Supplementary figure 3. Histopathologic findings in muscle biopsies of patient 37, 14, 15 and 39. (A) Hematoxylin and eosin (H&E) staining on the muscle biopsy of patient 37: nonspecific myopathic changes such as an increased number of internalised nuclei, fiber size variation and fiber splitting. (B) H&E staining showing dystrophic features on the muscle biopsy of patient 14: scattered necrotic muscle fibers (arrow) and endomysial fibrosis. (C, D) Trichrome staining on the muscle biopsy of patient 14: vacuoles containing basophilic material (arrow) in
scattered muscle fibers. (E) Trichrome staining on the muscle biopsy of patient 15: morphologically similar vacuoles are observed (arrow). (F) H&E staining on the muscle biopsy of patient 39: vacuoles (arrow) in scattered muscle fibers.