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Delayed Diagnosis of Shone Syndrome in a Patient Planning for Pregnancy

Niloufar Samiei[1], Sepehr Gohari[2,3], Hassan Ahangar[4*]

1. *Heart Valve Research Center, Rajaie Cardiovascular, Medical and Research Center, Iran University of Medical Sciences, Tehran, Iran*
2. *Student Research Committee, School of Medicine, Zanjan University of Medical Sciences, Zanjan, Iran*
3. *Dept. of Family Medicine, Alborz University of Medical Sciences, Alborz, Iran*
4. *Dept. of Cardiology, Mousavi Hospital, School of Medicine, Zanjan University of Medical Sciences, Zanjan, Iran*

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Corresponding Information:
Hassan Ahangar,
Dept. of Cardiology, Mousavi Hospital, School of Medicine, Zanjan University of Medical Sciences, Zanjan, Iran

E-Mail: hassan.ahangar@yahoo.com

ABSTRACT

Shone syndrome is a rare congenital cardiac abnormality; however, many of the cases remain undiagnosed until early and middle adulthood. Different imaging modalities are used to assess the related structural abnormalities. In this case study, we report a 32-year-old woman who was planning her first pregnancy. In light of her childhood heart problems, in addition to a history of extended penicillin prescriptions for several years, she was referred for complementary assessments. At the time of presentation, she was asymptomatic. Imaging results showed several structural obstructive left-sided lesions and pulmonary artery hypertension. Ultimately, the patient was diagnosed with congenital shone syndrome, which was initially misdiagnosed. Shone complex in our case was presented in its full form, which could be potentially fatal in case of pregnancy.

Keywords: Congenital heart disease, Echocardiography, Shone syndrome, Shone's complex, Pregnancy

Introduction

Shone syndrome (Shone complex) is a collection of left-sided obstructive heart lesions, which accounts for about 0.6% of congenital cardiac abnormalities (1). Formerly, four characteristics were defined for Shone syndrome: Coarctation of the aorta, parachute mitral valve, supramitral ring, and subaortic stenosis (2). Recently, four other lesions have been reported to contribute to this syndrome: Cor triatriatum, bicuspid aortic valve and small aortic valve annulus, hypoplastic (stiff) left ventricle, and small aortic arch. The presence of two or more of the main characteristics is defined as Shone syndrome (3, 4).

Case Presentation

We report the case of a 32-years-old woman who was planning for her first pregnancy and referred for complementary evaluations due to her prior cardiac disease. She had a history of Failure to Thrive (FTT) and dyspnea on exertion (Function class I-II) during early childhood and referred to cardiologist for diagnostic evaluations and at the age of 9 she was diagnosed with mild coarctation of the aorta [mean Pressure Gradient (PG)=15 mmHg], mild mitral stenosis (MS) [Mean gradient (MG)= 9 mmHg and mitral valve area= 1.6 cm^2] and mild mitral regurgitation (MR). She received medical care follow-up subsequent to her diagnosis and by the following 2 years echocardiographic imaging along with cardiac catheterization showed mild discrete coarctation of the aorta, bicuspid aorta valve and severe MS; however, information regarding the severity of MR was not provided. Percutaneous transvenous mitral commissurotomy (PTMC) was performed and trans-mitral pressure gradient reduced from 25 to 5 mmHg. Penicillin (1200000 unit monthly) was then prescribed until she was 18-year-old, with a possible diagnosis of rheumatic heart disease (RHD). At the time of admission, she had a complaint of occasional palpitation.

Physical exam was only remarkable for systolic murmur (III/VI) and diastolic rumbling murmur at the apex. The electrocardiogram had sinus rhythm with incomplete right bundle branch block (RBBB) morphology (rsr´ in lead V_1) and infrequent premature ventricular complex (PVC) with right ventricular outflow tract (RVOT) origin. The patient underwent resting transthoracic echocardiography (TTE) that demonstrated normal LV and RV size and systolic function (LVEF: 55%), severe left atrium (LA) enlargement (Suppl 1-Panel A), mild right atrium (RA) enlargement, bicuspid aortic valve (BAV) (Suppl 1-Panel B), coarctation of the aorta (peak PG= 39 mmHg; mean PG=21 mmHg) (Suppl 1-Panel C,D), progressive MS [Mitral valve area by 2D plantimetry= 1.9 cm^2, MG=11 mmHg and pressure half-time (PHT)= 124 msec], moderate MR (Suppl 1-Panel E), parachute like mitral valve and single papillary muscle (Suppl 1-Panel F-H), moderate so severe tricuspid regurgitation (TR) (Suppl 1-Panel I) and systolic pulmonary artery pressure (SPAP)=50 mmHG. As the patient was planning for pregnancy and there was a discrepancy between the medical history and the TTE findings, stress echocardiography was performed to assess the pregnancy stress tolerance and had the following findings: excellent functional capacity (estimated metabolic equivalents of task (METs=13.6, duration=10 min), Terminated at stage 4 of Bruce protocol, normal heart rate and blood pressure response, SPAP increased from 50 mmHg at rest to 85 mmHg at peak stress, trans-mitral MG increased from 11 mmHg at rest to 28 mmHg at peak stress and peak PG at coarctation site increased from 39 mmHg at rest to 60 mmHg at peak stress and mean PG increased from 21 mmHg at rest to 32 mmHg. Based on stress echocardiography results, pregnancy was considered high risk according to the induction of severe pulmonary hypertension with exercise. To assess the coarctation, an aortic computed tomography angiography (CTA) was performed which revealed 40% diameter reduction in descending thoracic aorta 13 mm post left subclavian artery suggestive of mild coarctation with post stenotic dilation. Additionally, a trans-esophageal echocardiogram (TEE) was done in order to better assess the cardiac structures. The TEE demonstrated a membranous partial cor-triatrum in LA (Suppl 2-Panel A) without obstruction extended at the interatrial septum (IAS) side of LA (Suppl 2-Panel B), significant LV inflow obstruction (area=0.7 cm^2, MG=23 mmHg) (Figure.1) with mild MS and moderate MR (Suppl 2-Panel C), all chorda tendineae attached to a single multi-head papillary muscle at posteromedial wall compatible with parachute mitral valve (Suppl 2-Panel D). Also, we detected BAV, small sub-aortic web (diameter=7 mm) (Figure.2, Suppl 2-Panel E) without left ventricular outflow tract (LVOT) obstruction, systolic turbulent ante-grade flow in descending aorta with 50 mmHg pressure gradient compatible with coarctation, moderate to severe TR (TRG= 50 mmHg) and SPAP= 55 mmHg at rest. Based on the mentioned findings, she was diagnosed with complete form of Shone Complex. Afterwards, the patient underwent catheterization procedure in which the PG reported to be 30 mmHg; consequently, interventional coarctoplasty was planned. Moreover, she scheduled for surgical mitral valve replacement due to the severe MS, MR and pulmonary hypertension.

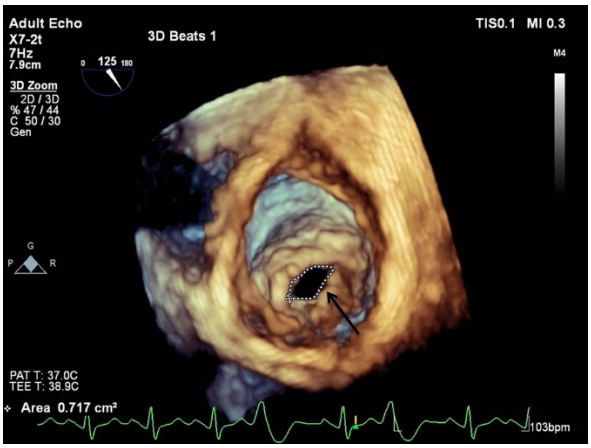

Figure 1. 3D TEE- Severe left ventricular inflow obstruction, The arrow shows the subvalvular mitral area (Area= 0.7 cm^2).

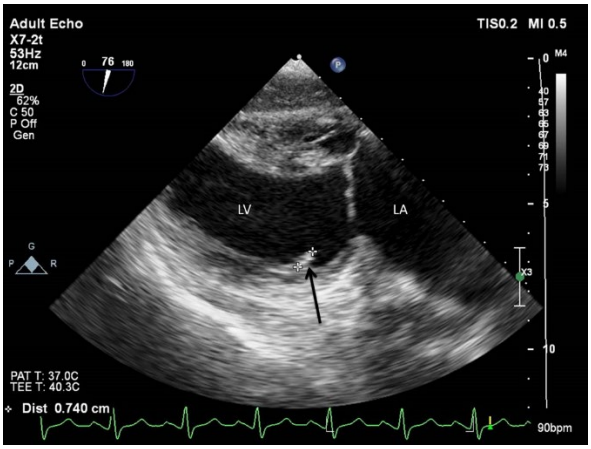

Figure 2. 2D TEE (Trans-gastric view)- The arrow shows small sub aortic web. LA (Left Atrium), LV (Left Ventricle).

Discussion

Shone complex is an under-recognized condition associated with relatively low mortality in adulthood but substantial morbidity (5). Different modalities of echocardiography, computed tomography angiography (CTA), and other diagnostic tools may be used to support the diagnosis of Shone complex. According to the number of the presented features, a series of surgical procedures would be performed to correct the abnormalities (6). This report highlights the challenges in the diagnosis of Shone complex due to the different

aspects of the disease. In addition, this report features the possibility of the prolonged use of unnecessary medications, such as penicillin, which could have been avoided by a more precise evaluation early on. Many Shone cases can be overlooked at early ages as a result of incomplete assessments and physicians only focusing on some aspects of this syndrome (3). Misdiagnosing the Shone complex presented in its full form could be potentially fatal for the mother and embryo during pregnancy. Many cases may be either asymptomatic or exhibit mild vague symptoms that are mainly disregarded. According to this case, one of the common findings of Shone complex in early childhood is coarctation of the aorta (7).

Conclusion

In the presence of coarctation of the aorta, the clinicians should highly suspect other complicated cardiac structural defects such as Shone complex.

Acknowledgments

None.

Conflict of Interest

None Declared.

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