Incomplete Shone's syndrome in a patient with left ventricular noncompaction

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Introduction

- Shone's syndrome is a rare congenital cardiac anomaly consisting of four obstructive defects including a supravalvular mitral ring, parachute mitral valve, subaortic stenosis, and aortic coarctation
- Patients with two or three of these defects have the incomplete form of Shone's syndrome
- Shone's syndrome frequently presents with other cardiac abnormalities, however it has rarely been described with concomitant left ventricular noncompaction

Case description

A 38-year-old male presented with left flank pain of one day duration. He additionally reported ongoing intermittent chest pain of several months duration.

Past medical history: heart failure with reduced ejection fraction secondary to non-ischemic cardiomyopathy

In patient work-up:

•**Labs:** BNP 590 pg/mL, troponin-I 0.35 ng/mL (falling to 0.27 ng/mL 5 hours later)

•**Chest CT:** left inferior renal pole infarct and aortic coarctation of the descending thoracic aorta

•**TTE:** LVEF of 35-40% along with several structural abnormalities, including: 1) prominent trabeculations of the left ventricle, 2) mitral valve with unusual configuration involving fusion of the lateral commissure, 3) aortic coarctation just after the origin of the left subclavian, 4) mildly obstructive subaortic membrane, and 5) bicuspid aortic valve

• **EKG:** sinus rhythm with a first degree atrioventricular block, a nonspecific intraventricular conduction block, and left ventricular hypertrophy by Cornell criteria

Figures

Figure 1. Aortic Coarctation

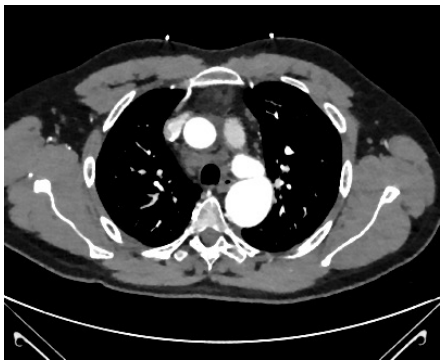


Figure 2. Subaortic Stenosis

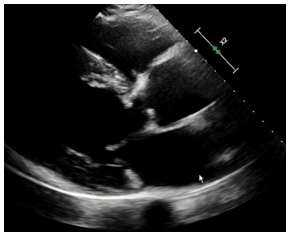
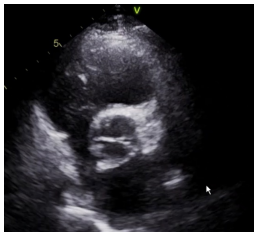


Figure 3. Bicuspid Aortic Valve



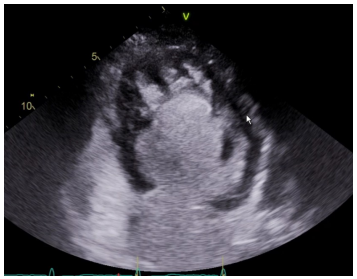


Figure 4. Left Ventricular Trabeculations

Management

- Treated with intravenous heparin for renal pole infarct and concern for non ST elevation myocardial infarction
- Surgical repair of the aortic coarctation
- Lifelong anticoagulation recommended by cardiology given risk of recurrent thromboembolic event
- Patient will continue to follow-up as outpatient with cardiothoracic surgery for serial echocardiographic surveillance of bicuspid aortic valve and subaortic membrane, which were mild on presentation and required no acute intervention

Discussion

- Shone's syndrome is known to present with other congenital cardiac abnormalities
- To our knowledge, a simultaneous presentation of Shone's syndrome and left ventricular noncompaction has only been described in one previous case report.
- An accurate diagnosis of these conditions is crucial, as Shone's syndrome creates risk of heart failure and pulmonary hypertension, while left ventricular noncompaction leads to increased risk of thromboembolic events and ventricular arrhythmias.

References

1. Toufan, M., Pourafkari, L., Baghbani-Oskouel, A., & Nader, N. D. (2018). Left ventricular non-compaction in a patient with Shone Anomaly. Türk Kardiyoloji Derneği Arşivi. https://doi.org/10.5543/tkda.2018.30136
2. Zhang, X., Ma, G., Zheng, L., Zhang, H., Sun, Y., Li, J., & Ma, N. (2020). Echocardiographic diagnosis of Shone's syndrome. Echocardiography-a Journal of Cardiovascular Ultrasound and Allied Techniques, 37(12), 2139–2143. https://doi.org/10.1111/echo.14899