Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy

Alonso-Jimenez, Alicia

Kroon, Rosemarie H.M.J.M.

Alejaldre-Monforte, Aida

Nuñez-Peralta, Claudia

Horlings, Corinne G.C.

Van Engelen, Baziel G.M.

Olive, Montse

González, Laura

Verges-Gil, Enric

Paradas, Carmen

Márquez, Celedonio

Garibaldi, Matteo

Gallano,

Background and objective Oculopharyngeal muscular dystrophy (OPMD) is a genetic disorder caused by an abnormal expansion of GCN triplets within the PABPN1 gene. Previous descriptions have focused on lower limb muscles in small cohorts of patients with OPMD, but larger imaging studies have not been performed. Previous imaging studies have been too small to be able to correlate imaging findings to genetic and clinical data. Methods We present cross-sectional, T1-weighted muscle MRI and CT-scan data from 168 patients with genetically confirmed OPMD. We have analysed the pattern of muscle involvement in the disease using hierarchical analysis and presented it as heatmaps. Results of the scans were correlated with genetic and clinical data. Results Fatty replacement was identified in 96.7% of all symptomatic patients. The tongue, the adductor magnus and the soleus were the most commonly affected muscles. Muscle pathology on MRI correlated positively w