



**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.