Whole body MRI WB-MRI *MRI in the diagnosis and monitoring neuromuscular diseases*

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Muscle imaging has recently become a part of the diagnostic work-up of acquired and inherited muscle diseases. Magnetic resonance imaging (MRI) is the method of choice because it is free of radiation or side-effects, and has excellent soft tissue contrast and resolution. It allows topography of fat infiltration into muscular mass and decrease of muscular volume (T1-weighted (T1w) sequences), tissue oedema or inflammation (STIR, parametric T2) and has a potential use to provide quantitative data to assess change over time (density weighted (PDw) imaging with water–fat separation) without requiring invasive procedures such as biopsy. While proton-density weighted (PDw) imaging with water–fat separation is being investigated both preclinically and in animal models to assess the course of neuromuscular disease, the major development of muscle MRI techniques at present is focused in the impact of the technique in diagnosis of muscle disorders. T1-weighted sequences are the most useful images. Sequential MRI studies, mainly in lower limbs, have reported distinct patterns of muscle involvement of thighs and legs in various forms of inherited myopathies (*COL6, SEPN1, RYR1, LMNA, DNM2)*. More recently, whole-body MRI (WB-MRI) techniques have been developed (Quijano-Roy et al 2012) and may enhance significantly the diagnostic potential of the sequential scanning techniques in many neuromuscular disorders with more diffuse involvement. WBMRI allows visualizing in axial and frontal views muscles of the body from head to toes. It offers not only the equivalent of multiple sequential MRI studies for all body regions (head, neck, trunk, girdles, limbs, thorax, abdomen), but also frontal views. We have recently reviewed the WBMRI examinations in 117 individuals including young children, patients with severe limb contractures, and children receiving ventilatory support or with metallic implants. 38 subjects had a genetically confirmed diagnosis (*RYR1, LMNA, COL6, DNM2, GAA, TPM2, SGCA, MYH7, NEB, SMN*). T1-TSE WBMRI sequences were reviewed and were abnormal in 67 % of the 106 patients, and unexpectedly in 27% of the 11 asymptomatic relatives. Diffuse striped signal abnormalities ('tiger-like') were very specific of ColVI-related myopathy. In addition, distinct involvement of muscles in the head, neck, trunk, girdles and limbs was observed in patients with *RYR1, SEPN1, GAA, LMNA or TPM2* mutations, leading to quite specific recognisable patterns in 34 patients. Abnormalities and pattern recognition were more frequent in patients studied due to selective spinal stiffness or scoliosis (80% abnormal, recognizable in 75% of cases) hyperlaxity syndrome (75%; 50%), or with confirmed myopathy but absence of these particular markers (71%; 40%). Pattern was compatible with the molecular diagnosis in 97%. Mild clinical involvement was revealed by muscle testing in 3 parents with abnormal WBMRI.

Overall, WB-MRI seems particularly useful in patients with indeterminate clinical or histological features of neuromuscular disease, especially if lower limb MRI features are not diagnostic. WB-MRI is a useful diagnostic tool when clinical neuromuscular findings suggest a disorder caused by mutations in genes which are difficult to analyse (RYR1, COL6, NEB), when gene variants of unknown pathogenicity are found or in cases where the disease has poor tissue specific markers (LMNA, SEPN1, GAA, SMN). Although additional observations in specific genetic entities are necessary and will improve confidence in use of WB-MRI to differentiate between different neuromuscular disorders, it

is already a valuable element in the diagnosis and monitoring of neuromuscular disorders in children. It has the potential to provide valuable assistance in interpretation of complex data from new molecular techniques such as whole-genome or exome sequencing and SNP-CGH array.
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