Cardiac impairment in Duchenne Muscular Dystrophy: A single-center retrospective experience

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ICNC 2022
17th INTERNATIONAL CHILD NEUROLOGY CONGRESS
ANTALYA, TURKEY | OCTOBER 3-7, 2022

INTRODUCTION

- Cardiomyopathy is the leading cause of mortality in boys with Duchenne Muscular Dystrophy (DMD) and myocardial involvement of DMD remains under-recognized and undertreated.
- Early diagnosis is critical for timely initiation of cardioprotective therapies. Screening of all DMD boys by electrocardiogram (ECG) and transthoracic echocardiography (TTE) is recommended annually after the age of 10 years. Myocardial fibrosis can be detected by cardiac magnetic resonance imaging (cMRI), is an early finding, even if conventional TTE is normal.
- In the current study, we aimed to evaluate cardiac functions in patients with DMD and to determine the degree of cardiac impairment.

MATERIALS & METHODS

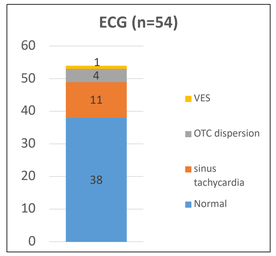
- A retrospective chart review of 54 DMD boys followed at Ankara City Hospital in the period of 2019-2022 was performed.
- Patients with at least 6 months of follow-up and one cardiac evaluation were included.
- The following clinical variables were collected: demographic features, family history of DMD, ambulatory status, genetic mutations, corticosteroid therapy, and cardiac assessments (TTE, ECG, and cMRI). Presence of late-gadolinium enhancement on cMRI was considered as LGE positivity (LGE+) and revealed myocardial fibrosis.

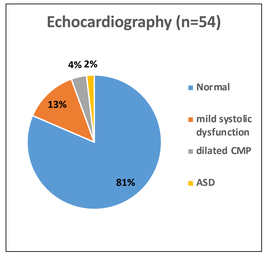
RESULTS

- Baseline clinical characteristics of the DMD boys are shown in Table 1.
- At last cardiac evaluation, 25 patients (46%) were >10 years of age.
- ECG revealed baseline tachycardia in 11 (20%), prolonged QTC interval in 4 (7%), and ventricular extrasystole in one patient.
- Abnormal findings were detected by TTE in 18% (n=10). Of those, 7 had mild systolic dysfunction, two had dilated cardiomyopathy and one had atrial septal defect. In the TTE, mean ejection fraction (EF), shortening fraction (SF), and left ventricular end-diastolic diameter (LVEDd) was 67% (48-74), 36% (24-42), and 32 mm (20-50), respectively.
- Cardiac MRI was performed in 15 boys aged between 7.5-17.5 years (mean 12.4 years). CMRI-derived LVEF was ≤55% in 40% and cMRI showed the presence of left ventricular wall hypokinesia in 25%. Focal LGE was detected in 12 of 15 (80%) DMD boys. Echocardiography was found to be normal in 8 out of 15 patients who underwent cardiac MRI. Abnormal cMRI findings lead to the initiation of prophylactic cardioprotective therapies in 6 patients. During follow-up period, one patient died from heart failure at 17.5 years of age.

Table 1. Clinical characteristics of DMD boys (n=54)

Age at diagnosis, mean (min-max), mo	38 (1-88)
Age at last visit, mean (min-max), yr	7.6 (1-17.7)
Follow-up, mean (min-max), mo	38 (6-76)
Type of mutation	
Deletion, n, (%)	43, (80)
Duplication, n, (%)	5, (9)
Point mutation, n, (%)	6, (11)
Ambulatory status	
Ambulant, n (%)	39 (72%)
Non-ambulant, n (%)	15 (28%)
Age at ambulation loss, median, years	10.6 (7.5-13)
Corticosteroid treatment, n (%)	36 (66%)
Cardioprotective therapy, n (%)	9 (17%)





Cardiac MRI findings (n=15)	
LVEF <55%	6/15 (40%)
Systolic dysfunction	8/15 (53%)
Diastolic dysfunction	9/15 (60%)
LGE	12/15 (80%)

CONCLUSIONS

- Current study shows that despite having normal baseline systolic function in TTE, variable degrees of systolic/diastolic dysfunction or focal myocardial fibrosis were detected by cMRI in almost half of our patients.
- In recent years, cardiac MRI with the assessment of late gadolinium enhancement has replaced echocardiography as the imaging modality of choice in the DMD Care Considerations Working Group recommendations from 2018.
- Early detection of subclinical cardiac dysfunction with sensitive imaging modalities in DMD is critical for early intervention.

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