

## EDITORIAL

# Magnetic resonance imaging in muscular dystrophies

Muscular dystrophies present with a broad often overlapping diagnostic spectrum which may ultimately require muscle biopsy.

Magnetic Resonance Imaging (MRI) is growing in popularity and becoming more and more followers, because its capacity to reveal characteristic findings which can address the diagnosis, aid in determining optimal biopsy sites and control therapeutic interventions.

Several papers published during the past few years have reported on the value of MRI in detecting patterns of muscle involvement in various muscular dystrophies and other inherited myopathies. Such a technique provides a high soft tissue contrast allowing excellent assessment of striated muscles concerning shape, volume (hypotrophy, hypertrophy) and tissue architecture (1-4).

Because of the lack of ionizing radiation, MRI has become a valuable imaging method in children, although sometimes sedation might be necessary. Basically, MRI is performed as a multi-sequence imaging protocol including T1-weighted (T1W) and T2-weighted (T2W) (turbo) spin echo as well as fat-suppressed (short tau inversion recovery or spectral fat suppression techniques) T2-weighted sequences (T2WFS). The image acquisition is performed in the axial plane with a slice thickness of 5-7 mm. If necessary, additional images in other anatomical planes (coronal, sagittal) can be easily acquired. MRI can be performed and rated in a standardized manner suggesting a good inter-rater and intra-rater (during follow-up) agreement.

In the early stages of Duchenne Muscular Dystrophy (DMD), MRI shows an early involvement of the gastrocnemii (5, 6). An abnormal signal in the gluteus maximus and adductor magnus, followed by involvement of the quadriceps, rectus femoris, and biceps femoris, with selective sparing of the sartorius, gracilis, semitendinosus, and semimembranosus is observed in the advanced stages (7-9).

A distinct pattern on muscle imaging characterized by prominent involvement of the gluteus maximus and medius, adductor magnus, biceps femoris long head,

semi-membranosus and vasti was also observed in individuals with Becker Muscular Dystrophy (BMD), a milder form secondary to mutations in DMD gene (10, 11).

A specific pattern of muscle fatty replacement and atrophy, particularly in upper girdle muscles have been reported in Facio-Scapulo-Humeral-Dystrophy (FSHD) patients. The most frequently affected muscles, including paucisymptomatic and severely affected patients, were trapezius, teres major and serratus anterior, in a characteristic asymmetric fashion (12).

In some forms of limb-girdle muscular dystrophies such as dysferlinopathies and anoctaminopathies, due respectively to mutations in DYSF and ANO5 genes, a predominant fatty degeneration of the gluteus minimus muscle and of the posterior segments of the thigh and calf muscles, with sparing of the gracilis muscle, was observed (13).

MRI was proved useful also in patients with oculopharyngeal muscular dystrophy (OPMD), where it revealed distal lower legs more severely fatty replaced than the thigh muscles. Soleus and long head of the biceps femoris was severely involved in all patients, whereas popliteus, gracilis and short head of biceps femoris were almost completely spared, even in advanced stages (14).

A few recent studies have reported muscle MRI findings in the most common forms of Congenital Muscular Dystrophies (CMD), those secondary to mutations in the collagen VI genes (Ullrich CMD), though no systematic studies have evaluated muscle MRI in all of the genetically recognized forms of CMD. Patients with Ullrich CMD show diffuse involvement of all the posterior and lateral muscles of the thigh with selective sparing of the sartorius, gracilis, and adductor longus, and often the rectus femoris (15-18). These signs have a significant overlap with those observed in Bethlem myopathy, a milder dominant condition allelic to Ullrich CMD.

It has been shown that muscle MRI is able to distinguish the various forms of congenital muscular dystrophy, despite a significant clinical overlap. For example,

patients with RSMD1, a condition secondary to deficiency in selenoprotein 1, who also have rigidity of the spine, early respiratory involvement, and normal or only mildly elevated CK, present a peculiar MRI muscle involvement (17, 18).

In the present issue we publish the article review of Díaz-Manera et al. on muscle MRI pattern in various forms of muscular dystrophies and the paper of Maggi et al. on the usefulness of MRI in muscle channelopathies.

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