

Cardiovascular profile in pediatric Myotonic Dystrophy type 1: are our patients at risk?

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Introduction

Myotonic dystrophy-1(DM1) is a multisystem disease associated with cardiac conduction and structural abnormalities. Our objective is to describe cardiovascular evaluation in our paediatric population, both echocardiography and EKG measurements, and to correlate these features with CTG expansion size.

Methods

This retrospective study included 27 paediatric DM1 patients. We reviewed clinical features, transthoracic echocardiography images, EKG and cardiac 24h-Holter. We analysed (1) echocardiographic measurements with LVEF, LVED-LVES diameters, systolic and diastolic echo analysis (2) EKG features: rhythm, PR interval, QRS interval, QT and QTc. (3) cardiac 24h-Holter monitoring features: HR variability, mean SDNN, maximum RR, and qualitative findings, (4) and the CTG expansion size.

Results

No significant differences by gender (m55.56%; f44.44%); age was 5-19yo (m14.1). Only two symptomatic patients explaining dizziness, incomplete right branch block was detected in these cases. No sudden death was detected and no pacemaker or ICD were implanted. EKG and 24h-Holter findings: mean HR in the basal 12 leads EKG was 82bpm (40-130). The most frequent feature was an incomplete RBBB (26%), and complete RBBB (11%). Five patients (18.5%, older patients) had abnormal 24h-Holter: 2 had second degree AV block Mobitz I, 1 had first degree AV-block, 1 had asymptomatic monomorphic PVC, and the last one had severe sinus bradycardia during the deep sleeping (min HR of 23bpm). Mean SDNN was obtained in 22/27 patients. Analysis by age group showed a decreasing mean SDNN with age, so younger patients had a larger value SDNN than the older (group <10yo mSDNN 170.7ms±39, and group >15yo SDNN of 147.3ms±37), with a significant variation between three age groups 5-10, 10-15, 15-20yo. No patients with structural heart disease. LVES and LVED diameters were normal, 11% had slightly reduced LVEF (52-55%) with no relation with conduction disorders. No significant abnormalities in diastolic echo analysis. We didn't find significant relation between neurological condition, EKG/echo features and the CTG expansion (but only had an exact size in 6/27 patients, 123-1667 CTG repeats).

Conclusions

(1) Pediatric patients showed mild conduction abnormalities that could be the precursors for high degree heart block, ventricular arrhythmias and sudden death in the DM1-adult population. (2) In our DM1-pediatric series, SDNN analysis showed that the lower values of SDNN the older DM1 patients. So, it is an abnormal SDNN behaviour that it could have the basis on para/sympathetic system, cardiac autonomic modulation and deterioration in baroreceptor function. (3) Echocardiographic structural abnormalities were described in DM1 population, but it had been detected in our paediatric series. (4) We cannot relate CTG repeats with the heart condition.