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Images in cardiology

Inferior vena cava stenosis: Echocardiographic diagnosis in Marfan syndrome

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ABSTRACT

Marfan syndrome is a genetic disease with variable clinical presentation. This case describes a 36-year-old lady who was diagnosed with Marfan syndrome based on revised Ghent criteria. She was found to have bicuspid aortic valve and sensorineural hearing loss. Inferior vena cava stenosis was suspected on echocardiography due to high velocity flow and visualization of a focal narrowing in the inferior vena cava proximal to hepatic vein entry. Inferior vena cava stenosis was confirmed by computed tomography. Echocardiographic features suggestive of inferior vena cava stenosis include detection of a focal narrowing and high turbulent flow, peak velocity > 1.5 m/s and S/D wave fusion on spectral Doppler.

Keywords: Marfan, IVC, echocardiography, sensorineural, ectopia lentis

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INTRODUCTION

Marfan syndrome is an inherited autosomal dominant disorder of connective tissue disease associated with a wide clinical variability and a prevalence of 1:5000.¹ We are not aware of a description of any association between Marfan syndrome and inferior vena cava (IVC) disorders. In our case study we show evidence of congenital inferior vena cava stenosis in Marfan syndrome.

THE CASE

A 36-year-old female was labelled as a case of possible Marfan syndrome at age of 11, based on the finding of bilateral ectopia lentis when she complained of reduced visual acuity. Ectopia lentis was confirmed by an ophthalmologist based on slit lamp examination. During the same period she was noted as having hearing difficulties. Bilateral moderate sensorineural hearing loss was confirmed by tympanometry. The patient's intelligence was normal. All routine basic laboratory tests were checked and found to be normal, including urine sulfur containing amino acid for homocystinurea. The patient lacked any musculoskeletal signs of Marfan syndrome and family history of Marfan syndrome was also negative. However, she had seven siblings, of which two sisters died in infancy of unknown reasons. Father and mother were cousins.

The patient was lost on follow-up until she was referred to the cardiology service for atypical chest discomfort. On echocardiogram, aortic root diameter was found to be 35 mm (Figure 1) based on measurement utilizing diastole and leading edge-to-leading edge measurement of the sinuses of Valsalva.²

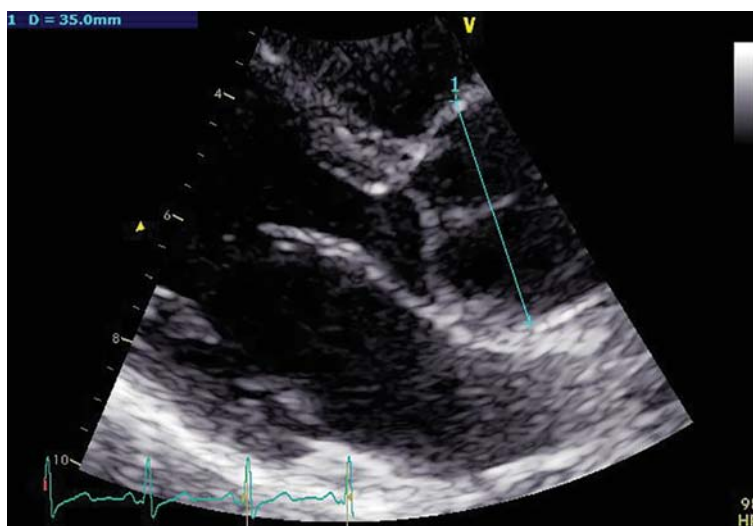


Figure 1. Aortic root measurement at sinus of Valsalva utilizing leading edge-to-leading edge technique. Diameter (D) of 35 mm is measured.

Consequently, her aortic root Z score was 2.6. Thus, Marfan syndrome was confirmed based on the revised Ghent nosology.³ The aortic valve was bicuspid, without any hemodynamically significant dysfunction (Figure 2).

Furthermore, interestingly, a narrowing of the inferior vena cava (IVC) proximal to hepatic vein entry was noticed on sub-costal views measuring 5.2 mm compared to proximal segment of 15 mm and distal segment of 15 mm (Figure 3).

Doppler studies of the IVC revealed a high velocity turbulent flow with a peak velocity of 2.2 m/s, a peak gradient of 19.3 mmHg and a mean gradient of 16.9 mmHg with fusion of S/D waves on parasternal RV inflow (Figure 4). Such findings were highly suggestive of IVC stenosis.

Accordingly, other imaging modalities were requested for further evaluation. Computed tomographic angiography of the abdomen confirmed the presence of IVC stenosis proximal to hepatic vein entry. However no IVC interruption was noted (Figure 5).

Magnetic resonance imaging (MRI) of the spine did not show evidence of dural ectasia. The patient denied history suggestive of significant IVC stenosis i.e. lower limb edema, varicose veins and abdominal pain.



Figure 2. Bicuspid aortic valve seen during systole.

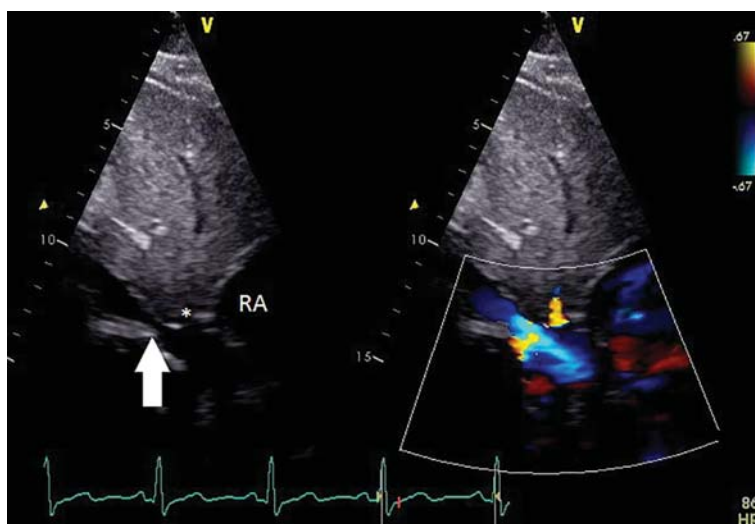


Figure 3. 2D and 2D with color mapping image showing a narrowed IVC (arrow). Hepatic vein is seen (asterisk). RA: Right atrium.

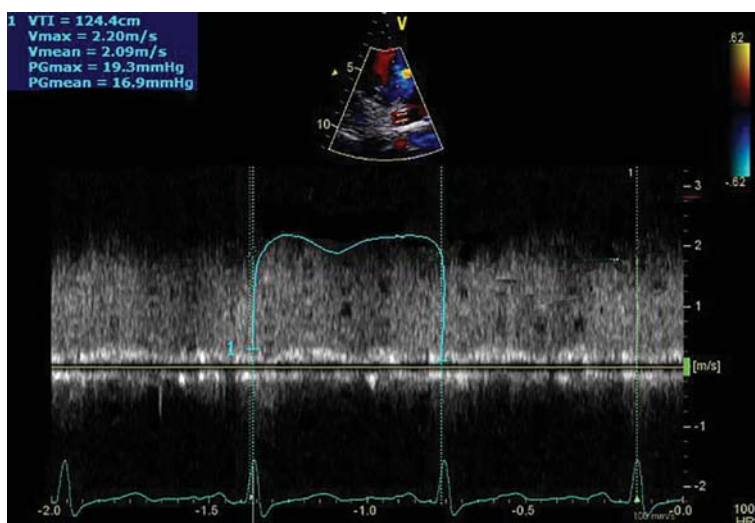


Figure 4. Pulsed wave Doppler of the IVC flow in parasternal RV inflow view.

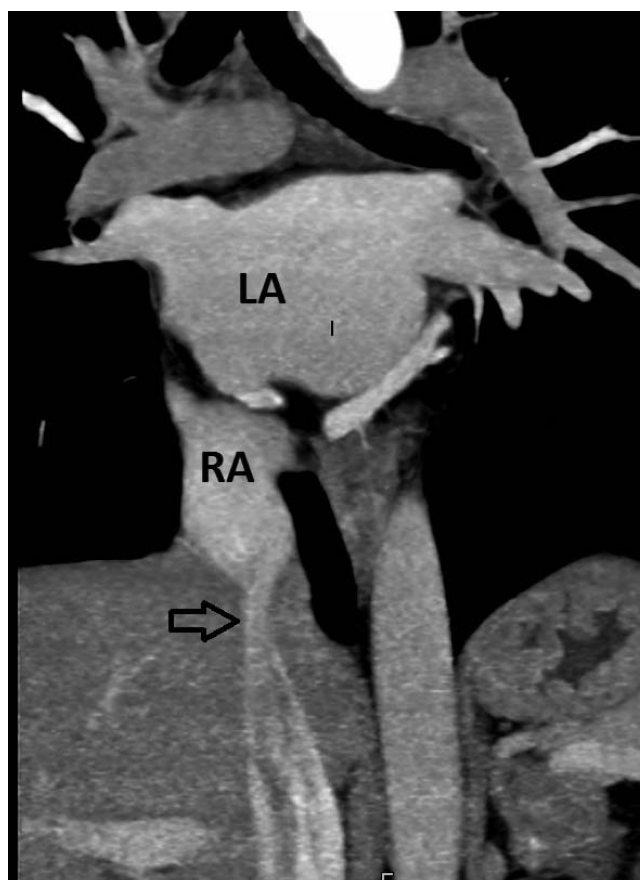


Figure 5. CTA showing focal IVC stenosis (arrow). Measurements were taken and corresponded to those of echocardiogram. RA: right atrium, LA: left atrium.

DISCUSSION

Is it really Marfan? Upon extensive review of the literature of Marfan syndrome, no association between sensorineural hearing loss and Marfan syndrome was noted. However, conductive hearing loss in association with Marfan syndrome was described.⁴ To the best of our knowledge, no known association between bicuspid aortic valve, ectopia lentis and sensorineural hearing loss has been previously described. However, bicuspid AV prevalence in Marfan is higher than in the general population (4.7% vs 0.5%).⁵ Congenital IVC stenosis was not reported to be associated with Marfan nor bicuspid AV.

Congenital IVC stenosis is characterized by narrowing with or without a web formation mostly at the diaphragmatic level or hepatic segment of the IVC.⁶ The prevalence of interruption or congenital stenosis of the IVC is 0.15% in the general population. Symptoms include leg swelling (33%); leg pain (17%); varices of lower extremities (17%); abdominal pain (17%); and, rarely, hematochezia (8%). The most common cause of presenting symptoms was DVT.⁷

Table 1. Normal IVC Doppler velocity measured at IVC orifice (n = 16).

Peak	cm/s
Inspiration	67.9 ± 12.8 (47–98)
Expiration	34.5 ± 7.0 (25–52)

Adopted from Minagoe S et al. Obstruction of inferior vena caval orifice by giant left atrium in patients with mitral stenosis. A Doppler echocardiographic study from the right parasternal approach. *Circulation*. 1992 Jul;86(1):214–225.

Table 2. Echocardiographic findings suggestive of IVC stenosis.

Findings	Echocardiographic technique
Demonstration of focal IVC narrowing	2D echocardiography
Demonstration of turbulent flow	Color Doppler
Peak velocity > 1.5 m/s	Spectral Doppler (PW and CW)
Loss of S and D wave distinction	Spectral Doppler (PW and CW)

Multidetector row computed tomography (MDCT) and magnetic resonance imaging (MRI) are the most reliable methods for identification of these anomalies.⁸

Specific echocardiographic criteria of IVC stenosis is lacking. In a study of impact of a large left atrium in mitral stenosis on the IVC, normal IVC velocities were documented (Table 1).⁹ Giving the lack of echocardiographic criteria to diagnose IVC stenosis and upon review of different venous stenosis cases that include superior vena cava and pulmonary veins, we recommend evaluation of multiple parameters of IVC in 2D, color Doppler and spectral Doppler (Table 2) in cases where IVC stenosis is suspected.

However, the hemodynamic state of the patient, volume status, and RA pressure should be taken into consideration at all times as these parameters may influence IVC flow.

CONCLUSION

We believe that our case is complex and deserves extensive genetic testing of all possible syndromes responsible for sensorineural hearing loss, and those for ectopia lentis, including testing for fibrillin-1 (FBN1) mutation of Marfan syndrome.

The revised Ghent criteria has put more weight on the aortic root diameter. The presence of bicuspid aortic valve in our case can cause aortic root dilatation by itself¹⁰ and makes it possible that Marfan syndrome in this particular case is over diagnosed.

Meanwhile, we decided to treat our patient as a case of Marfan syndrome. Since, our patient is asymptomatic regarding IVC stenosis we decided not to intervene. IVC stenosis is a rare disorder and once suspected by echocardiographic criteria, MDCT or MRI examination should be made to confirm the diagnosis.

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ABBREVIATIONS

IVC Inferior vena cava
 2D Two-dimensional
 MDCT Multi detector computed tomography
 CTA Computed Tomographic angiography
 MRI Magnetic resonance imaging
 RA Right atrium
 LA Left atrium

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