The clinical presentation of facioscapulohumeral muscular dystrophy (FSHD) is quite variable in terms of severity and progression. We aimed to evaluate muscle involvement pattern and correlate the lesions on muscle image with clinical features and D4Z4 fragment size in FSHD. Twenty-four FSHD patients regularly followed at a medical center were enrolled in this study. The grading of the muscle image detected by computed tomography (CT) was based on a four-point semi-quantitative visual scale. The results of muscle grading were correlated with D4Z4 fragment size, disease duration, clinical severity scores, serum creatine kinase (CK) levels and muscle power. On muscle CT, the most affected muscle was the trapezius, followed by hamstrings. Interestingly, CT identified hamstrings involvement rather than shoulder-girdle in clinically asymptomatic subjects. CT image also showed that axial muscle was affected in one-third of patients which appeared even earlier than clinical manifestation and correlated well with disease duration. Strong correlations between CT findings, serum CK level and clinical severity scores were also found. Asymmetric involvement was more evident on CT image than manual muscle strength testing (MMT). Inverse correlation between CT grade and D4Z4 fragment size was clearly demonstrated. Our results revealed a preferential pattern of muscle involvement for FSHD. Early muscle involvement could be identified on CT image in clinically asymptomatic individuals. Therefore, muscle CT is helpful for the early intervention of FSHD even at a preclinical status.

doi:10.1016/j.nmd.2011.06.859

MR as biomarker for disease evaluation in facioscapulohumeral dystrophy
B.H. Jansen 1, R.J.W. Arts 1, N.B.M. Voet 1, C.I.H. Nabuurs 1, B.G.M. Engelen 1, A. Heerschap 1
1 Radboud University Nijmegen Medical Centre, Radiology, Nijmegen, Netherlands; 2 Radboud University Nijmegen Medical Centre, Neurology, Nijmegen, Netherlands; 3 Radboud University Nijmegen Medical Centre, Rehabilitation, Nijmegen, Netherlands

Facioscapulohumeral dystrophy (FSHD) is the third most common muscular dystrophy and is typically characterized by asymmetry of skeletal muscle involvement. Our method was to explore whether MR imaging and spectroscopy can serve as a non-invasive biomarker to assess disease severity, enable therapy evaluation and study disease pathobiology. Thirty genetically proven FSHD patients were examined on a Siemens Trio 3T system. Measurements were performed on the thigh muscles of the patients. Fat infiltration was quantitatively determined with 31-phosphorus MRS. Strength of the quadriceps and hamstrings muscles was assessed by a myometry setup. Three-hundred and fifty-three thigh muscles were investigated, of which 232 twice. Analysis of the fat fraction showed that muscles of FSHD patients are nearly binary divided between high and low fat fraction. Differences were found between the first and second measurement (100 days). Muscles with an intermediate fat infiltration showed the largest increase in fat fraction over time. This suggests that once disease onset is triggered it inevitably and rapidly leads to complete fat infiltration of the muscle. Fat fraction of the quadriceps and hamstring muscles correlated with the muscle strength. PCr/ATP ratio showed a significant decrease with increasing fat fraction. Fat fraction determined with MR can serve as a biomarker since it was sensitive enough to measure changes over a short period of time and correlates with muscle strength. The decrease in PCr/ATP ratio may be an attempt to compensate for overload in development towards a FSHD condition, for instance by adapting to a more oxidative phenotype.

doi:10.1016/j.nmd.2011.06.860