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Brief Report

Isolated obstruction of the right ventricular infundibulum in a patient with Williams' syndrome

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Abstract Williams' syndrome is a rare condition that is associated with severe cardiovascular manifestations. We report a patient with Williams' syndrome who also has isolated obstruction of the right ventricular infundibulum, an association that, to the best of our knowledge, has not been previously reported.

Keywords: Heart defects; congenital; right ventricular outflow obstruction

Williams' syndrome is an autosomal dominant condition which occurs in 1 in every 10,000 live births. In its full-blown form, it includes cardiovascular anomalies, elfin face, mental deficiency, short stature, characteristic dental malformations, and infantile hypercalcaemia. The condition is a contiguous gene syndrome associated with a heterozygous microdeletion in the chromosomal region 7q11.23, an area which encompasses the elastin gene.[1] We present a patient with Williams' syndrome who has isolated right ventricular infundibular obstruction, an association, to the best of our knowledge, not previously reported.

Case report

Our male patient presented at 25 years of age after a murmur was noted on routine examination. He has classical phenotypical features of Williams' syndrome, and cardiovascular examination showed a harsh systolic murmur, graded at 2 out of 6, loudest at the upper left sternal edge with no radiation. Echocardiography was difficult due to the patient being overweight, but showed acceleration of flow in the right ventricular outflow tract, with a peak velocity of 3.9 metres per second. Acceleration appeared to commence from well below the pulmonary valve. Cardiac catheterisation showed isolated pulmonary infundibular obstruction, with a hypertrophied and heavily trabeculated right ventricle, and dilation of the pulmonary trunk and left and right pulmonary arteries (Fig. 1). The peripheral pulmonary arteries were normal. Pullback from pulmonary trunk to the right ventricle showed isolated infundibular obstruction, with a pulmonary arterial pressure of 12 over 4, and a mean of 8, millimetres of mercury, and a right ventricular pressure of 50 over 6 millimetres of mercury under general anaesthesia. Simultaneous aortic pressure was 90 over 45 millimetres of mercury, and there were no left-sided lesions, including supravalvar aortic stenosis and renal arterial stenoses.

Discussion

Mortality in Williams' syndrome is mostly due to cardiovascular complications. Cardiovascular manifestations tend to be worse in males,[2] and include subvalvar,[3] valvar and supravalvar aortic stenosis that may be associated with an aortic valve having only two leaflets,[4] peripheral pulmonary stenosis, discrete stenosis of the pulmonary arteries,[5] hypoplastic pulmonary arteries,[6] mitral valvar prolapse with or without severe regurgitation,[5] coarctation or hypoplasia of the aorta,[7] and ostial stenosis of the coronary arteries.[8] The two commonest cardiovascular lesions are peripheral pulmonary arterial stenosis, which tends to improve with time, and supravalvar aortic stenosis which tends to progress.[2]

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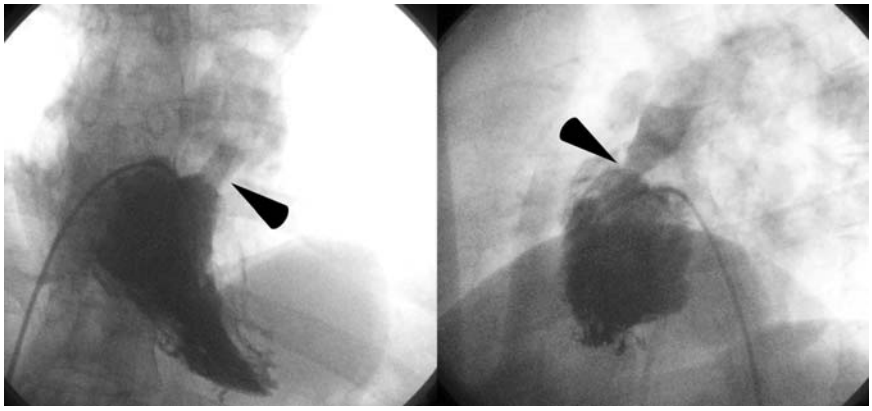

Figure 1.
Right ventricular angiogram. The left panel shows the postero-anterior view and the right panel shows the lateral view. The level of the stenoses is denoted by the arrows. Note the severe degree of narrowing of the right ventricular outflow tract especially on the lateral view, and the dilation of the pulmonary trunk.

Renal malformations, and renal arterial stenosis, are also associated with this condition,[7] and such involvement may or may not be associated with systemic hypertension, implying that hypertension may occur simply as a manifestation of generalized arteriopathy rather than due to renal hypoperfusion.[4] Stroke may occur independently of hypertension due to stenoses of the cerebral arteries.[9] Our case is unusual in that an extensive search of the literature failed to reveal any previously reported case of isolated right ventricular infundibular obstruction. Intervention for this young man is planned for the near future.

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