Review Article

ULTRASOUND EVALUATION OF MUSCLE ARCHITECTURE IN CHILDREN WITH MUSCULAR DYSTROPHY A COMPREHENSIVE REVIEW

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ABSTRACT

Background: Muscular dystrophies are a heterogeneous group of inherited neuromuscular disorders characterized by progressive muscle weakness and degeneration. Early diagnosis and continuous monitoring are essential for optimizing clinical management, especially in children. Muscle ultrasound has emerged as a valuable, non-invasive imaging modality for evaluating muscle architecture and pathology. This review provides a comprehensive overview of the role of ultrasound in assessing muscle architecture in pediatric muscular dystrophy, including principles of imaging, normal and pathological muscle characteristics, disease-specific ultrasound findings, quantitative techniques, advantages, limitations, and future perspectives. The growing use of ultrasound offers significant potential for early diagnosis, disease monitoring, and assessment of therapeutic response in children with muscular dystrophy.

Keywords: muscle ultrasound, muscular dystrophy, pediatric neuromuscular disorders, muscle architecture, echogenicity

INTRODUCTION

Muscular dystrophies are a group of inherited neuromuscular disorders characterized by progressive muscle weakness due to degeneration of muscle fibers and their replacement by adipose and fibrous tissue. These disorders frequently manifest in childhood and include Duchenne muscular dystrophy (DMD), Becker muscular dystrophy (BMD), limb-girdle muscular dystrophy (LGMD), congenital muscular dystrophy (CMD), and Emery–Dreifuss muscular dystrophy [1,2]. Among these, DMD is the most common and severe form, affecting approximately 1 in 3,500 live male births worldwide [3].

Early diagnosis and continuous monitoring are essential to optimize clinical management, initiate corticosteroid therapy, and evaluate emerging disease-modifying treatments such as exon-skipping and gene therapy [4]. Conventional diagnostic methods including serum creatine kinase estimation, electromyography, muscle biopsy, and genetic testing are effective but may be invasive, costly, or unsuitable for repeated follow-up in children [5]. Imaging modalities play a crucial complementary role in the evaluation of muscular dystrophy. Magnetic resonance imaging (MRI) provides detailed assessment of muscle composition but is limited by high cost, long examination times, and the need for sedation in young children [6]. In contrast, muscle ultrasound is a safe, non-invasive, portable, and cost-effective technique that allows real-time visualization of muscle architecture without radiation exposure [7].

Muscle ultrasound has demonstrated high sensitivity in detecting pathological changes such as increased echogenicity resulting from fatty infiltration and fibrosis, often before the onset of overt clinical weakness [8]. Recent advances in quantitative ultrasound analysis have further enhanced its role as a potential biomarker for disease severity and progression [9]. This review aims to summarize the current literature on ultrasound evaluation of muscle architecture in children with muscular dystrophy.

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REVIEW OF LITERATURE

Normal skeletal muscle on ultrasound appears relatively hypoechoic with fine hyperechoic linear striations representing fibroadipose septa. The muscle fibers are arranged in a parallel pattern, with clear delineation of muscle fascia and underlying bone [7]. In children, normal muscle echogenicity is lower than that of adults, and muscle thickness increases with age and growth. In muscular dystrophy, progressive muscle fiber degeneration leads to fatty infiltration and fibrosis, resulting in characteristic ultrasound changes. These include increased muscle echogenicity, loss of normal striated architecture, reduced muscle thickness, and acoustic shadowing in advanced disease [10].

Several studies have demonstrated that children with DMD exhibit markedly increased muscle echogenicity, particularly in proximal muscles such as the quadriceps and calf muscles [8,11]. Pseudohypertrophy of the gastrocnemius muscle is a common finding and correlates with fatty infiltration rather than true muscle hypertrophy. Selective involvement of certain muscles, such as early involvement of the vastus intermedius compared to relative sparing of the rectus femoris, has also been described [12].

Ultrasound findings in BMD (Becker and Limb-Girdle Muscular Dystrophy) are similar to those in DMD but less severe and more slowly progressive [13]. In LGMD, muscle involvement is typically patchy, predominantly affecting the pelvic and shoulder girdle muscles, with variable echo-

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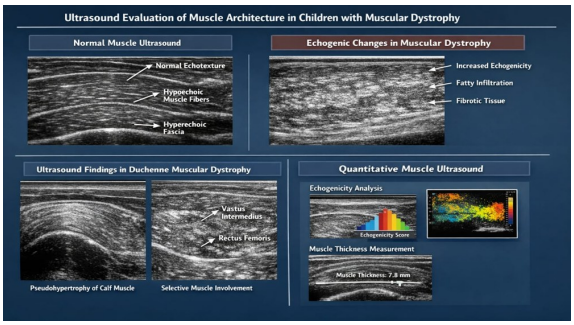
genic changes depending on the genetic subtype [14].

Quantitative techniques, including grayscale analysis and muscle thickness measurements, have been developed to improve objectivity and reproducibility. Increased grayscale values correlate with histopathological fat and fibrotic content and have been shown to correlate with functional impairment [9,15]. Quantitative ultrasound has also been proposed as an outcome measure in clinical trials.



DISCUSSION

Muscle ultrasound has emerged as a valuable imaging modality for the assessment of muscle architecture in children with muscular dystrophy. Numerous studies have demonstrated that ultrasound is sensitive in detecting increased muscle echogenicity due to fatty infiltration and fibrosis, which are hallmark pathological features of dystrophic muscle [7,10]. Importantly, these changes can be detected in early or even preclinical stages, emphasizing the role of ultrasound in early diagnosis [8].

In Duchenne muscular dystrophy, characteristic patterns of selective muscle involvement observed on ultrasound correlate well with histopathological findings and MRI-based fat fraction analysis [6,11]. Several studies have also shown significant correlations between ultrasound echogenicity and functional outcome measures such as muscle strength and ambulatory status [15,16]. This supports the use of ultrasound as a clinically meaningful tool for disease monitoring. Quantitative muscle ultrasound represents a major advancement by reducing operator dependency and enabling standardized assessment of disease severity and progression [9].

Its feasibility for repeated examinations makes it particularly suitable for pediatric patients and longitudinal follow-up. However, limitations remain, including difficulty in evaluating deep muscles and lack of universal standardization of imaging protocols [10]. Despite these limitations, emerging techniques such as ultrasound elastography and automated image analysis are expected to further enhance diagnostic accuracy and reproducibility [17]. When used in conjunction with clinical assessment and other imaging modalities, muscle ultrasound provides valuable complementary information and has the potential to become an integral component of pediatric neuromuscular evaluation.

CONCLUSIONS

Muscle ultrasound is a valuable, non-invasive imaging modality for the evaluation of muscle architecture in children with muscular dystrophy. It enables real-time visualization of structural changes such as increased echogenicity, altered muscle thickness, and loss of normal muscle architecture resulting from fatty infiltration and fibrosis. These ultrasound features closely reflect the underlying pathological processes and can often be detected at an early stage, even before the onset of significant clinical weakness.

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