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Clinical outcome study of dysferlinopathy: Lower limb water T2 predicts functional decline in patients with dysferlinopathy

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Water-T2 (T2H2O) mapping is used in muscular dystrophies to assess disease activity. It has been suggested as a surrogate outcome measure for clinical trials. However, the prognostic utility of T2H2O to identify changes in muscle function over time has not been described. A cohort of 18 patients (7 male) from two sites (Newcastle and Paris) with genetically confirmed dysferlinopathy were assessed as part of the Jain Foundation Clinical Outcomes Study of dysferlinopathy. Imaging used 3.0 T MRI clinical scanners with acquisition parameters standardised across sites. A multi-spin-echo sequence, with 17 equidistant echoes at 9.5ms spacing, was used for T2H2O mapping. T2H2O value was defined as higher or lower than the median in each muscle bilaterally. The degree of deterioration on four functional tests over three years was assessed in a linear model against covariates of high or low T2H2O at baseline, age, disease duration and baseline function. The T2H2O threshold which best predicted functional decline was determined. Higher T2H2O value correlated with greater functional decline in 21/35 muscles, and was never associated with slower decline ($p < 0.05$, correlation coefficients > 0.6). Higher T2H2O values in adductor magnus, vastus intermedius, vastus lateralis and vastus medialis were the most sensitive, being associated with greater decline in timed tests. Patients with a higher than median T2H2O value (40.6 milliseconds (ms)) in these muscles deteriorated 11 points more on the North Star Ambulatory Assessment for Dysferlinopathy (NSAD) and lost an additional 86 metres on the six-minute walk than those with a lower T2H2O value ($p < 0.05$). In dysferlinopathy, T2H2O did not correlate with current functional ability. However, T2H2O at baseline was higher in patients who worsened more rapidly on functional tests. With its capacity to predict progression, T2H2O mapping could be used to improve prognostication, patient selection and disease modelling for clinical trials.