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Images in Cardiology

Myriad manifestations of Williams syndrome

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ABSTRACT

4 months male child presented with failure to thrive. On general examination child had normal O2 saturation with characterstic elfin facies. Further evaluation of the patient showed major manifestations of Williams syndrome in form of supravalvar aortic stenosis, branched pulmonary artery stenosis along with cardiomyopathy. Although the entity is known, this article shows comprehensive diagnostic workup with the aid of multimodality imaging techniques. The genetic diagnosis of Williams syndrome was confirmed using fluroscent in situ hybridisation techniques (FISH). In this patient most of the manifestations of elastin vasculopathy were noted in the form of involvement of ascending aorta, pulmonary arteries and myocardium. We also want to emphasis the importance of echocardiography in newborn patients with dysmorphic facies as Williams syndrome can be easily missed in neonatal period.

A 4 month old male child presented with symptoms of heart failure and poor weight gain. On examination, O_2 saturation in both limbs was 99% and there was no significant blood pressure difference in all four limbs. He had a characterstic 'elfin facies' with a sunken nasal bridge, a long philtrum, wide mouth, prominent lower lip, small chin and low set ears (Fig. 1) .2D echocardiography showed situs solitus, concentric left ventricular hypertrophy with mild narrowing in the supravalvular region (Fig. 2) with systolic gradients of 26 mm Hg across the segment (Fig. 3) (Online Video 1). The right pulmonary artery (RPA) after its origin showed significant short segment stenosis with peak systolic gradients of 32 mmHg. The left pulmonary artery (LPA) after its origin showed mild narrowing (Figs. 4 and 5) (Online Video 2). Cardiac CT demonstrated supravalvular aortic stenosis

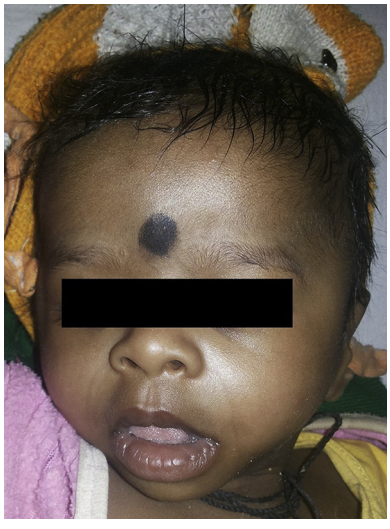

Fig. 1 – Characterstic elfin facies.

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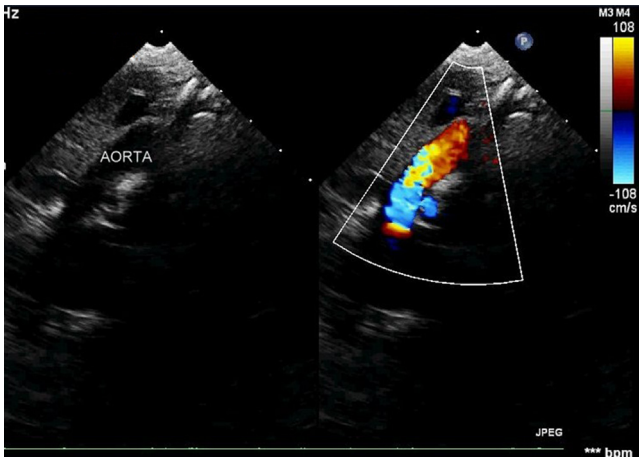

Fig. 2 – 2D echocardiography showing mild narrowing in the supravalvular region.

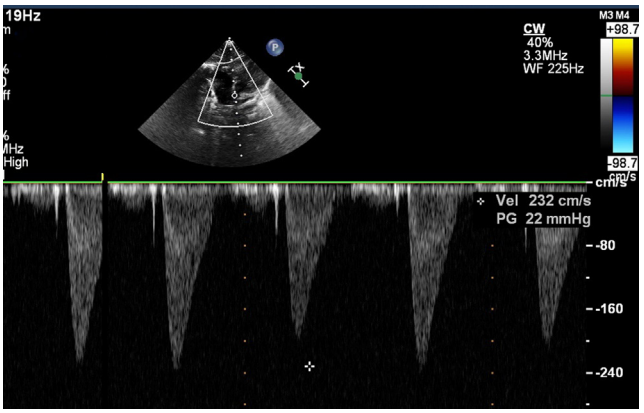

Fig. 5 – Doppler investigation showing peak systolic gradients of 32 mmHg across the narrowed segment of RPA.

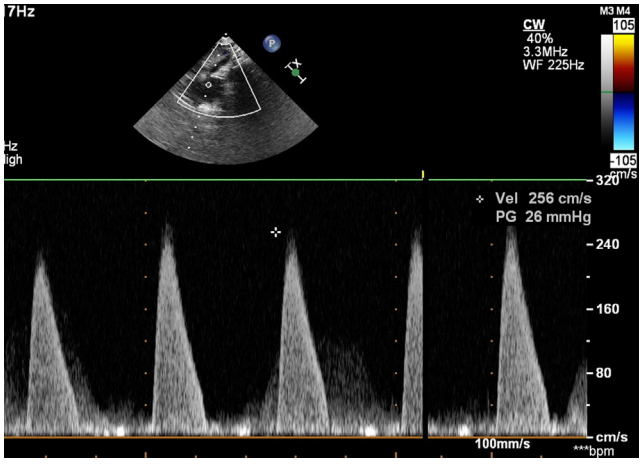

Fig. 3 – Doppler showing gradients of 26 mm Hg across narrowed supravalvular region.

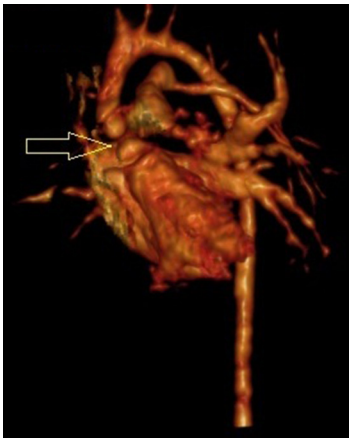

Fig. 6 – Cardiac CT with 3D reconstruction showing supravalvular aortic stenosis (SVAS).

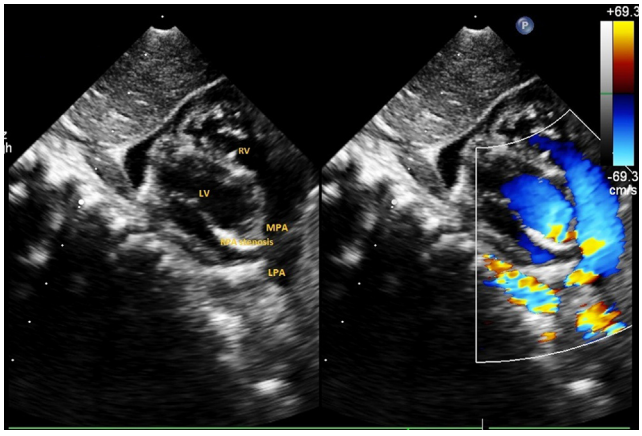

Fig. 4 – 2D echocardiography with color Doppler showing bilateral PAS.

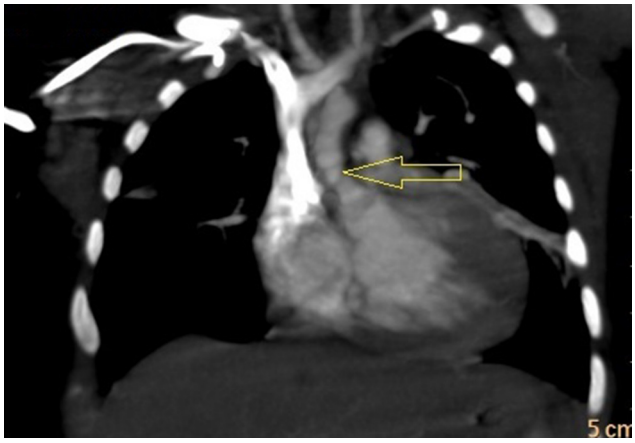

Fig. 7 – Cardiac CT (Coronal section) showing SVAS.

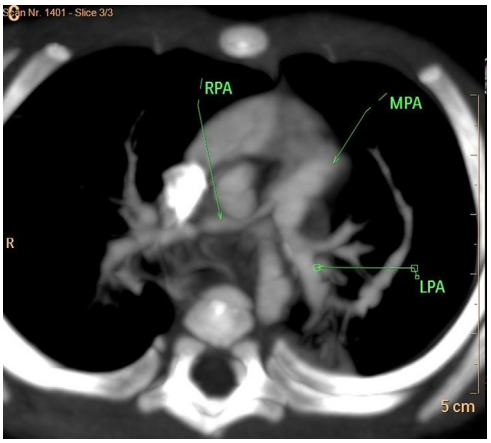

Fig. 8 – Cardiac CT (Transverse section) showing bilateral PAS.

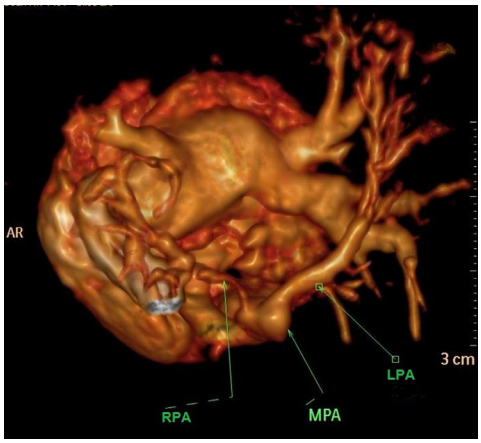

Fig. 9 – Cardiac CT with 3D reconstruction showing bilateral pulmonary artery stenosis (PAS) involving RPA more than LPA.

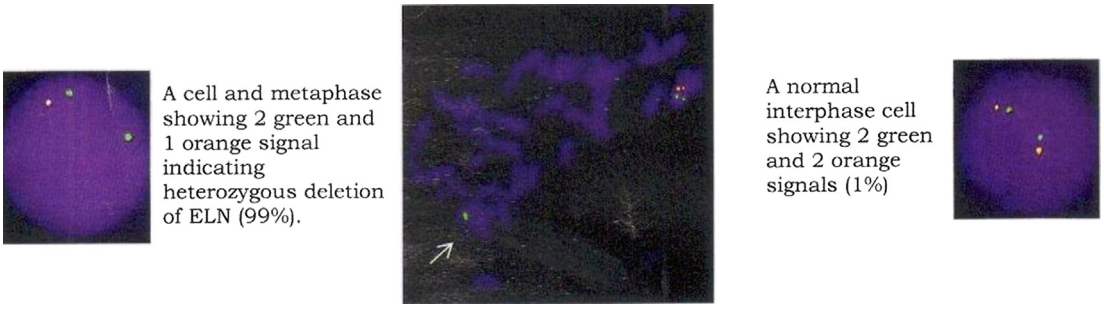

Fig. 10 – Fluorescent in-situ hybridization studies showing deletion of elastin gene (Chromosome 7q11.23).

(SVAS) with hour glass appearance of the aorta (Figs. 6 and 7) with bilateral pulmonary artery stenosis (PAS) involving RPA more than LPA (Figs. 8 and 9). Fluorescent in-situ hybridization (FISH) studies (Fig. 10) showed heterozygous deletion of elastin gene (Chromosome 7q11.23) and confirmed the diagnosis of Williams syndrome (WS).

Supplementary data related to this article can be found online at http://dx.doi.org/10.1016/j.ihj.2015.02.026.

WS is a genetic disorder occurring with a frequency of 1 in 20,000–50,000 live births. Manifestations of WS include congenital heart disease, hypertension, dysmorphic facial features, infantile hypercalcaemia and mental retardation. Apart from supravalvular AS and branched PA stenosis other cardiac abnormalities observed are bicuspid aortic valve, mitral valve regurgitation, coarctation of the aorta, ventricular or atrial septal defects. In neonates, cardiovascular symptoms were evident in 47% of WS children.[1] PA stenosis often tends to regress spontaneously and SVAS tends to progress with time. In this patient most of the manifestations of elastin vasculopathy were noted in the form of involvement of ascending aorta and pulmonary arteries. The concentric left ventricular hypertrophy observed in our patient may be an expression of hypertrophic cardiomyopathy which is known to be associated with WS.[1] In neonatal period all newborn patients with dysmorphic facies should be evaluated with echocardiography so that the cardiac abnormalities are not missed.

Conflicts of interest

All authors have none to declare.

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