Beyond Ejection Fraction: Analysis of Cardiac Strain for Early Detection of Myocardial Dysfunction in Duchenne Muscular Dystrophy

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Short Editorial related to the article: Regional Left Ventricular Myocardial Strain Alterations in Patients with Duchenne Muscular Dystrophy and Correlation with Electrocardiographic and Motor Function Alterations

Duchenne muscular dystrophy (DMD) is a progressive, degenerative, X-linked genetic neuromuscular disorder that affects motor, respiratory, and cardiac function.[1,2] DMD is the most severe presentation of neuromuscular diseases related to dystrophin mutations.[1,2] Clinically, it manifests as progressive impairment of motor function in boys, with wheelchair dependence before the age of 12.[3]

Advances in multidisciplinary care and ventilatory management have improved survival, making cardiac complications a leading cause of mortality in patients with Duchenne muscular dystrophy (DMD).[4-6] However, to date, no temporal correlation has been established between cardiac and skeletal muscle impairment, making the screening for cardiac dysfunction challenging, since the classic symptoms of heart failure are masked by progressive motor limitation.

In this context, the study by Costa et al.[7] makes a relevant contribution by investigating regional left ventricular (LV) myocardial strain using speckle tracking in patients with DMD and its correlation with electrocardiographic changes and motor function.

In this study,[7] a significant sample size was evaluated for a rare disease, considering the logistical limitations inherent in recruiting this population. The authors performed a correlation between changes in regional myocardial deformation and electrocardiographic and motor alterations. The role of myocardial strain assessment in these patients warrants further investigation, particularly for the early detection of cardiac dysfunction and the timely initiation of cardioprotective therapy to prevent adverse clinical outcomes, underscoring the clinical relevance of this study.[8,9]

The authors demonstrate that although left ventricular ejection fraction (LVEF) was preserved in most patients, Speckle-tracking analysis revealed a reduction in global longitudinal, circumferential, and radial myocardial deformation in 70 to 80% of cases. This finding reinforces a concept already established in the cardiology literature: LVEF is a late marker of myocardial dysfunction, and advanced cardiac imaging techniques are able to detect subclinical impairment years before its reduction.

However, the most original aspect of the study consists of the proposal of a new regional myocardial deformation index (REGLSi), calculated from the ratio of segmental strain of the inferior and inferior lateral walls of the LV – characteristically the most compromised in DMD – to the average strain of the other segments. Anatomopathological studies have demonstrated the preferential development of myocardial fibrosis in the posterobasal segments, a particular pattern distinct from that observed in other non-ischemic cardiomyopathies.[10] Thus, by analyzing segmental myocardial deformation through a continuous and reproducible index, this study demonstrates the potential of a parameter more sensitive than qualitative segmental contractility scores, which identified abnormalities in only 15% of the evaluated cohort, while 80% of patients presented segmental strain abnormalities assessed by the speckle tracking technique.

Another interesting aspect of this study was the convergence between the electrocardiographic findings – prominent R wave in V1 and pathological Q wave in V5-V6, present in the vast majority of patients – and the preferential involvement of the inferior and lower lateral wall strain. These findings corroborate the hypothesis that these alterations are related to the subendocardial distribution of dystrophin in the Purkinje fibers.[11,12] If confirmed by prospective studies correlating these findings with histological evaluation or cardiac magnetic resonance imaging, this association could suggest electrocardiographic evaluation – a low-cost and widely available examination – as an initial screening tool for selecting DMD patients who should be evaluated early by advanced echocardiographic techniques.

Conversely, this temporal dissociation, reflected by the lack of correlation between motor function and cardiac parameters, may reflect the natural history of the disease. Despite sharing the same primary genetic mutation, cardiac and skeletal muscle involvement may follow distinct and independent trajectories of functional deterioration. This observation is a relevant finding and reinforces the importance of cardiological surveillance, not only clinical, since patients present as self-limiting, but also supported by cardiac imaging exams and conducted systematically and independently from diagnosis.

Keywords

Duchenne Muscular Dystrophy; Echocardiography; Global Longitudinal Strain; Cardiomyopathies

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As with any pioneering study, some questions remain. External validation of REGLSi in multicenter cohorts, with standardization of equipment and analysis software, is an indispensable step before its incorporation into clinical practice, given the known interobserver and intervendor variability of myocardial strain measurements. The lack of data on left atrial and right ventricular strain, as well as its correlation with serum biomarkers and cardiac magnetic resonance imaging—the gold standard for myocardial fibrosis characterization—highlights important avenues for future research to further establish the diagnostic and prognostic value of this technique.

The data from this study emphasize the need to define the ideal time to initiate cardiac pharmacoprotection for patients with DMD, currently still conditioned by LVEF.[13,14] Regional deformation indices, if prospectively validated, could improve patient care by anticipating this therapeutic decision, with a potential impact on the survival of a population where cardiovascular mortality is a key factor.

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