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CLINICAL CASE

Hypertrophic Cardiomyopathy in a Behçet's Patient; a Chance or Chess

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ABSTRACT

Hypertrophic Cardiomyopathy (HCM) is an inherited autosomal dominant cardiomyopathy characterized by marked genotypic and phenotypic heterogeneity. On the other hand, Behçet's disease (BD) is a recurrent multisystemic inflammatory disorder associated with vasculitis and recurrent arterial and venous thrombosis. The etiology of BD is linked to viral, toxic, bacterial, and immunological factors. To date, no cases have been reported describing an association between BD and Hypertrophic Cardiomyopathy (HCM).We present a case of 31-year-old male with Behçet's disease and a history of recurrent DVT who presented with atypical chest pain and dyspnea that has been ongoing for 1 year. An echocardiography study revealed an asymmetrical increase in left ventricular wall thickness and significant provocable left ventricular outflow tract obstruction (LVOTO). A diagnosis of hypertrophic obstructive cardiomyopathy was confirmed by cardiac magnetic resonance (CMR). The potential relationship between Behçet's disease and hypertrophic cardiomyopathy remains unclear. While this coexistence may be purely coincidental, the absence of previous reports on this association raises the possibility of an underlying pathophysiological link that has yet to be explored.

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KEYWORDS Behçet's disease, Hypertrophic cardiomyopathy

Background

Hypertrophic cardiomyopathy (HCM) is an inherited autosomal dominant cardiomyopathy characterized by marked genotypic and phenotypic heterogeneity with variable clinical expressions ranging from asymptomatic status to progressive heart failure or sudden death. On the other hand, Behçet's disease is known as a recurrent multi systemic and inflammatory disease associated with vasculitis and recurrent thrombotic arterial and venous thrombosis. The etiology of Behçet's disease is associated with viral, toxic, bacterial, and immunological factors[1]. As far, this is the first reported case of Behçet's disease associated with Hypertrophic obstructive cardiomyopathy.

Case presentation

A 31-year-old male with a past medical history only significant for Behçet's disease that was diagnosed 7 years ago based on a history of recurrent painful oral ulcers, genital ulcers, and recurrent unprovoked bilateral lower extremity deep venous thromboses. The patient was referred for cardiology evaluation due to chest pain and dyspnea that has been ongoing for 1 year. He describes his chest pain as a heaviness sensation in the precordial area, episodic in nature, and only lasts a few minutes for each episode. It is not precipitated or worsened by exertion or deep breathing. He also reports dyspnea on moderate exertion with no orthopnea or paroxysmal nocturnal dyspnea. He denies any history of syncope or pre-syncope, fever, cough, or hemoptysis. He is a current smoker with a smoking index of 150 cigarette-years. He does not have a family history of similar conditions or sudden cardiac death. His vitals were generally unremarkable with blood pressure 110/70 mmHg, equal on both sides. Complete physical examination was only noticeable for bilateral gaiter area hyperpigmentation, multiple venous ulcers at the dorsal aspect of the right foot and lax calf muscles. Cardiac examination was remarkable for systolic ejection murmur, with maximal intensity at the left lower sternal border, which intensifies on the straining phase of Valsalva. ECG showed a normal sinus rhythm with no specific changes (figure 1). A combined Transthoracic echocardiogram (figure 2), and Transesophageal echocardiogram (figure 3, and video 1) showed normal left ventricular dimensions and systolic function (EF 69%), asymmetric septal hypertrophy with a maximal thickness of 1.4 cm, posterior wall thickness of 1.1 cm, mitral valve systolic anterior motion (SAM), and significant Left ventricular outflow tract obstruction (LVOTO) with a maximum provoked gradient of 98 mmHg which raises concerns for a diagnosis of Hypertrophic Obstructive Cardiomyopathy (HOCM). Cardiac Magnetic Resonance (figure 4) confirmed HOCM diagnosis by asymmetric septal hypertrophy (mid-septum of 1.5 cm) and a few tiny areas of left ventricular fibrosis.

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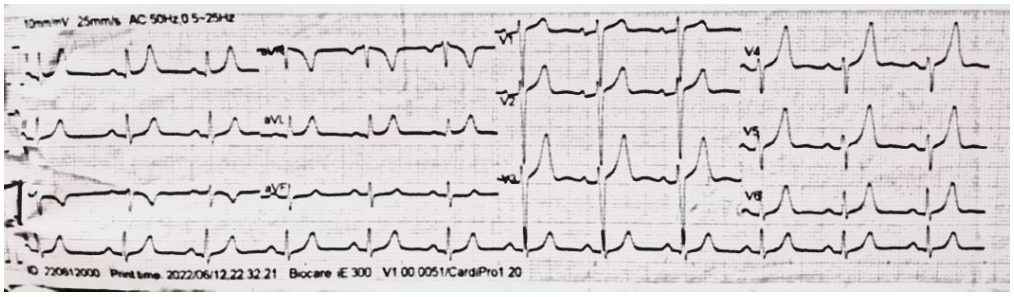

Figure 1 ECG showing sinus rhythm and no specific changes

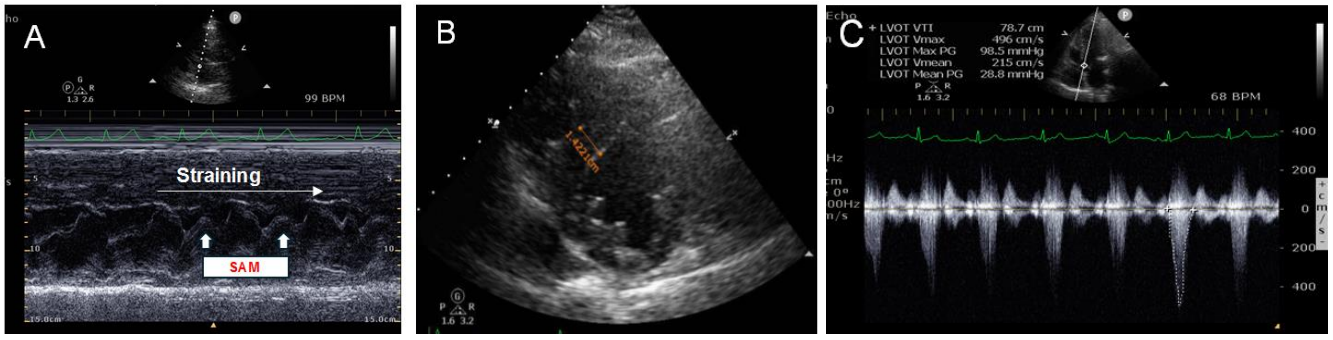

Figure 2 Transthoracic Echocardiogram. A) M-mode on MV leaflets showing significant SAM during Valsalva (arrowheads). B) Short axis on left ventricle, showing asymmetrical septal hypertrophy. C) CW Doppler on LVOT during Valsalva, showing significant obstruction with a peak gradient of 98 mmHg.

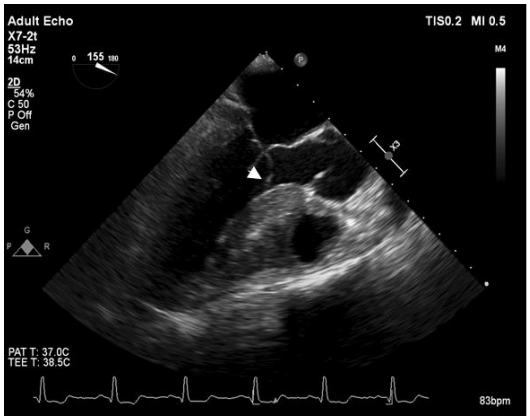

Figure 3 Transesophageal echocardiogram shows systolic anterior motion (SAM) at mid-systole (arrowhead).

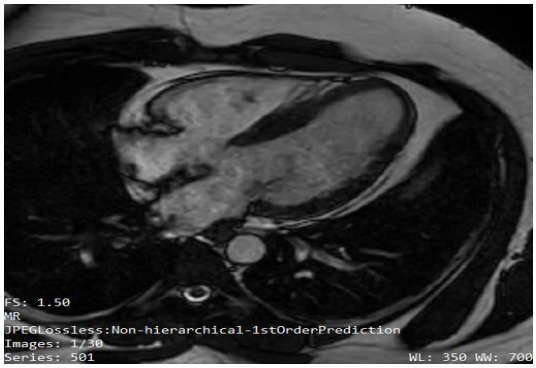

Figure 4 Cardiac Magnetic Resonance: 4-chamber view showing asymmetrical septal hypertrophy.

Discussion

Hypertrophic cardiomyopathy is a complex genetic cardiovascular disorder. It is defined as an increase in wall thickness of ≥15 mm anywhere in the left ventricle, in the absence of another cause of hypertrophy[2]. HCM may be presented as an isolated disease or associated with other clinical syndromes such as mitochondrial myopathies, glycogen storage diseases in children, and Fabry, and amyloid cardiomyopathy in adults[3]. Many HCM patients are asymptomatic, however, some may experience angina, dyspnea, palpitations, or syncope. On the other hand, Behçet's disease, as an inflammatory disease, can affect the heart by causing vasculitis, and arterial and/or venous thrombosis, which may involve the coronary arteries and cause ischemic cardiomyopathy[4]. Behçet's disease may cause secondary amyloidosis[5], which is one of the HCM mimics, however, it has different features by echocardiography and CMR. Also, while searching for a link between the two conditions, we found a potential genetic association between Behçet's disease and HCM that could involve HLA mutations. Recent studies suggest HLA variants in HCM patients[6], while it is well-documented that HLA-B51 increases the risk of BD[7]. In this case, according to the American Heart Association guidelines[8], genetic testing for the common pathogenic genes for HCM in addition to the HLA genes - in this specific case - should be offered to the patient, however, it wasn't done due to financial limitations. Furthermore, additional research is needed to explore whether shared genetic or inflammatory pathways contribute to this rare co-occurrence.

Another important observation in this patient is the presence of significant LVOTO (provocative gradient = 98 mmHg) disproportionate to a mild increase in left

ventricular wall thickness (1.5 cm by CMR)[9]. While increased wall thickness has been known as the most common factor responsible for LVOTO, there are other important factors, including mitral valve abnormalities, such as anterior MV leaflet length, abnormal chordal attachment, and papillary muscle abnormalities. This was described in a study by Parag Patel et al. on 121 patients who had significant LVOTO without significant LV hypertrophy[10]. In our case, the presence of an elongated anterior mitral leaflet (32 mm) with a longer coaptation length (8 mm) likely contributed to a significant LVOTO. Knowing the mechanism of LVOTO is crucial in decision-making regarding surgical intervention because, in this category of patients, additional procedures on MV and papillary muscles will be needed.

Conclusion

The potential relationship between Behçet's disease and hypertrophic cardiomyopathy remains unclear. While this coexistence may be purely coincidental, the absence of previous reports on this association raises the possibility of an underlying pathophysiological link that has yet to be explored. Given the rarity of this combination, further research is warranted to investigate whether Behçet's disease may contribute to or influence the development of hypertrophic obstructive cardiomyopathy. We hope this case serves as a starting point to encourage deeper exploration into this intriguing and potentially significant connection.

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