Ventricular noncompaction and Williams syndrome are genetic disorders with typical clinical and echocardiographic cardiovascular manifestations. Here, we describe a young patient with rare association of clinical phenotype suggestive of Williams syndrome and right ventricular noncompaction.

Keywords: Echocardiography, noncompaction, Williams syndrome

INTRODUCTION

Ventricular noncompaction is a genetic cardiomyopathy with various mutations in genes associated with neuromuscular (82%) and congenital heart diseases (12%), namely bicuspid aortic valve, aortic coarctation, Ebstein’s anomaly, and Tetralogy of Fallot. Complete atrioventricular (AV) block has been observed in 22% of patients by He et al. Williams syndrome is a genomic disorder caused by a de novo microdeletion of chromosome 7q11.23. The deletion leads to haploinsufficiency of approximately 28 genes, including elastin which leads to the severe thickening, fibrosis of the vascular media layer in medium- and large-sized arteries. The clinical features of Williams syndrome may include short stature, characteristic dysmorphic elfin face, intellectual disability with an outgoing personality, musculoskeletal abnormalities, infantile hypercalcemia which resolves during childhood and cardiovascular malformations. The typical heart defects associated with Williams syndrome (50%–80%) are supravalvar aortic stenosis, pulmonary arterial stenosis, diffuse aortic hypoplasia, aortic coarctation, hypertension, congenital septal defects, and rarely aortic and mitral valve stenosis or regurgitation.

CASE REPORT

A 31-year-male presented with exertional shortness of breath class II for one year. His clinical examination revealed short stature with typical elfin face with no overt mental retardation. Examination revealed a blood pressure of 140/90 mmHg, pulse of 50 bpm, regular, jugular venous pressure elevated, mild pedal edema, pectus excavatum, kyphosis, aortic ejection systolic murmur, apical pan-systolic murmur, and a mid-diastolic murmur. In addition, he had upper limb wasting. Electrocardiography showed complete heart block. His echocardiogram showed hypoplastic ascending aorta and pulmonary arteries, severe calcification of mitral valve, aortic valve, aorta, basal ventricular and atrial septum, severe valvular mitral and aortic stenosis, moderate mitral regurgitation, mild aortic regurgitation, rupture of anterior tricuspid leaflet chordae with severe tricuspid regurgitation, with the right ventricular (RV) systolic pressure of 41 mmHg and good biventricular function. In addition, there was severe noncompaction of the right ventricle with multiple trabeculae and deep intertrabecular spaces in the entire free wall, apex as well as RV side of septum. The end-systolic ratio of the noncompacted to compacted myocardial layer was 3:1. There was color flow into the trabecular recesses. There was minimal circumferential pericardial effusion. Computed tomography scan showed diffuse hypoplasia of aorta with extensive calcifications involving thoracic aorta and its major branches.

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study done showed upper limb myopathy. His forced vital capacity/forced expiratory volume in 1 s ratio was <1. In view of his severe comorbid problems, he was advised medical follow-up by cardiac surgeons. He was offered palliative surgery, but he and family declined. Blood investigations showed hemoglobin 14 g/dl (n = 14–18.1), creatinine 85 µmol/L (n = 45–100), calcium corrected 2.54 mmol/L (normal range, 2.1–2.6), erythrocyte sedimentation rate 37 mm/h (normal range, 2–25), C-reactive protein 44.7 mg/L (normal <5), and creatinine phosphokinase was normal. Patient refused genetic testing. Family screening did not reveal any abnormalities. Later, the patient went to an outside country for surgery, but unfortunately expired postsurgery.

**DISCUSSION**

This patient had all typical phenotypes of Williams syndrome. In addition, his echocardiogram showed extensive dystrophic calcification localized to heart and aorta with normal corrected calcium levels indicating chronic fibrosis leading to calcification. There were severe mitral, aortic valvular lesions and the rare involvement of tricuspid valve disease which is not reported in Williams syndrome. Furthermore, he had evidence of RV noncompaction which is again not reported previously in patients with Williams syndrome. The complete AV block may be either due to severe septal calcification or as an association of ventricular noncompaction as noted previously. The upper limb myopathy may be an association of either syndromes. It is observed that the creatine kinase is generally elevated in the majority of patients with myopathy, but may be normal in slowly progressive myopathies. As genetic testing was not done, the clinical phenotype suggests Williams syndrome. This case report bring out an intriguing question whether there is a genetic linkage between Williams syndrome and ventricular noncompaction which needs further confirmatory studies.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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