Supplementary Material

Clinical features and genetic results of other members of the pedigree

Both of the proband's uncles (II-3, II-7) had a history of gait abnormalities and muscle weakness in distal lower limbs for more than 30 years. They could still walk by themselves. Electrophysiological studies of his uncle (II-3) revealed sensorimotor demyelinating polyneuropathy with axonal degeneration (Supplementary Table 4 and 5 in the online-only Data Supplement). One of his nephew (IV-4) was 20 years old and had gait abnormalities at the age of 15. The proband's mother, one of his sister (III-2) and both of his daughters (IV-5, IV-6) had no symptom had no muscle atrophy but pes cavus. Electrophysiologic findings of both of his daughters supported evidence of a mild axonal neuropathy (Supplementary Table 6 and 7 in the online-only Data Supplement). A hemizygous GJB1 p.E208K mutation was also identified in his uncle (II-3), a heterozygous missense p.E208K mutation was identified in his mother (II-2), sister (III-2), and both of his daughters (IV-4, IV-5).