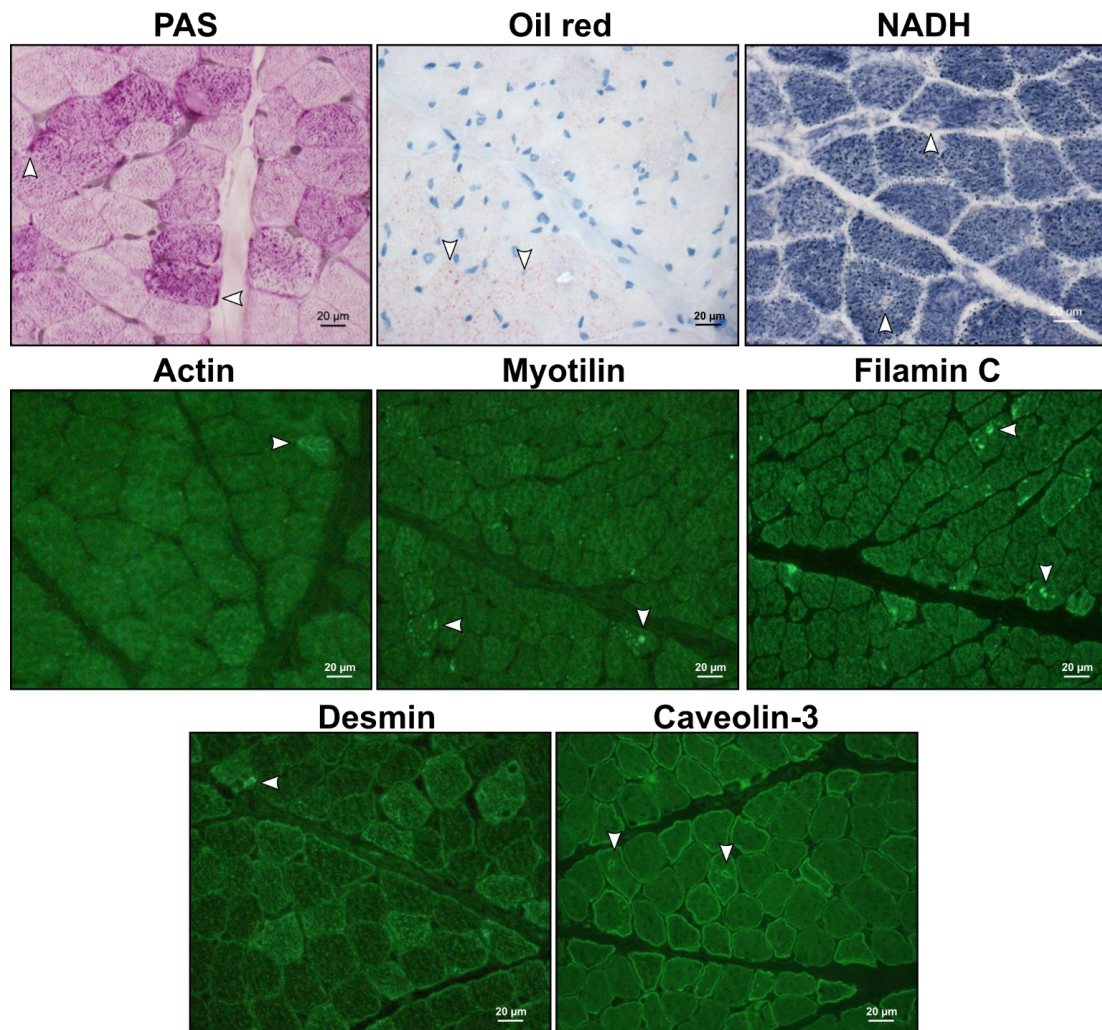


Muscle histology B-II-1



Supplementary Figure 1: Histological and immunofluorescence findings from individual B-II-1: The periodic acid–Schiff (PAS) reaction displayed deposits in a proportion of muscle fibres indicative for accumulation of polysaccharide-structures (white arrows). Oil red staining identified muscle cells with sarcoplasmic lipid accumulation (white arrows). NADH-reaction unraveled muscle fibres with reduced focal reactivity (white arrow). Immunofluorescence study of actin shows increased reactivity of in a minority of muscle cells (white arrow). Prominent build-up of deposits of myotilin, filamin C and desmin in a proportion of muscle fibres are respectively highlighted (white arrows). Immunofluorescence studies of caveolin-3 revealed an immunoreactivity of the rim of vacuoles (white arrows). Scale bar: 20 µm