male patient with Williams syndrome, in whom the diagnosis was based on angiography due to loud murmur and increased RV flow revealed by echocardiography[8].

The term double chambered right ventricle refers to the narrowing inside the right ventricle resulting from hypertrophy and an abnormal position of the muscular structures dividing its cavity into a high-pressure inflow chamber and a low-pressure outflow chamber. This narrowing has a progressive nature. The relationship between ventricular septal defect (particularly in long-terms cases), including those closing spontaneously, with subvalvular/membranous narrowing of the left ventricle outflow tract or other defects causing intraventricular blood flow distur-

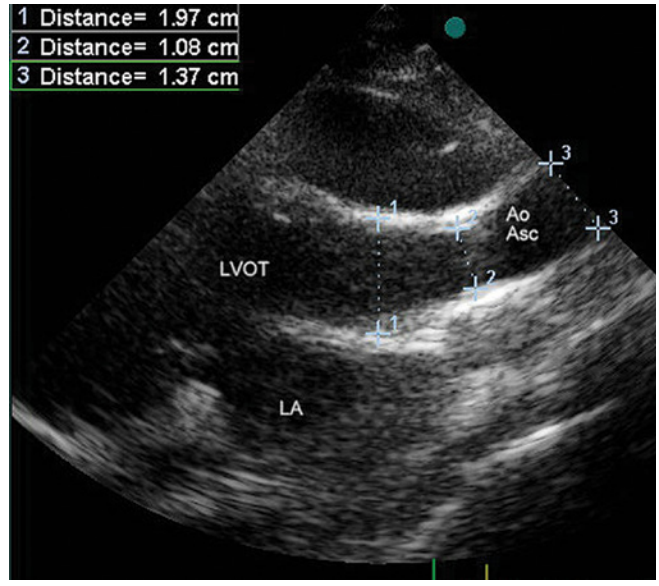

Fig. 6. *The ascending aorta in long-axis view. The walls are hyperechogenic, the proximal part of the ascending aorta seems slightly narrowed*

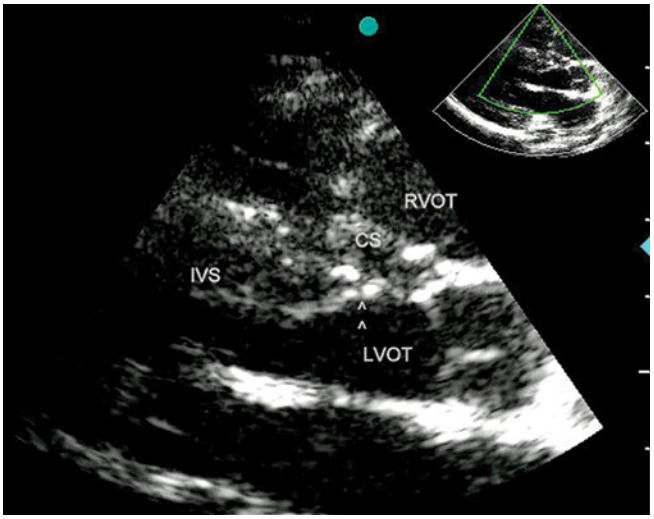

Fig. 7. *Left ventricular long axis: the main parts of the ventricular septum are displaced and connected with a thin band of tissue – a picture suggesting a spontaneous closure of the defect located in this area. IVS: the main body of ventricular septum. CS: conal septum, LVOT: left ventricular outflow tract, RVOT: right ventricular outflow tract, arrowheads point the location of potential VSD*

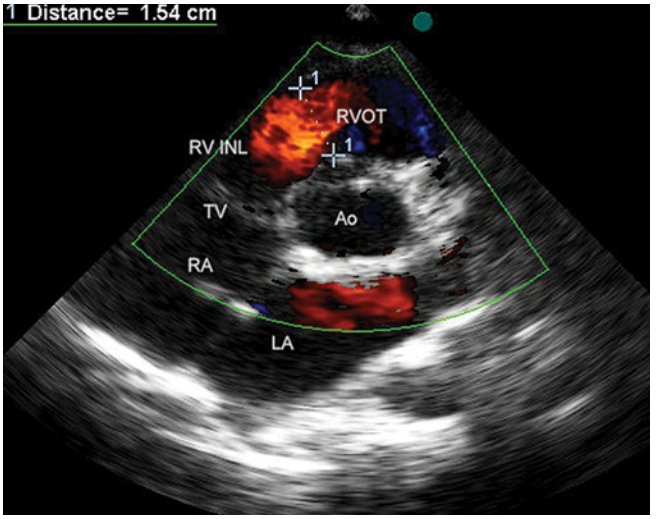

Fig. 8. *Color Doppler of the flow through the RVOT ten months after surgery. A view analogical to the one used in Fig. 2. Only mild turbulences of the flow are visible, no residual structures obstructing RVOT are present*

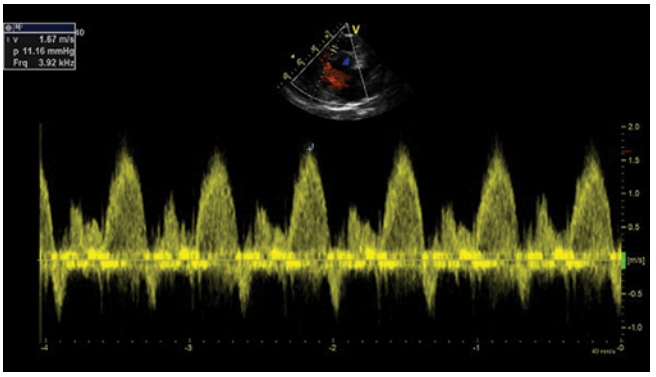

Fig. 9. *Velocity of the flow recorded simultaneously with spectral Doppler does not exceed 1.67 m/s – residual maximum gradient of 11 mmHg*

bances, such as Down syndrome, Noonan syndrome and twin-to-twin transfusion syndrome, is emphasized[9]. The incidence of ventricular septal defect in Williams syndrome is estimated at about 11%, which is much higher than the observed rates in the general population[6], suggesting that there is a relationship between these anomalies and confirming the presence of at least an indirect correlation between Williams syndrome and double chambered right ventricle. On the other hand, usually small, hemodynamically insignificant muscular defects are observed[1–3,6], and it is hard to relate their presence with elastin deficiency in Williams syndrome. The question remains whether detection of an increased number of muscular septal defects is associated with a more careful examination of children with Williams syndrome or is related to the syndrome itself[6]. In the presented case, the echocardiographic examination along with intraoperative inspection revealed anatomic factors causing intraventricular critical stenosis, as well as intervetricular septum malformations suggesting spontaneous defect closure. The literature review confirms that this is a typical natural history of double chambered right ventricle, which allows us to assume that the narrowing develops as a consequence of ventricular septal defect rather than Williams syndrome. Late diagnosis of both documented DCRV cases corresponds to the progressive character of the disease.

Conclusions

Patients with Williams syndrome should be screened for atypical heart malformations, especially when new clinical symptoms emerge that do not correspond to previous medical findings.

Conflict of interest

The authors do not report any financial or personal connections with other persons or organizations which might negatively affect the content of this publication and/or claim authorship rights to this publication.

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