



**Supplementary Fig. 1.** Electrophysiological findings of proband and comparison of brain MRI in both siblings. Findings of a nerve conduction study (NCS) and electromyography (EMG) of the proband (A). The NCS showed no responses in the left common peroneal nerve or the left superficial peroneal nerve. The EMG findings were normal. Brain MRI findings for the proband (B) and his sister (C). Axial (left) and sagittal (right) T1-weighted images of both siblings demonstrated diffuse cerebellar atrophy, but atrophy of the cerebral cortex and brainstem was not observed.