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COMMENTARY

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A new era: Speckle tracking echocardiography and cardiomyopathies

Uğur Aksu | Mehmet Hakan Uzun

Faculty of Medicine, Department of Cardiology, Afyonkarahisar Health Sciences University, Afyon, Turkey

Correspondence
Uğur Aksu, Afyonkarahisar Sağlık Bilimleri Üniversitesi Tıp Fakültesi, Afyon,Turkey.
Email: aksuuu001@msn.com

Abstract

Limb-girdle muscular dystrophy (LGMD) is a heterogeneous inherited disorder affecting the skeletal muscle and frequently also involve the heart and in LGMD; development of dilated cardiomyopathy is common and usually the predominant feature. Arrhythmias and conduction disease can be associated with the development of cardiomyopathy.

KEYWORDS
cardiomyopathies, strain, strain rate imaging

Limb-girdle muscular dystrophy (LGMD) is a heterogeneous inherited disorder affecting the skeletal muscle and frequently also involve the heart and in LGMD; development of dilated cardiomyopathy is common and usually the predominant feature. Arrhythmias and conduction disease can be associated with the development of cardiomyopathy.[1,2]

Limb-girdle muscular dystrophy (LGMD) is first defined in 1954 as a new entity, distinct from the other dystrophies. It is classified as autosomal dominant LGMD (LGMD1 or AD-LGMD) and autosomal-recessive LGMD (LGMD2 or AR-LGMD). With a wide range of clinical and genotypic variability, LGMDs involve progressive proximal muscle weakness in common.[3]

Different subtypes are associated with varying degrees of cardiac involvement. More than 30 different subtypes are defined, with each linked to a specific gene loci.[1] Nonetheless, almost every subtype has overlapping phenotypic expressions, due to the nature of proteins which are coded, are responsible for the integrity of the sarcomere-sarcolemma-sarcoplasm-extracellular matrix complex. Difference between subtypes varies in age of onset, rate of progression, and affected muscle groups.[4]

Autosomal dominant forms represent less than 10% of the LGMD cases.[1] They usually have adult-age onset, with milder symptoms. Autosomal recessive forms are much more common, with a cumulative prevalence of 1:15,000. There are currently 23 autosomal recessive forms of LGMD subtypes in literature (LGMD2A to LGMD2W). Subtypes LGMD2I and LGMD2E have higher prevalence of cardiac involvement; patients with LGMD2A and LGMD2B have much lower and milder prevalence of cardiac involvement.[3–5]

LGMD2A, also known as Calpainopathy which is caused by Calpain 3 (CAP3N) gene mutations, is the most common form of LGMD cases. Calpain system is composed of μ-calpain, m-calpain and calpastatin. This system participates in cytoskeletal/membrane attachments, signal transduction pathways and apoptosis.[4] Calpain 3 is expressed mostly in the skeletal muscles, but a previous study has shown that cardiac muscle contains 46% of Calpain 3a mRNA as skeletal muscle.[5]

LGMD2B is caused by mutations in the Dysferlin (DYSF) gene. This protein is involved in calcium-mediated sarcolemma resealing.[1] This subgroup also has heterogeneous clinical presentations, varying from mild late-onset forms to severe functional disabilities.[6]

Although LGMD2A and LGMD2B have relatively lower prevalence of cardiac involvement, it should be noted that some patients within these subtypes may develop severe cardiac dysfunction. Therefore, developing a cost-effective scanning protocol for early changes in cardiac functions is crucial in the prevention and/or disease progress modification in such patients. Speckle-tracking echocardiography (STE) is a non-invasive imaging method, able to detect subclinical myocardial dysfunction. In this recent research, it has shown that LGMD2A patients have insignificant changes in STE measurements, while LGMD2B patients show statistically significant changes in measured values, pointing towards early subclinical myocardial fibrosis development.

The roles of STE in detecting these aforementioned changes have been a hot topic of debate in recent publications, with strong evidence of correlation between cardiac magnetic-resonance imaging (cMRI) and STE. Due to the advantages of STE over cMRI; such as cost-effectiveness and the absence intravenous contrast agent

administration, this method in screening patients under increased risk of developing cardiac dysfunction is a promising feature.

In this issue of *Journal of Clinical Ultrasound*, a new research article titled"Evaluation of Cardiomyopathy with Two-Dimensional Speckle Tracking Echocardiography in Limb-Girdle Muscular Dystrophy Type 2A and 2B" has been published. Authors have compared echocardiographic findings in 3 groups; between LGMD2A, LGMD2B and a healthy control group. The median age was 33 (22–39 interquartile range [IQR]) in the LGMD2A group, 33 (27–38 IQR) in the LGMD2B group, and 28 (25–35 IQR) in the control group. Apical 4-chamber longitudinal strain (LS), Apical 2-chamber LS, Apical 3-chamber LS, left ventricular global longitudinal strain (LVGLS)-mid-myocardial, LVGLS-endocardium, and LVGLSepicardium were lower (less negative) in the LGMD2B group compared to the control group ($p = 0.006$).

Authors have shown that, myocardial dysfunction may develop in patients with LGMD2B and this entity can be detected in early stage by using STE imaging. Strain imaging, compared with conventional echocardiographic parameters, is more sensitive in detecting early and subclinical changes in diseases, that is, myocardial infarction, hypertension, myopathies and storage diseases, which can alert the physician to take appropriate precautions before clinical myocardial dysfunction develops.[7,8] Findings being reported by the authors are consistent with current literature.

Also, current guidelines suggest using STE imaging over conventional echocardiographic parameters, due to detecting myocardial dysfunction in the early stages. In addition, STE can be applied on all four chambers. Due to routine usage of automatic tracing systems, calculated values are less dependent on the operator skill and knowledge, thus leading to a more standardized approach. In the near future, it is expected to have more applications with wider patient groups.[7,8]

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

ORCID

Uğur Aksu https://orcid.org/0000-0003-0918-5032

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