Supplementary figure 3. T1-weighted axial magnetic resonance imaging (MRI) of the lower limbs of patient 38 identified with a homozygous TRIM32 variant of unknown significance. MRI images at mid-thigh level on the left and mid-calf level on the right for: (A) patient 16 acquired at approximately 29 years of disease duration; (B) patient 38 at 11 years of disease duration; (C) patient 40 at 8 years of disease duration. (A) MRI images of patient 16 showed end-stage involvement of all muscles at the level of the thigh and the calf, with the exception of marked sparing of the gracilis muscle and the lateral gastrocnemius. Arrows indicate relatively spared muscles. (B) MRI images of patient 38 revealed a diffuse mild fatty infiltration of muscles of the anterior and posterior compartment of the thigh. Arrows indicate predominantly involved muscles at the calf level, i.e. muscles of the anterior and lateral compartment,
accompanied by asymmetric involvement of the medial gastrocnemius muscle (atrophy more prominent than fatty replacement). (C) MRI images of patient 40 showed end-stage involvement of all thigh muscles. At the calf level, end-stage involvement of muscles of the lateral compartment and of the gastrocnemius and soleus muscles was noted, with the remaining muscle groups being moderately involved. GR, gracilis muscle; LG, lateral gastrocnemius; MG, medial gastrocnemius.