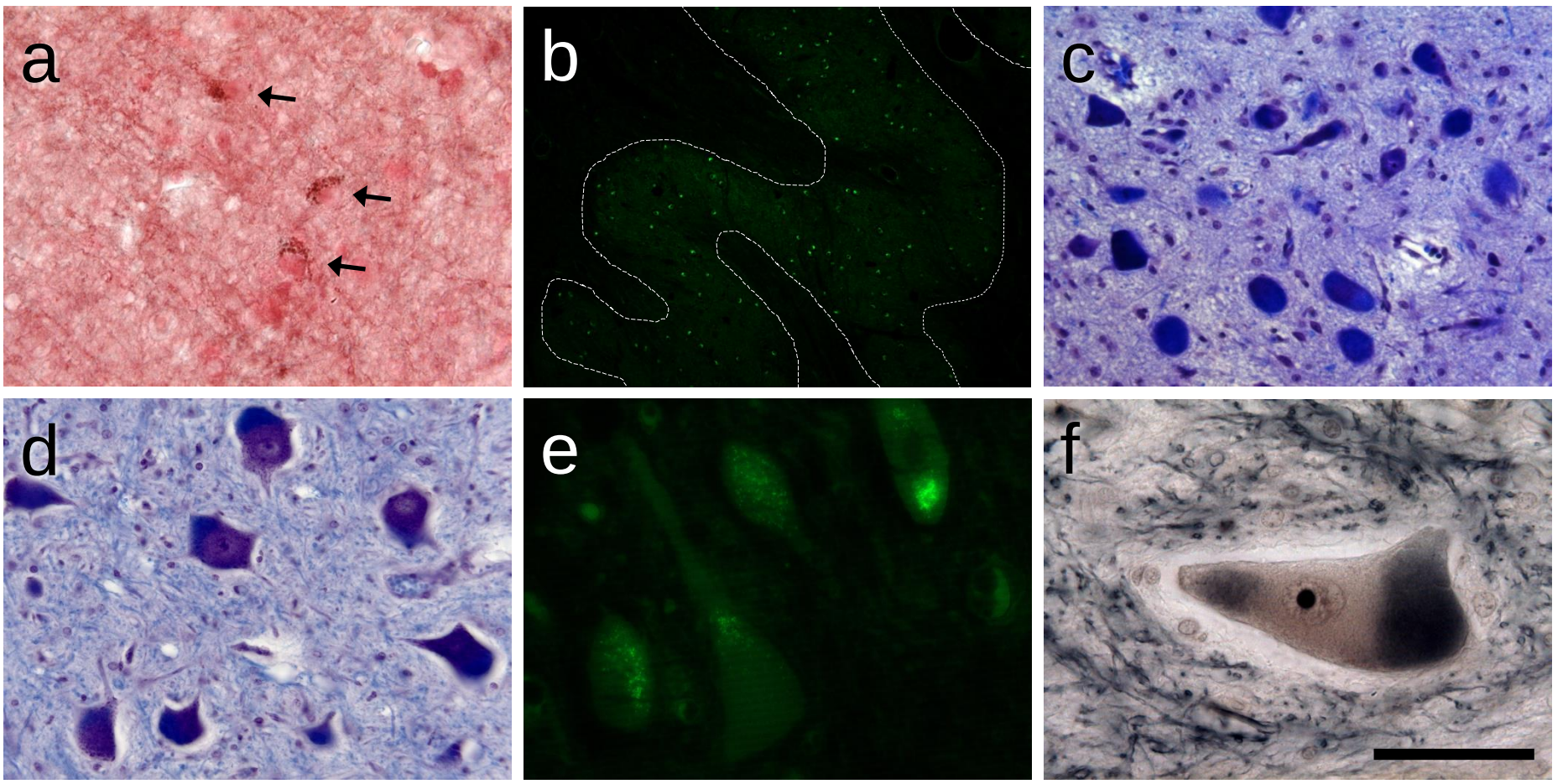
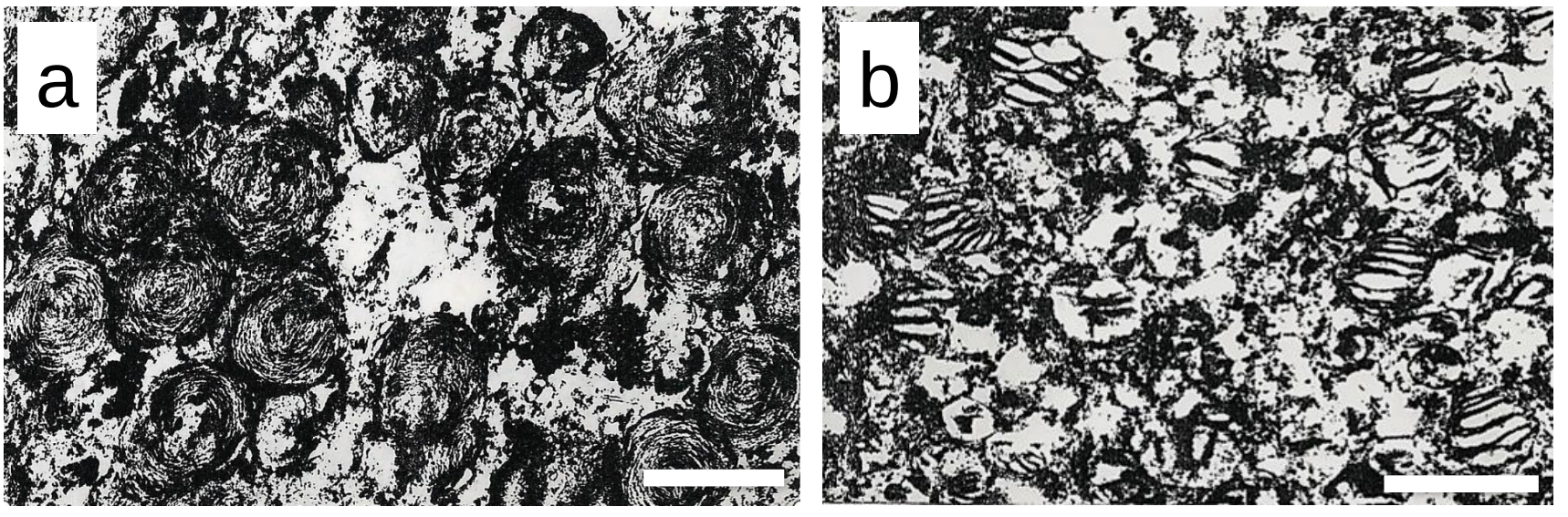
Supplement A



Neuronal storage material in patient 1. Neuronal inclusions with the same characteristics as those seen in her sister are present in the central nervous system of patient 1. The neurons were affected amongst other regions in the cortex (a, sudan black, arrows), nuclei olivary (b, green autofluorescence), nuclei nervi vagi (c, Kluver Barrera) and in the anterior horn of the spinal cord (d, Kluver Barrera; e, green autofluorescence; f, Heidenhain-Woelcke). Scale bar: 50 µm in a, e, f; 1000 µm in b; 125 µm in c, d.

Supplement B



Ultrastructural morphology of neuronal storage material. Both panels show cytoplasmic inclusions in neurons of the spinal cord anterior horn in glutaraldehyde-fixed, resin-embedded material of patient 1. In addition to the abundant neuronal lipofuscin granules (*Fig. 6*), there were occasional structures resembling membranous cytoplasmic bodies (a) and zebra bodies (b). Scale bars: 1 µm.