

## MRI Quantitative Measurements

### Quantitative fat measurements

Quantitative fat measurement should be included as **Supplemental** for FSHD. Quantitative fat measurement by MRI is increasingly being used in muscle disease as a sensitive early measure of disease progression. There are studies validating procedures in normal volunteers (Morrow, Sinclair et al. 2014), and showing sensitivity to change in limb girdle dystrophy 2I, along with a consensus statement about choice of technique (Willis, Hollingsworth et al. 2013; Willis, Hollingsworth et al. 2014). Duchenne muscular dystrophy has shown this to be reliable and sensitive to disease progression (Gaeta, Messina et al. 2012; Fischmann, Hafner et al. 2013). In FSHD there are two studies documenting a technique for making quantitative fat measurements and showing sensitivity of a select group of muscles (intermediate fat content) to disease progression over 3 months (Kan, Scheenen et al. 2009; Janssen, Voet et al. 2014). Ultimately the specific technique will depend on equipment and software at individual sites (the Dixon technique is perhaps currently the most common).

Fischmann, A., P. Hafner, et al. (2013). "Quantitative MRI and loss of free ambulation in Duchenne muscular dystrophy." J Neurol **260**(4): 969-974.

Gaeta, M., S. Messina, et al. (2012). "Muscle fat-fraction and mapping in Duchenne muscular dystrophy: evaluation of disease distribution and correlation with clinical assessments. Preliminary experience." Skeletal Radiol **41**(8): 955-961.

Janssen, B. H., N. B. Voet, et al. (2014). "Distinct disease phases in muscles of facioscapulohumeral dystrophy patients identified by MR detected fat infiltration." PLoS One **9**(1): e85416.

Kan, H. E., T. W. Scheenen, et al. (2009). "Quantitative MR imaging of individual muscle involvement in facioscapulohumeral muscular dystrophy." Neuromuscul Disord **19**(5): 357-362.

Morrow, J. M., C. D. Sinclair, et al. (2014). "Reproducibility, and age, body-weight and gender dependency of candidate skeletal muscle MRI outcome measures in healthy volunteers." Eur Radiol **24**(7): 1610-1620.

Willis, T. A., K. G. Hollingsworth, et al. (2013). "Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study." PLoS One **8**(8): e70993.

Willis, T. A., K. G. Hollingsworth, et al. (2014). "Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study." PLoS One **9**(2): e90377.