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CASE REPORT

Facing the challenges of ventricular hypertrophy: The eyes don't lie

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Abstract We describe the case of a 47-year-old man with new-onset heart failure who was found to have severe biventricular wall thickening. We present comprehensive data from invasive and non-invasive multimodality imaging, genetic and histologic tests, and briefly describe their importance in the final diagnosis.

To our knowledge, this is the first case of the Portuguese variant of familial amyloid polyneuropathy presenting with heart failure in the fifth decade of life.

This is an unusual case report, but also an illustration of how to approach any patient with suspected infiltrative cardiomyopathy.

PALAVRAS-CHAVE
Insuficiência cardíaca;
Hipertrofia ventricular;
Cardiomiopatia infiltrativa;
Polineuropatia amiloidótica familiar;
Ecocardiografia

Resolver os desafios da hipertrofia ventricular: os olhos não mentem?

Resumo Descrevemos o caso de um doente do sexo masculino de 47 anos, com insuficiência cardíaca de novo, que apresentava marcado aumento da espessura da parede de ambos os ventrículos. Apresentamos dados dos exames de imagem, estudo genético e análise histológica que nos guiaram no diagnóstico, explicando o nosso precurso pelos diagnósticos diferenciais e a interpretação dos diferentes testes.

Tanto quanto sabemos, é o primeiro caso descrito de um doente com a variante portuguesa da polineuropatia amiloidótica familiar que se apresenta com insuficiência cardíaca na quinta década de vida.

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Este é um caso clínico invulgar, mas também um exemplo ilustrativo da abordagem de um doente com suspeita de cardiomiopatia infiltrativa.

Case report

Increased ventricular wall thickness can be a physiologic response to pressure overload or to exercise, but it can also be pathologic, resulting from myocyte hypertrophy or deposition of an abnormal substance. Finding its exact cause can be challenging, but is crucial in determining the appropriate management.

We describe the case of a 47-year-old Caucasian man complaining of tiredness and dyspnea on moderate exertion lasting several months, progressively worsening and associated with orthopnea.

A history of erectile dysfunction, night sweats and paresthesias in both hands and feet was also reported. He had also experienced bilateral calf pain during the previous month, with no specific relieving or aggravating factors, and had noted blurred vision in the left eye for the previous six months.

He denied chest pain, fever, muscular weakness or other symptoms. His medical history was positive for type 2 diabetes and his family history was unremarkable.

On physical examination, bilateral rales and hypoesthesia of the hands and feet were noted. The chest X-ray revealed an increased cardiothoracic index and arterial blood gas analysis showed mild hypoxemia. Blood analysis revealed increased proBNP (1781 pg/ml), without anemia or renal impairment. The electrocardiogram showed normal sinus rhythm, QS waves in the inferior leads, poor R progression in the precordial leads and T-wave inversion in I and aVL (Figure 1).

The transthoracic echocardiogram (Figure 2) showed severe biventricular concentric thickening with normal ejection fraction. Diastolic assessment indicated restrictive physiology; mild pericardial effusion was also noted.

At this point, our findings suggested an infiltrative cardiomyopathy, such as amyloidosis, Fabry disease or glycogen storage disease.[1,2] The neuropathy and visual changes could fit with a systemic disease. Interestingly, the ECG did not show decreased voltages, which is common in familial amyloidosis.

Hypertrophic cardiomyopathy was another hypothesis. Against it was the ECG showing poor R-wave progression and echocardiographic findings suggestive of an infiltrative disease, with symmetric biventricular wall thickening and pericardial effusion. We excluded hypertensive heart disease, since the patient did not have hypertension. Blood analysis and thoracic computed tomography showed no changes suggestive of hemochromatosis or sarcoidosis. Thyroid disease and HIV, HCV or HBV infection were also excluded.

Cardiac magnetic resonance (CMR) showed marked concentric left ventricular (LV) hypertrophy, mild right ventricular (RV) hypertrophy and normal ejection fraction. There was biatrial enlargement, thickening of the interatrial septum and mild pericardial effusion (Figure 3). Short-tau inversion-recovery (STIR) sequences showed no myocardial edema or inflammation. In late gadolinium enhancement study, there was difficulty in setting the optimal inversion time to correctly null the myocardium, with a relatively dark blood pool and diffuse subendocardial enhancement of the interatrial septum and the LV and RV walls (Figure 4). The diffuse subendocardial late gadolinium enhancement, coupled with abnormal myocardial and blood-pool contrast kinetics, was most consistent with cardiac amyloidosis. The fact that the optimal inversion time that would null the normal myocardium could not be found is an indirect sign suggesting amyloidosis, because if abnormal fibrils are widespread in the intercellular space, the gadolinium may not be taken up by healthy myocardium.[3]

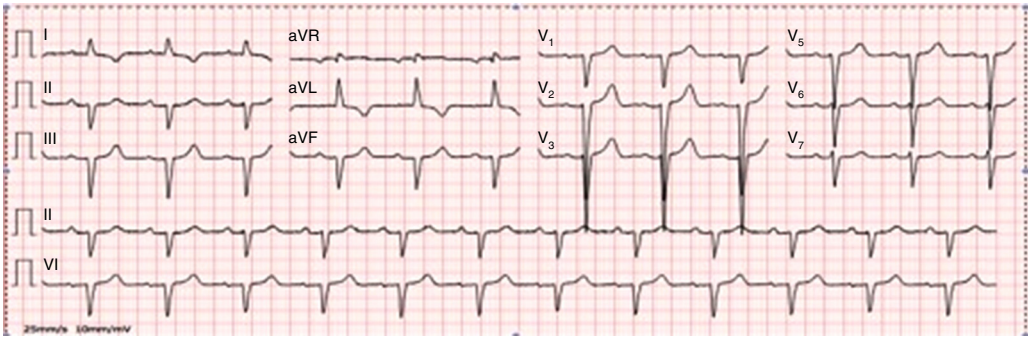

Figure 1 12-lead electrocardiogram depicting poor R-wave progression in the precordial leads and a pseudo-infarct pattern in the inferior leads.