Supplementary Table 1. Clinical features of each Charcot-Marie-Tooth disease type 1 patients with *GJB1* variants

Family individual no.	Sex/ age (y)	AAO (y)	Family history	Initial symptoms	Muscle atrophy (UL/LL)	Sensory deficit (UL/LL)	Pes cavus	CNS manifestation	CMAP (mV)	MNCV (m/sec)	SNAP (uV)	SNCV (m/sec)	CMTNS	*GJB1* mutation	ACMG classification
F1–II:1	M/34	33	+	Gait abnormality	++/++	--/--	-	-	0.9	39.1	Absent	Absent	11	c.9G>A (p. Trp3*)	P
F2–III:5	M/32	30	+	Weakness in legs	++/++	--/++	+	-	ND	ND	ND	ND	NA	c.29T>C (p.Leu10Pro)	LP
F3–III:3	M/38	11	-	Weakness in legs	++/++	--/++	-	-	4.9	53.2	3.5	44.4	18	c.30C>T (p.Leu10Leu)	VUS
F4–II:1	M/14	10	+	Paroxysmal aphasia, limbs weakness	++/++	--/++	-	Episodic dysphasia, WM lesions	2.1	38.0	2.9	40.0	13	c.59T>C (p.Ile20Thr)	LP
F4–I:2	F/38	NA	+	None	--/--	--/--	+	-	ND	ND	ND	ND	NA	c.59T>C (p.Ile20Thr)	LP
F5–II:1	F/36	31	+	Weakness in legs	++/++	--/--	+	-	2.4	34.9	3.4	35.9	11	c.207C>A (p.Phe69Leu)	LP
F6–III:2	M/43	10	+	Frequent falls, limbs weakness	++/++	++/++	+	-	1.6	37.2	9.4	38.7	14	c.224G>A (p.Arg75Gln)	P
F7–III:3	M/17	14	+	Gait difficulty	--/--	--/++	+	Abnormal BAEPs	5.2	34.5	3.6	40.6	15	c.379A>T (p.Ile127Phe)	P
F7–III:4	M/10	7	+	Slender limbs	++/++	--/--	+	-	7.2	41.7	7.1	41.7	4	c.379A>T (p.Ile127Phe)	P
F7–II:8	F/34	NA	+	None	--/--	--/--	+	-	3.6	35.7	Absent	Absent	6	c.379A>T (p.Ile127Phe)	P
F7–II:6	M/42	38	+	Weakness in legs	--/-++	--/++	+	-	ND	ND	ND	ND	NA	c.379A>T (p.Ile127Phe)	P
F7–II:4	M/38	10	+	Gait disturbances	++/++	++/++	+	-	ND	ND	ND	ND	NA	c.379A>T (p.Ile127Phe)	P
F7–I:2	F/65	NA	+	None	--/--	--/--	+	-	4.9	39.2	1.3	41.5	3	c.379A>T (p.Ile127Phe)	P
F8–III:1	M/36	23	+	Distal limbs weakness	++/++	++/++	+	Nystagmus, gait ataxia	0.3	27.3	2.3	32.1	16	c.403_404insT (p.Tyr135Leufs*12)	P
F9–II:2	F/34	22	+	Unstable walking	--/++	--/--	+	Recurrent epileptic seizures	3.8	53.6	2.2	34.5	8	c.415G>A (p.Val139Met)	P
F9–II:1	F/36	NA	+	Slender limbs	++/++	--/--	+	-	3.5	49.7	15.2	47.6	2	c.415G>A (p.Val139Met)	P
F9–I:1	M/58	20	+	Distal limbs weakness, foot drop	++/++	--/--	-	-	ND	ND	ND	ND	NA	c.415G>A (p.Val139Met)	P
F10–III:6	M/30	15	+	Foot deformity	--/++	--/--	+	Paroxysmal dizziness, abnormal signal of CC	1.59	31.0	0.75	33.7	10	c.425G>A (p.Arg142Gln)	P
F10–III:7	F/26	NA	+	None	--/--	--/--	+	-	4.1	31.6	3.4	42.2	5	c.425G>A (p.Arg142Gln)	P
F11–II:2	F/44	33	+	Gait difficulty, weakness in legs	++/++	--/++	-	-	2.2	30.8	4.7	34.5	18	c.428_430delTGT (p.Leu143del)	P
F11–II:1	F/49	45	+	Weakness in legs	--/++	--/--	+	-	ND	ND	ND	ND	NA	c.428_430delTGT (p.Leu143del)	P
F11–III:2	F/20	19	+	Gait disturbance	--/++	--/--	+	-	ND	ND	ND	ND	NA	c.428_430delTGT (p.Leu143del)	P
F12–III:2	M/13	11	+	Weakness in legs, foot drop	--/++	++/++	-	-	0.9	28	1.34	37.9	11	c.533A>G (p.Asp178Gly)	P
F13–III:1	F/36	32	+	Limbs weakness	++/++	++/++	-	Abnormal WM signal	1.14	33.4	1.69	36.8	12	c.580A>G (p.Met194Val)	P
F13–II:3	M/57	Early	+	Weakness in legs	--/++	++/++	+	-	1.52	32.7	0.43	35.5	13	c.580A>G (p.Met194Val)	P
F14–II:1	M/21	11	+	Weakness in legs	--/++	--/--	-	-	1.83	33.9	1.39	36.4	11	c.590C>T (p.Ala197Val)	P
F14–I:2	F/46	42	+	Mild lower limb weakness	--/--	++/++	+	-	1.5	46.8	3.3	43.3	11	c.590C>T (p.Ala197Val)	P

All *GJB1* mutations in male patients are hemizygous, and heterozygous in female patients.
AAO, age at onset; ACMG, American College of Medical Genetics and Genomics Standards and Guidelines; BAEPs, brainstem auditory evoked potentials; CC, corpus callosum; CMAP, median compound muscle action potential; CMTNS, Charcot-Marie-Tooth neuropathy score version 2; CNS, central nervous system; early, early childhood; F, female; *GJB1*, gap-junction beta-1 gene; LL, lower limbs; LP, likely pathogenic; M, male; MNCV, median motor nerve conduction velocity; NA, not available; ND, not done; P, pathogenic; SNAP, median sensory nerve action potential; SNCV, median sensory nerve conduction velocity; UL, upper limbs; VUS, variant of uncertain significance; WM, white matter.