Septal defect with polyvalvular involvement: A cardiac imaging hallmark of Trisomy 18

Usha Nandhini Sennaiyan[1], S. Kiruthika[2], Mani Ram Krishna[1]

[1]Department of Pediatric Cardiology, R.K. Hospital for Women and Children, Thanjavur, Tamil Nadu, India, [2]Department of Pediatrics, Thanjavur Medical College, Thanjavur, Tamil Nadu, India

ABSTRACT

Congenital Heart Diseases occur in close to 90% of children with Trisomy 18. A ventricular septal defect along with abnormalities of more than one cardiac valve is considered to be an imaging hallmark of Trisomy 18. We present echocardiographic images of an infant with Trisomy 18 who had a large ventricular septal defect and abnormalities of all cardiac valves.

Keywords: Myxomatous AV valves, bicuspid semilunar valves, chromosomal abnormalities

A 4-month-old infant was referred to us for evaluation of heart failure. Her birth weight was 2.4 kg, and she weighed only 2.9 kg. There were dysmorphic features including low set ears, small head, micrognathia, short palpebral fissures, and clenched fists. There were no foot abnormalities. The infant was tachypneic with an enlarged liver and easily palpable femoral pulse. The first heart sound was normal. There was an ejection click, and the pulmonic component of the second heart sound was loud. There was a pan systolic murmur at the left para-sternal border and a mid-diastolic murmur at the apex.

An echocardiogram was performed. There was usual atrial and abdominal arrangement. The atrioventricular and ventriculoarterial connections were concordant. The atrioventricular valves had redundant and myxomatous leaflets [Figure 1a and b; Videos 1 and 2]. The mitral valve was competent, and there was mild tricuspid valve incompetence. There was a large mal-aligned peri-membranous ventricular septal defect. The aortic valve was bicuspid [Figure 2a and Video 3]. The pulmonary valve was tricuspid [Figure 2b]. However, there was fusion of the left and right posterior cusps, resulting in a functionally bicuspid valve [Figure 2a and Video 4]. Both the outflows were unobstructed, and the valves were competent. There was a patent arterial duct, and the aortic arch was unobstructed. The presence of a ventricular septal defect along with bicuspid semilunar valves in a dysmorphic child is considered a feature of Trisomy 18. Chromosomal microarray analysis was

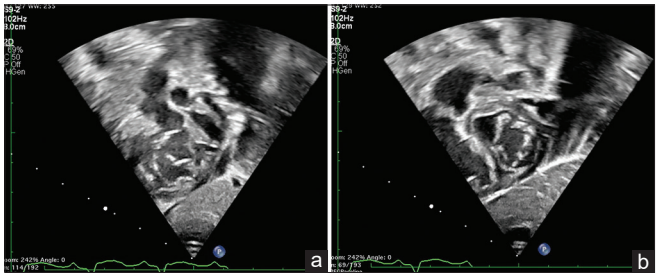

Figure 1: (a) Two-dimensional echocardiography view in end diastole demonstrating myxomatous tricuspid valve leaflets. (b) Two-dimensional echocardiography in the subcostal in the subcostal view in end diastole demonstrating myxomatous and redundant mitral valve leaflets

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Address for correspondence: Dr. Mani Ram Krishna, Dr R.K. Hospital for Women and Children, No 7, V.O.C. Nagar, Thanjavur - 613 007, Tamil Nadu, India. E-mail: mann_comp@hotmail.com

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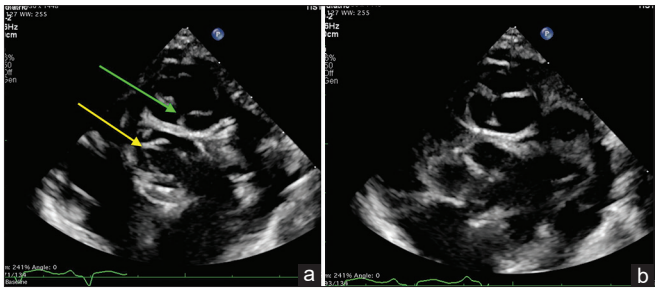

Figure 2: (a) Two-dimensional echocardiography in the para-sternal short axis view in systole. The aortic valve is bicuspid (yellow arrow). The pulmonary valve is also functionally bicuspid with the fusion of the right and left posterior leaflets (green arrow). (b) Two-dimensional echocardiography in the modified para-sternal short axis view in diastole. This shows that the pulmonary valve has three cusps with an inverted Mercedes Benz appearance

performed on peripheral venous blood. This identified a duplication spanning 77,878 kilobase pairs which was consistent with Trisomy 18.

Trisomy 18 is the second most common autosomal trisomy with a live birth incidence of around 1 in 6000.[1] It is commonly caused by nondisjunction and more rarely by mosaicism and partial trisomy of the long arm of chromosome 18. Congenital heart diseases are the most common structural abnormalities in Trisomy 18 present in >75% of cases[2] with shunt lesions and polyvalvular involvement present in a vast majority.[3] Other causes of polyvalvular disease include rheumatic heart disease in older children and adults, inflammatory disorders as well as the less common polyvalvular heart disease syndrome which is inherited in an autosomal dominant fashion.

The presence of a posttricuspid shunt lesion (ventricular septal defect or persistent arterial duct) with abnormalities of the heart valves is considered as a cardiac hallmark of Trisomy 18. In an early autopsy series, all valves were noted to be dysplastic with evidence of vacuolar degeneration of the spongiosa layer and a distinct lack of elastic tissue.[4] This was purported to be due to an arrest in embryonic development of the valve tissue in the second trimester. In the largest autopsy series on Trisomy 21, Stella Van Praagh *et al.* observed a very high incidence of redundant leaflets in all four cardiac valves with the tricuspid valve most frequently involved.[3]

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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