East African Scholars Journal of Medicine and Surgery
Abbreviated Key Title: EAS J Med Surg
ISSN: 2663-1857 (Print) & ISSN: 2663-7332 (Online) |
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-1 | Issue-6 | Nov-Dec-2019 | DOI: 10.36349/EASJMS.2019.v01i06.002

Letter to the Editor

CTG-Repeat Size is Crucial for Assessing Cardiac Disease in Myotonic Dystrophy Type 1 by MRI

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Keywords: multisystem disease, phenotype, genotype, cardiac, heart failure, arrhythmias.

In a recent article, Chmielewski *et al*., reported about cardiac involvement in 57 patients with myotonic dystrophy type 1 (MD1) as assessed by cardiac MRI (cMRI) (Chmielewski, L., *et al*.,2019). Non-ischemic late gadolinium enhancement (LGE) was the only independent predictor of atrial fibrillation (AFIB) or atrial flutter (AFLU), known to predict cardiac events in MD1 patients (Chmielewski, L.,*et al*.,2019). We have the following concerns and comments.

We do not agree with the statement that cardiac involvement in MD1 only includes conduction defects and arrhythmias (Chmielewski, L., *et al*.,2019). Cardiac involvement in MD1 also includes various types of cardiomyopathy, such as hypertrophic cardiomyopathy (O'Cochlain, D.F.,*et al*.,2004; Nakada, T., & Yonesaka, S. 1999), dilative cardiomyopathy(Papa, A.A.,*et al*.,2018), Takotsubo syndrome (TTS) (Finsterer. J.,*et al*.,2014), and left ventricular hypertrabeculation (LVHT), also known as noncompaction(Finsterer, J. 2009; Sá, M.I.,*et al*.,2007).

Type and degree of cardiac involvement may correlate with the size of the CTG-expansion (Melacini, P.,*et al*.,1995). Thus, it is crucial to know the CTG-repeat lengths in each of the included patients and to correlate cMRI parameters with the CTG-repeat size. Knowing CTG-repeat sizes is particularly crucial in the light of a positive correlation between CTG-repeat length and occurrence of ventricular arrhythmias (Melacini, P.,*et al*.,1995).

It could be also helpful to compare echocardiographic with cardiac MRI findings, since discrepancies may occur between both techniques (Xu, Y.,*et al*.,2018).

In the skeletal muscle, MD1 not only manifests as weakness, myotonia, and pain, but also with wasting, hypotonia, cramps, fatigue, and exercise intolerance (Laberge, L.,*et al*.,2009).

To exclude cardiac disease other than that related to MD1, it is crucial to be informed about the results of coronary angiography in the 57 included patients and about stress tests. We also should know the cardiovascular risk factor profile of these patients, such as smoking, diabetes, arterial hypertension, and hyperlipidemia. Important is also to report the medication of each included patient, as it may strongly influence the phenotype. Particularly we should know the cardiac medication as it may influence the cMRI findings. We should also know if there was cardiac disease in any of the first-degree relatives not carrying the CTG-repeat expansion.

Quick Response Code

Journal homepage:
http://www.easpublisher.com/easms/

Article History
Received: 22.10.2019
Accepted: 05.11.2019
Published: 15.11.2019

Cardiac involvement in MD1 also manifests as LVHT (Finsterer, J. 2009; Sá, M.I.,*et al*.,2007) .

Since LVHT is frequent in MD1 patients (Sá, M.I.,*et al*.,2007), we can expect that at least one or two of the included patients had LVHT. Echocardiography and cMRI should be revised for LVHT. Particularly panels a and b of figure 1 suggest LVHT, which is most frequently located in the apex and lateral wall of the left ventricle.

Recognising LVHT is crucial as it is associated with cardioembolism, heart failure, and ventricular arrhythmias, including sudden cardiac death (SCD). LVHT thus determines the outcome, and those having LVHT require particular surveillance, monitoring, and treatment.

Overall, this interesting study could be more meaningful if CTG-repeat lengths were provided, if CTG-repeat lengths were correlated with cMRI parameters, if echocardiography and cMRIs were revised for all types of cardiomyopathy, including LVHT, if the results of coronary angiography were provided, if the current medication was reported, and if the genetic and cardiac findings in first-degree relatives were mentioned.

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