Decreased global strains of the left ventricle in asymptomatic female carriers for duchenne muscular dystrophy gene using feature-tracking: a prospective study

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Background: Duchenne muscular dystrophy (DMD) is an X-linked recessive disease manifested in males predominantly by skeletal muscle wasting but also dilated myocardium. Heterozygous female carriers often have normal echocardiographic findings. Nevertheless, untreated could develop in 4th and 5th decade of life to cardiomyopathy, probably due to progressive myocardial fibrosis. CMR feature tracking (FT) is an innovative technique that measures the myocardial wall deformation and could help to detect the contractile dysfunction, even in its early stages, due to its high sensitivity. Thus, it could identify asymptomatic carriers at higher risk of developing cardiomyopathy later in life.

Aim: This prospective study aimed to assess left ventricular systolic function (LV) in DMD carriers using CMR-FT.

Methodology: Global longitudinal strain (GLS), global circumferential strain (GCS) and global radial strain (GRS) were evaluated by CMR-FT. End-systolic (ESV), end-diastolic (EDV) LV volumes and ejection fraction (EF) LV were also measured using CMR-FT.

There were 37 carriers, and 20 healthy controls enrolled in the study. The measured parameters showed a Gaussian distribution and were compared using a t-test, followed by Benjamini-Hochberg correction.

Results: Groups carriers and controls did not differ in basic demographic data (age: 39.5±9.1 vs. 39.1±10.7 years). In contrast, there were found statistically significant differences in EF LV (56±5 vs. 60±3%), and all strain parameters (GLS (−19.4±2.4 vs. −22.3±2.2%), GCS (26.8±3.3 vs. 29.5±2.0%) and GRS (59.8±12.9 vs. 70.9±14.8%, all $p<0.05$)).

Conclusion: Carriers had significantly lower values of all strains compared to controls using CMR-FT, although they had preserved EF LV. CMR-FT could be beneficial for early diagnosis of the developing heart disease in asymptomatic patients with a genetic predisposition.