

Pathophysiological insights into cardiac involvement in patients with Duchenne or Becker muscular dystrophy

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Introduction: Genetic dystrophin defects may cause myocardial fibrosis from the subepicardial layer due to intramural extension, resulting in potential regional myocardial dysfunction. For the early diagnosis and proper management of cardiac involvement in patients with Duchenne or Becker muscular dystrophy (DMD/BMD), the relationship between the extent of myocardial fibrosis and wall strain should be elucidated.

Methods: Cardiac magnetic resonance imaging (CMR) with late gadolinium enhancement (LGE) and two-dimensional speckle tracking echocardiography (STE) were performed in 7 patients with DMD/BMD (median age 11.7 years, 2.8 - 20.9 years) without any symptoms of heart failure. Each AHA segment except for apical segment was scored to determine the transmural extent of LGE (0=no LGE, 1=1-25%, 2=26-50%, 3=51-75%, and 4=76-100%). The relationship between the LGE score and STE strain (peak radial [RS] and peak circumferential strain [CS]) in a total of 84 myocardial segments was studied.

Results: Forty-three segments in four patients were LGE positive with predominant subepicardial involvement. The number of segments in each LGE score was n=6 (LGE score of 1), n=8 (2), n=12 (3), and n=17 (4). The RS was significantly decreased in the segments with LGE score >1 (Figure). The CS was not significantly different among the LGE score groups. ROC curve analysis revealed the cut-off RS value to predict an LGE score >2 was 22% (AUC 0.79), and the cut-off value for an LGE score of 4 was 12% (AUC 0.74).

Conclusions: Our data suggested that fibrosis of the subepicardial layer results in decreased RS, which can be detected by STE. Severely reduced RS may suggest intramural extension, which should be confirmed by CMR-LGE. Further investigation is required for detailed pathophysiological insights into cardiac involvement in patients with DMD/BMD.

