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Plenary Abstract

Emerging Role of Muscle Imaging in Diagnosis and Evaluation of Pompe Disease Patients

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The adult form of type II glycogenosis (GSDII) is a slowly progressive disease, with a few notable exceptions of patients who remain remarkably stable for decades and others who quite suddenly start to decline very quickly. A number of GSDII patients with non-typical presentation are misdiagnosed or diagnosed late. Whole-body muscle NMR imaging has proven useful in identifying as yet unrecognised GSDII patients by detecting muscle fatty infiltration patterns that are highly suggestive of the disease. In particular, Pompe patients show early fatty infiltration of tongue and subscapularis. Quantitative NMR imaging has interesting diagnostic applications but its primary role is to provide non-invasive biomarkers and assess disease progression and/or response to intervention. Studies have reported the effect of enzyme substitution therapy on muscle mass using NMRI. Imaging biomarkers may be of particular importance for adult-form GSDII therapeutic trials, where obvious clinical benefit has been difficult to document. Muscle water T2 increase is a non-specific process but it closely relates to "disease activity", whatever the underlying mechanisms: inflammation, necrosis, oedema. In FSHD patients, it has been suggested that abnormal T2 is predictive of fatty degenerative changes in subsequent years. We analysed the imaging data collected in the cohort of adult GSDII patients followed at the Institute of Myology, Paris, France. We focused our attention on the possible link between elevated muscle water T2 and the progression of fatty infiltration between yearly scans. NMR imaging was performed on a 3T Magnetom Trio TIM scanner, and muscle imaging was part of the routine yearly follow-up of GSDII patients. Despite the slow course of the disease in adults, a high percentage of muscles were found with an abnormally high T2: 32% of muscles had a water T2 above 39 ms. The progression of fatty degenerative changes as estimated by the fat fraction derived from Dixon acquisitions was remarkably slow: 0.9 ± 0.2%/yr. Nonetheless, the independent impact of elevated muscle water T2 and of enzyme substitution therapy on muscle fatty degenerative changes was evidenced. Muscles with elevated T2 experienced a fatty infiltration progression accelerated on average by 0.61%/yr ($p = 0.02$). Enzyme therapy slowed down the fatty degenerative changes on average by 0.68%/yr ($p = 0.01$). These original data show that quantitative NMR imaging can successfully monitor muscle lesions during natural history as well as assess the impact of treatment on muscle degenerative changes in GSDII patients.

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