Autoimmune/Chronic Inflammatory Disorders and Heart Disease

Cardiac magnetic resonance findings in patients with type 1 myotonic dystrophy

Leali M.[1]; Aimo A.[2]; Ricci G.[3]; Vergaro G.[2]; Todiere G.[2]; Grigoratos C.[2]; Aquaro GD.[2]; Siciliano G.[3]; Emdin M.[1]; Passino C.[1]; Barison A.[2]

[1]Sant"Anna School of Advanced Studies, Pisa, Italy
[2]Fondazione Toscana Gabriele Monasterio, Pisa, Italy
[3]University of Pisa, Department of Clinical and Experimental Medicine, Pisa, Italy

Funding Acknowledgements: Type of funding sources: None.

Background: Heart disease is a major determinant of prognosis in type 1 myotonic dystrophy (DM1), second only to respiratory complications. Cardiac imaging, possibly including cardiac magnetic resonance (CMR), is recommended in patients with DM1. However, limited information is available on CMR findings and their prognostic significance in DM1.

Methods: We identified all patients with DM1 evaluated from 2009 to 2020 in a CMR laboratory with an established collaboration with a Neuromuscular Disorder Unit.

Results: Thirty-four patients were retrieved (21 males, aged 45 ± 12). At the time of CMR examination, 97% had neuromuscular symptoms (mean duration 16 ± 13 years), 12 (35%) presented with atrioventricular block (n = 11 1st degree, n = 1 2nd degree type 1), 15 (44%) with intraventricular conduction disturbances (n = 5 left bundle branch block, n = 5 right bundle branch block, n = 3 left anterior fascicular block, n = 2 other non-specific intraventricular conduction delay), 4 (12%) with atrial fibrillation or flutter. No patient had a device. At CMR, 5 (15%) patients had left ventricular (LV) systolic dysfunction (LV ejection fraction [LVEF] <50%) and 5 (15%) a depressed right ventricular (RV) function (RVEF <50%). Compared to age- and sex-specific reference values for our laboratory, 12 (35%) patients showed a decreased LV end-diastolic volume index (LVEDVi), 7 (21%) a decreased LV mass index (LVMi), and 29 (83%) a decreased LVMi/LVEDVi ratio. Nine (26%) patients had mid-wall late gadolinium enhancement (LGE, mean extent 4.5 ± 2.0% of LVM; n = 8 septal, n = 4 inferolateral, n = 2 inferior, n = 1 anterolateral), and 14 (40%) some areas of fatty infiltration (n = 9 involving the LV, n = 13 the RV). Native T1 in the interventricular septum (1,041 ± 53 ms) approached the upper reference limit (1,089 ms), and the extracellular volume was slightly increased (33 ± 2%, reference values <30%). Over a median follow-up of 3.3 years (interquartile interval 1.6-4.7), 2 (6%) patients died, one for infectious and respiratory complications and the other for unknown causes, 5 (15%) patients underwent pacemaker implantation for conduction disturbances, and 4 (12%) had a documentation of high-risk (Lown class ≥4) ventricular ectopic beats (VEBs). Among all CMR variables collected, higher values of LVMi/LVEDVi ratio emerged as univariate predictor of all-cause death (p = 0.044). At logistic regression analysis, anteroseptal wall thickness was associated with the need for pacemaker implantation (p = 0.028), while LGE mass was associated with high-risk VEBs (p = 0.026).

Conclusions: Patients with DM1 display several structural and functional cardiac abnormalities, with variable degrees of cardiac muscle hypotrophy, fibrosis and fatty infiltration. The possibility to predict the need for pacemaker implantation, ventricular arrhythmias and all-cause or cardiovascular mortality should be verified in larger cohorts.

Abstract Figure. CMR findings (mean ± sd) and follow-up.