Clinical and Molecular Characterization of *PMP22* point mutations in Taiwanese patients with Inherited Neuropathy

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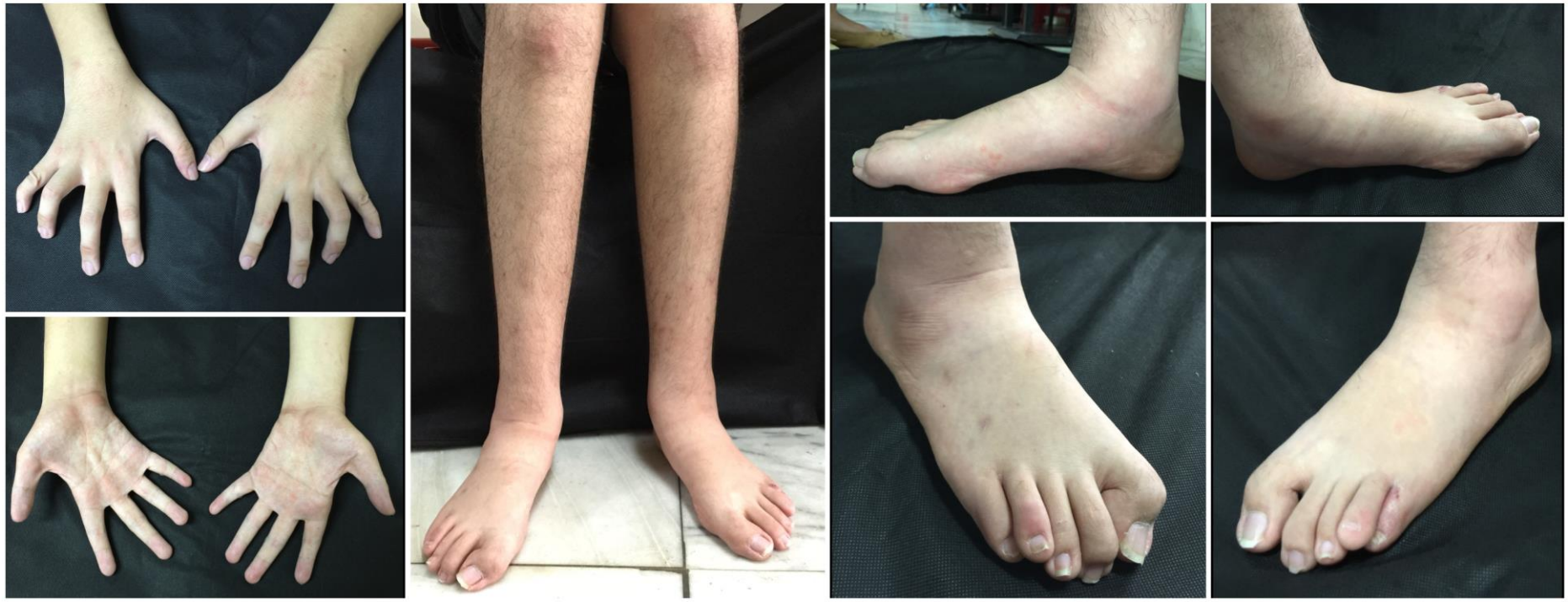
Supplementary Material

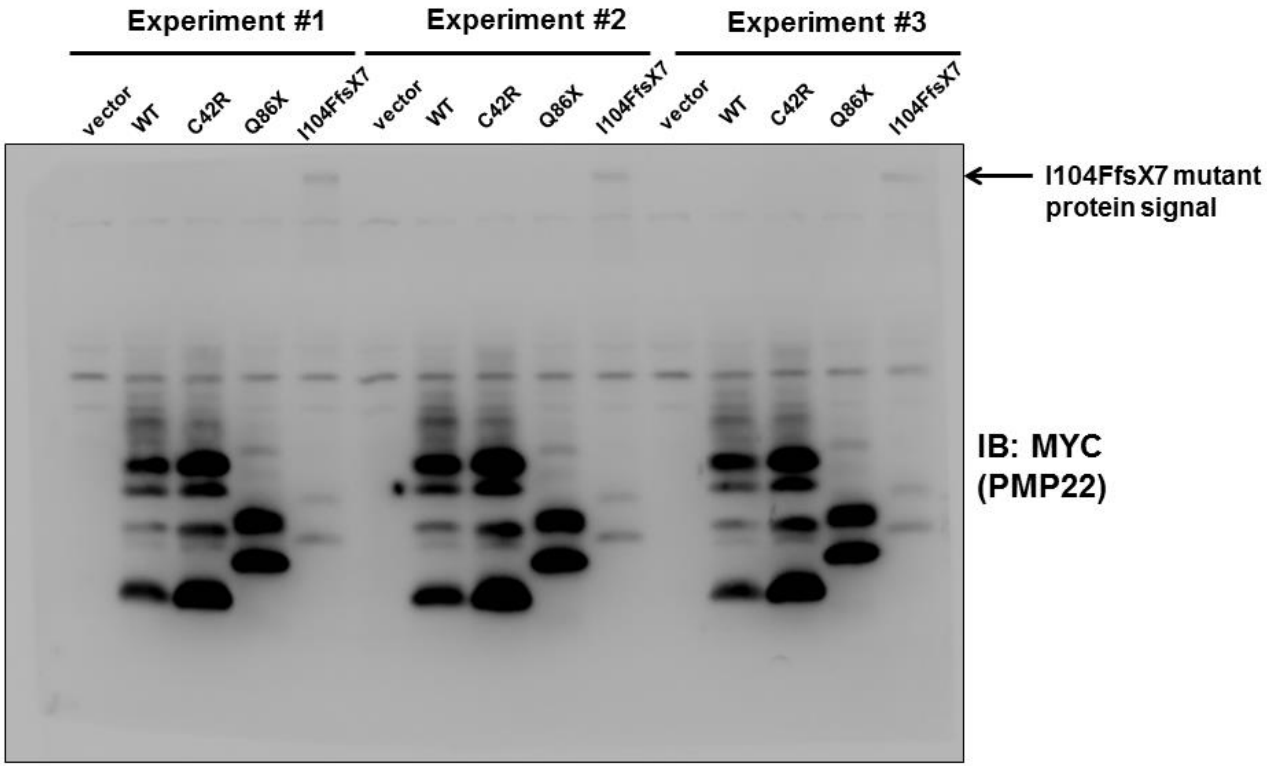
Supplementary Video Legend

The video of the patient D at age 26 years demonstrating pronounced atrophy of the leg muscles, bilateral foot drop and a steppage gait.

Supplementary Fig. S1. Atrophy of the intrinsic hand muscles as well as muscles in the feet and legs, and hammer toes in patient D harboring the *PMP22* p.I104FfsX7 mutation.



Supplementary Fig. S2. Western blot analysis of PMP22 protein expression in Neuro-2a cells showing I104FfsX7 mutant stuck at the interface between stacking gel and separating gel.



Supplementary Fig. S3. Cycloheximide (CHX)-chase assays showing different degradation rates of WT and mutant PMP22 proteins in RSC96 cells.