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ORIGINAL ARTICLE

Cardiovascular profile in myotonic dystrophy type 1: Analysis of a case series in a specialized center☆

Lilian Gomes[a,*], **Telmo Pereira**[b,c], **Luís Martins**[a]

[a] *Hospital de São Sebastião, Santa Maria da Feira, Portugal*
[b] *Departamento de Cardiopneumologia, Escola Superior de Tecnologia da Saúde de Coimbra, Coimbra, Portugal*
[c] *Departamento de Cardiopneumologia, Universidade Metodista de Angola, Luanda, Angola*

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KEYWORDS
Myotonic dystrophy;
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Abstract
Introduction: Myotonic dystrophy is a multisystem disease associated with cardiac abnormalities that are responsible for high morbidity and mortality. It commonly affects conduction tissue, resulting in changes in heart rate that tend to progress with age.
Objective: The aim of the study was to assess overall cardiovascular risk and the risk of arrhythmias in patients with myotonic dystrophy type 1 (DM-1) and to correlate them with genetic study (CTG expansion size).
Methods: This retrospective study included 31 DM-1 patients referred to the cardiology department of Centro Hospitalar Entre Douro e Vouga by the neurology department for screening for heart disease. Patients' medical records were consulted for the diagnostic tests performed in the diagnostic cardiology consultation: electrocardiogram (ECG), high-resolution ECG, heart rate variability (HRV), Holter 24-hour ambulatory ECG and transthoracic echocardiogram (TTE); results of genetic testing were also consulted.
Results: Of 31 patients studied, 38% had first-degree atrioventricular block (AVB) and 51% had intraventricular conduction disturbances (62% had late potentials). TTE revealed no structural heart disease. Rare supraventricular and ventricular ectopic beats were the most common arrhythmias on 24-hour Holter monitoring. The sample showed lower HRV, reflecting vagal dysfunction. Patients with larger CTG expansions had more cardiac abnormalities.

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* Corresponding author.
E-mail addresses: liliangomes@sapo.pt, liliangomes@ufp.edu.com (L. Gomes).

Conclusions: Patients with DM-1 had arrhythmic events, with AVB and more significantly intraventricular block, although none had malignant arrhythmias or structural heart disease. No patient died. Patients with larger CTG expansions had greater involvement of cardiac conduction tissue.

PALAVRAS-CHAVE
Distrofia muscular miotónica;
Miocardiopatia;
Disritmias

Perfil cardiovascular na distrofia muscular miotónica tipo 1: estudo de uma série de casos seguida num centro especializado

Resumo
Introdução: A distrofia miotónica é uma doença multissistémica, que se associa a alterações cardíacas que são responsáveis por elevada taxa de morbilidade e mortalidade. O atingimento do tecido de condução é frequente, traduzindo-se em alterações no ritmo cardíaco, que tendem a progredir com a idade.
Objetivo: O presente trabalho propõe-se avaliar o perfil cardiovascular global, bem como o risco de arritmias cardíacas, em indivíduos com distrofia miotónica de steinert tipo 1 (DMS1), correlacionando os diversos achados com o estudo genético (tamanho da expansão CTG) comparando possíveis diferenças entre géneros.
Métodos: Estudo retrospetivo que incluiu 31 pacientes DMS1 referenciados ao serviço de cardiologia do Centro Hospitalar Entre Douro e Vouga pelo serviço de neurologia para rastreio de doença cardíaca. Todos os doentes realizaram uma consulta clínica e foram submetidos a exames complementares de diagnóstico, nomeadamente eletrocardiograma (ECG), eletrocardiograma de alta resolução (ECGAR), variabilidade da frequência cardíaca (VFC), monitorização eletrocardiográfica ambulatória contínua (MEAC-Holter) e ecocardiograma transtorácico (ETT). Foi ainda consultado o exame genético previamente efetuado.
Resultados: Dos 31 doentes estudados, 38% apresentaram BAV 1.º grau e 51% apresentaram perturbação da condução intraventricular no ECG; 62% dos doentes apresentaram potenciais ventriculares tardios; não se verificou doença cardíaca no ETT; as extrassístoles supraventriculares e ventriculares raras foram as arritmias mais frequentes no Holter de 24 h; a amostra apresentou valores diminuídos para a VFC, o que reflete a presença de disfunção vagal. Os doentes com maiores expansões CTG apresentaram maior número de anormalidades cardíacas.
Conclusões: Pacientes com DMS1 apresentam alterações cardíacas, embora a gravidade das alterações documentadas esteja aquém do sugerido na literatura. Este aspeto poderá refletir a qualidade do seguimento clínico oferecido aos doentes por um centro com uma consulta especializada.

Introduction

Myotonic dystrophy type 1 (DM-1), also known as Steinert disease, is a genetic disease caused by abnormal expansion of the cytosine-thymine-guanine (CTG) triplet in the *DMPK* (dystrophia myotonica-protein kinase) gene on chromosome 19, which is mainly expressed in skeletal and cardiac muscle.[1] DM-1 is the commonest form of muscular dystrophy in adults, with a prevalence of 2–14:100 000 individuals and an incidence of 1:8000.[2–5] It is a hereditary multisystemic disease with autosomal dominant transmission and incomplete penetrance.

Although the main clinical manifestations of DM-1 are myotony and muscle weakness secondary to involvement of skeletal muscle, other organs and systems may be affected, particularly the cardiovascular and respiratory systems.[6] The reduced life expectancy of these patients is largely due to high mortality resulting from respiratory infections, cancer and sudden cardiac death (SCD).[5]

Heart disease is common in DM-1, although its actual prevalence is difficult to estimate. There is evidence that conduction disturbances are the most frequent cardiac manifestation, but other complications have been reported, including tachyarrhythmias.[6,7] Some findings also indicate that progressive muscular impairment is associated with worsening arrhythmic events, the prevalence of which can reach 80% of cases.[8] Arrhythmias are the second leading cause of death in DM-1 after pneumonia, causing up to 30% of deaths. Most patients (over 60%) who die from cardiac cause suffer SCD.[9]