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Brief Report

Severe pulmonary hypertension secondary to a parachute-like mitral valve, with the left superior caval vein draining into the coronary sinus, in a girl with Turner's syndrome

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Abstract A 17-year-old girl with Turner's syndrome underwent two cardiac operations due to severe mitral stenosis with pulmonary hypertension, caused by a parachute-like mitral valve. The anomaly was associated with persistence of the left superior caval vein, which drained to the coronary sinus, and non-compaction of the left ventricular myocardium. The association of these lesions is rare in patients with Turner's syndrome.

Keywords: Mitral valve; XO syndrome; left superior vena cava

CARDIOVASCULAR DISORDERS ARE THE MAIN cause of increased mortality in patients with Turner's syndrome. They occur in one quarter to one third of the patients,[1,2] with the spectrum ranging from bicuspid aortic valve, coarctation of the aorta, partially anomalous pulmonary venous connection, hypertension and dilation of the ascending aorta, to hypoplasia of the left heart. The mitral valve is usually of normal morphology, although frequently with prolapse.[3] The combination of a parachute-like mitral valve with persistence of the left superior caval vein draining to the coronary sinus in girls with Turner's syndrome is rare.

Case report

A 17-year-old girl with Turner's syndrome, with the 45 XO karyotype, was evaluated due to increasing pulmonary hypertension. She had undergone a plastic repair of a parachute-like mitral valve at the age of 9 years. The preoperative pulmonary arterial pressure was 100/75 mmHg, with a maximum wedge pressure of 35, and a mean pressure of 22 mmHg. At surgery, there were two papillary muscles supporting the mitral valve, but the posteromedial one was much smaller than the other. The tendinous cords were short and fused to membrane-like structures, so that not only the valvar orifice, but also the secondary openings within the cords appeared to be stenotic. A left superior caval vein entered the enlarged coronary sinus. Several incisions were made parallel to the cords, creating secondary openings. The patient recovered completely, and her pulmonary arterial systolic pressure decreased to 20 mmHg. Two years later, however, there was recurrence of mitral stenosis and insufficiency, leading to a rise in the pulmonary arterial pressure. At this stage, neither the diastolic pressure gradient nor the area of the valve area suggested severe stenosis. Over time, however, the pulmonary hypertension worsened, and atrial fibrillation developed. Eight years after the operation, the patient developed dyspnoea and orthopnoea. A loud second sound was then audible over the pulmonary valve, along with a musical systolic murmur of grade 3 out of 6, and a soft diastolic murmur graded at 1 to 2 out of 6. There was no opening snap. The electrocardiogram showed atrial fibrillation, along with an rsR' pattern in V1 with clockwise rotation. The chest-X-ray revealed cardiomegaly, with a dilated pulmonary trunk and features of pulmonary venous congestion. Echocardiography demonstrated gross dilation of the left atrium, measured at 55 mm, 188% of our normal values, a left ventricular end-diastolic

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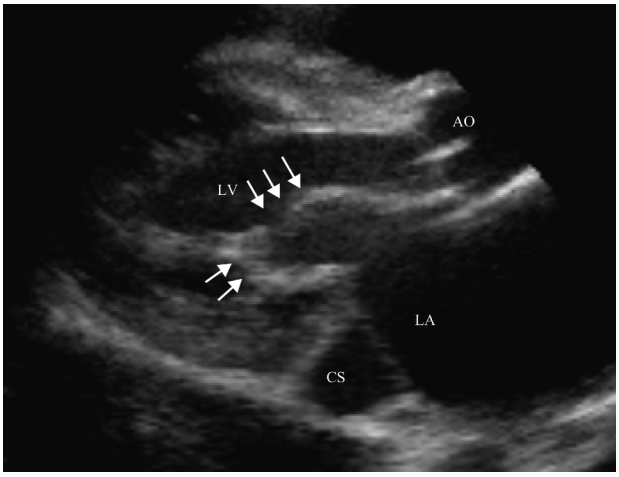

Figure 1.
The long-axis parasternal echocardiogram shows the abnormally oriented mitral valve (arrows). LA: left atrium; LV: left ventricle; AO: aorta; CS: coronary sinus.

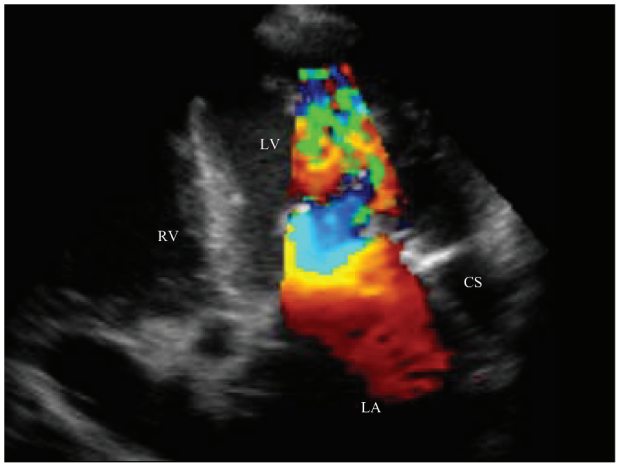

Figure 2.
The four-chamber apical view with colour-flow Doppler. LA: left atrium; CS: coronary sinus; LV: left ventricle; RV: right ventricle.

dimension of 61 mm, 133% of normal, and an end-systolic dimension of 44 mm, 145% of normal. The shortening fraction, at 28%, indicated mild systolic dysfunction of the left ventricle. Multiple trabeculations and recesses were observed in the left ventricular apex, consistent with non-compaction of the left ventricular myocardium. The mitral valve remained asymmetric, with one of the papillary muscles remaining hypoplastic. The orifice of the asymmetric mitral valve had formed along an abnormal vertical plane in the left ventricle (Fig. 1), producing an abnormal course of the blood (Fig. 2). The area of the valvar orifice was calculated as 1.76 cm^2/m^2, representing moderate stenosis. The pulmonary pressure was estimated at 90/55 mmHg. Repair was not possible because of the shortened and fibrotic cords and the extent of deformation of the valve. Instead, the valve was replaced by a 27 mm St. Jude bileaflet prosthesis, followed by an intraoperative ablation for treatment of the atrial fibrillation. One year postoperatively, the patient was clinically well. Her pulmonary hypertension had resolved completely, and sinus rhythm was maintained without antiarrhythmic medication. Her systemic blood pressure was 125/75 mmHg, with continued use of enalapril, furosemide, and spironolactone. Echocardiography now demonstrated a decrease in the size of the left atrium to 44 mm, 151% of normal, but the left ventricular dimensions and systolic function had not improved.

Discussion

More than one-fifth of girls with Turner's syndrome have congenital cardiac malformations, most commonly coarctation of the aorta or aortic valvar abnormalities.[1] The spectrum of left heart involvement ranges from an asymptomatic bicuspid aortic valve to full-blown hypoplasia of the left heart. The spectrum of heart defects may be associated with a particular karyotype.[4] Although persistence of the left superior caval vein is less typical,[5] it has been detected in one-tenth of patients with Turner's syndrome,[6] this being ten times the frequency in the general population. The association of a parachute-like mitral valve and a left superior caval vein, however, is rare, and to our knowledge has not been previously described in patients with Turner's syndrome.

Persistence of the left superior caval vein, or more exactly an enlarged coronary sinus, can produce obstruction of both the left ventricular inflow[7] and outflow.[8] It has been hypothesized that the enlarged coronary sinus can change the normal hemodynamics in the right atrium during fetal life, leading to a smaller than normal right-to-left shunt and diminished left ventricular flow.[9] We suspect that the non-compaction of the left ventricular myocardium, the development of the asymmetric parachute-like mitral valve, and persistence of the left superior caval vein, may have been embryologically correlated in this patient. Asymmetric papillary muscular involvement may represent a persistent embryonic situation that was caused by a disturbed delamination of either the anterior or the posterior part of the trabecular ridge from the ventricular wall.[10] This disturbance could have been associated with the arrest of ventricular myocardial compaction. If an arrest of compaction could be caused by disturbed hemodynamics, both the asymmetric mitral valve and the non-compaction could have resulted from the widened coronary sinus and its impact on left atrioventricular flow.[7]

It is often difficult properly to assess the significance of mitral stenosis caused by a parachute-like mitral valve. The assessment of the orificial area alone may underestimate the severity of the stenosis. Atypical multilinear directions of the stenotic flow may lead to the underestimation of the Doppler diastolic pressure gradient. This should be considered especially when the presence of an enlarged coronary sinus, with persistence of the left superior caval vein, makes the problem with the mitral valve even more complex.

References

1. Mazzanti I, Cacciati E. Congenital heart disease in patients with Turner's syndrome. Italian Study Group for Turner Syndrome (ISGTS). J Pediat 1998; 133: 688–692.
2. Sybert VP. Cardiovascular malformations and complications in Turner syndrome. Pediatrics 1998; 101: E11.
3. Bastianon V, Pasquino AM, Giglioni E, et al. Mitral valve prolapse in Turner syndrome. Eur J Pediatr 1989; 6: 533–534.
4. Gotzsche CO, Krag-Olsen B, Nielsen J, Sorenson KE, Kristensen BO. Prevalence of cardiovascular malformations and association with karyotypes in Turner's syndrome. Arch Dis Child 1994; 71: 433–436.
5. Hilton T, Castello R, Ohar J, Kong B, Kern M, Labovitz A. Persistent left superior vena cava in Turner's syndrome: a transesophageal echocardiographic study. Am Heart J 1992; 123: 234–236.
6. Douchin S, Rossignol AM, Klein SK, Siche JP, Baguet JP, Bost M. Heart malformations and vascular complications associated with Turner's syndrome. Prospective study of 26 patients. Arch Mal Coeur Vaiss 2000; 93: 565–570.
7. Cochrane AD, Marath A, Mee FBB. Can a dilated coronary sinus produce left ventricular inflow obstruction? An unrecognized entity. Ann Thorac Surg 1994; 58: 1114–1116.
8. Agnoletti G, Annecchino F, Preda L, Borghi A. Persistence of the left superior caval vein: can it potentiate obstructive lesions of the left ventricle? Cardiol Young 1999; 9: 285–290.
9. Macedo AJ, Oosthoek PW, Wenink ACG, Bartelings MM. Does persistent left superior vena cava to coronary sinus influence the growth of the left heart in the fetus? Cardiol Young 1996; 6 (Suppl 1): S43.
10. Oosthoek PW, Wenink ACG, Wisse LJ, Gittenberger-de Groot AC. Development of the papillary muscles of the mitral valve: morphogenetic background of parachute-like asymmetric mitral valves and other mitral valve anomalies. J Thorac Cardiovasc Surg 1998; 116: 36–46.