

Figure e1: Histology. Muscle biopsy (patient 2) of the left *vastus lateralis* demonstrates wide variation in fibre size and shape due to the presence of scattered angular, atrophic fibres, occasionally in groups, as

well as abundant pyknotic nuclear clumps, both suggestive of neurogenic atrophy. No ragged red fibres or COX negative fibres were observed; no quantitation of COX staining intensity was performed. Cross-sections in **A** and **B** indicate myosin ATPase activity while the cross-section in **C** is stained with hematoxylin and eosin.

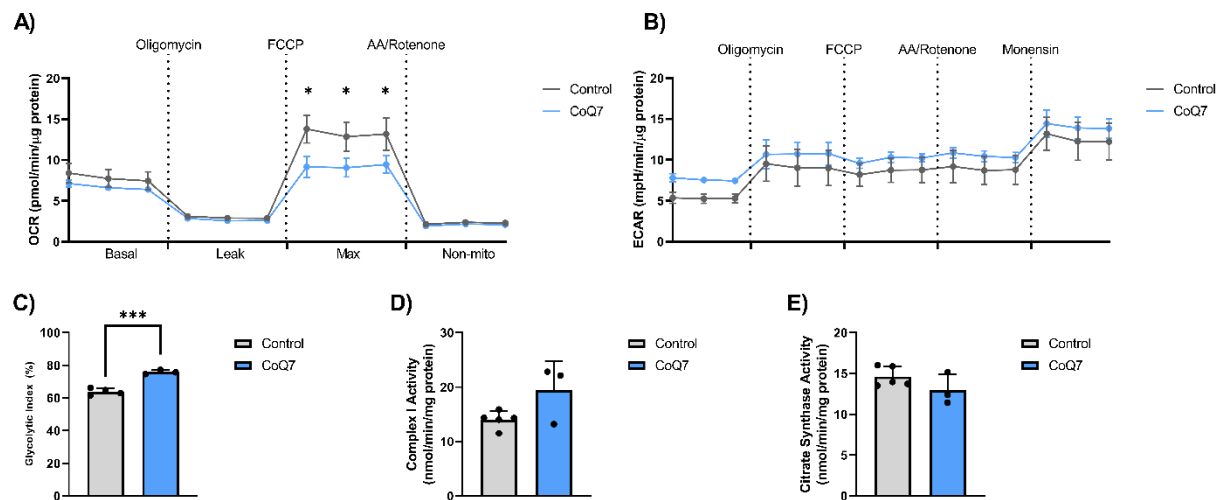


Figure e2: Supplemental Seahorse analysis on cultured skin fibroblasts. A: Oxygen consumption rate data from technical replicates underlying **Fig. 6A**. **B:** Extracellular acidification rate data for technical replicates underlying **Fig. 6B**. **C:** Glycolytic index. **D:** Complex I activity. **E:** Citrate synthase activity. All values are presented as means \pm SD. Comparisons between groups were as determined using a two-tailed Student's *t*-test for independent samples. * - $P < 0.05$, *** - $P < 0.001$.