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CASE REPORT

When what appears to be mitral stenosis is not: Diagnostic and therapeutic implications☆

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Abstract The authors report the case of a 53-year-old man, with a long-standing history of mild mitral stenosis, admitted for worsening fatigue. Transthoracic echocardiography (limited by poor image quality) showed mitral annular calcification, leaflets that were difficult to visualize and an estimated mitral valve area of 1.8 cm^2 by the pressure half-time method. However, elevated mean transmitral and right ventricle/right atrium gradients were identified (39 and 117 mmHg, respectively). This puzzling discrepancy in the echocardiographic findings prompted investigation by transesophageal echocardiography, which revealed an echogenic structure adjacent to the mitral annulus, causing severe obstruction (effective orifice area 0.7 cm^2). The suspicion of supravalvular mitral ring was confirmed during surgery. Following ring resection and mitral valve replacement there was significant improvement in the patient's clinical condition and normalization of the left atrium/left ventricle gradient.

Supravalvular mitral ring is an unusual cause of congenital mitral stenosis, characterized by an abnormal ridge of connective tissue on the atrial side of the mitral valve, which often obstructs mitral valve inflow. Few cases have been reported, most of them in children with concomitant congenital abnormalities. Diagnosis of a supravalvular mitral ring is challenging, since it is very difficult to visualize in most diagnostic tests. It was the combination of clinical and various echocardiographic findings that led us to suspect this very rare condition, enabling appropriate treatment, with excellent long-term results.

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PALAVRAS-CHAVE
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Quando parece estenose mitral mas não é – implicações diagnósticas e terapêuticas

Resumo Os autores reportam o caso de um doente de 53 anos, com diagnóstico prévio de estenose mitral ligeira, admitido por agravamento do cansaço. O ecocardiograma (Eco) transtorácico, limitado pela janela acústica, mostrava válvula mitral com ecogenicidade aumentada a nível do anel, deficiente observação dos folhetos e área valvular estimada de 1,8 cm^2 por tempo de hemipressão. No entanto, a identificação de elevados gradientes transmitral médio e ventrículo direito/aurícula direita (atingindo respetivamente 39 e 117 mmHg) intrigou os autores. No Eco transesofágico foi observada estrutura hiperecogénica sobre o anel mitral a condicionar obstrução grave (área do orifício efetivo de 0,7 cm^2), o que levantou a suspeita de anel supramitral. Esta foi confirmada durante a cirurgia. Após a ressecção do anel e implantação de prótese mecânica, verificou-se uma franca melhoria clínica e normalização do gradiente aurícula esquerda (AE)/ventrículo esquerdo (VE).

O anel supramitral é uma causa invulgar de estenose mitral congénita, caracterizado pela presença de uma membrana fibrosa adjacente à face auricular da válvula mitral. Existem poucos casos reportados na literatura, sendo a maioria descritos em idade pediátrica e em associação a outras anomalias congénitas. O diagnóstico é desafiante, dado que o anel raramente é visualizado nos exames complementares. Uma elevada suspeita clínica e a integração dos vários achados ecocardiográficos são aspetos fundamentais para a sua identificação, permitindo o tratamento adequado com bons resultados a longo prazo.

Introduction

Rheumatic disease is still the most common cause of mitral stenosis, particularly in developing countries,[1] and congenital etiology is rare.

Supravalvular mitral ring, also called supramitral ring or supramitral membrane,[1] is a rare form of congenital mitral stenosis. It is characterized by the presence of a ridge of connective tissue on the atrial side of the mitral valve, often attached to the valve annulus and/or leaflets,[2,3] and may obstruct flow to the left atrium.

Supravalvular mitral ring was first described by Fisher in 1902,[4] and fewer than 100 cases had been reported by 2002.[5] The largest series to date included only 25 patients over a 20-year period,[6] and there are no data on its actual incidence or predisposition by gender or race.[1–3] In 90% of cases, it is associated with other congenital malformations, particularly Shone's complex, and/or mitral valve anomalies.[1,2] Isolated occurrence, first described by Chung et al. in 1974,[7] is rare.

Case report

A 53-year-old man had a history of bilateral vision loss due to juvenile glaucoma, intrinsic asthma, and suspected pulmonary sarcoidosis (under corticosteroid therapy). He had been previously followed in cardiology consultations for ''a murmur since childhood'', and had been diagnosed with mild mitral stenosis based on a valve area of 1.8 cm^2 estimated by the pressure half-time method.

In 2012, he was admitted to the emergency department for worsening fatigue and dyspnea. On physical examination, he was dyspneic, with mild hypoxemia at rest (PO_2 60 mmHg), and hemodynamically stable. Cardiac auscultation revealed regular rhythm with a harsh mid-diastolic murmur, particularly audible over the apex, with presystolic accentuation, and pulmonary auscultation revealed rales in the left lung base and diffuse wheezing; there were no signs of right heart failure. Diagnostic exams included: electrocardiography showing sinus rhythm, heart rate (HR) of 80 bpm, and criteria for bi-atrial dilatation and partial right bundle branch block; laboratory tests showing elevated inflammatory markers; and chest X-ray showing increased cardiothoracic ratio and straightening of the left heart border, bilateral hilar enlargement, cephalization of the pulmonary vasculature and a reticulonodular/micronodular interstitial pattern, particularly in both lower pulmonary fields (similar to previous exams). The patient was admitted to the pneumology department with a diagnosis of respiratory infection.

Antibiotic therapy was begun but the clinical setting persisted, and so transthoracic echocardiography (TTE) was performed, which, although limited by poor image quality, revealed no left ventricular (LV) dilatation and good global and segmental systolic function; mild left atrial (LA) dilatation (area in apical 4-chamber view: 22.8 cm^2); mitral valve leaflets that were difficult to visualize but with apparently normal opening (Figure 1); a convergence zone on the atrial side of the mitral annulus plane that was difficult to visualize due to enhanced echogenicity; transmitral flow with peak diastolic gradient of 41 mmHg and mean of 21 mmHg (HR 104 bpm); valve area of 1.8 cm^2 estimated by the pressure half-time method, with no mitral regurgitation; right ventricle/right atrium (RV/RA) systolic gradient of 117 mmHg, excluding pulmonary stenosis (Figure 2); and right chambers of normal dimensions and contractility.