Supplementary Table 1. TUBB4A mutation phenotype with hypomyelination and spastic paraplegia

Genotype	Onset age, yr	Family history	Delay motor milestone	Spasticity	Polyneuropathy	Dystonia	Ataxia	Cognitive impairment	Brain MRI		References
									Basal ganglia atrophy	Cerebellar atrophy	
D355V	13	De novo	ND	+	ND	-	-	+	-	+	Di Bella et al.,[1] 2021
G96R	13–17	De novo	-	+	ND	+	-	Minimal	-	-	Lu et al.,[2] 2017
D355V	Childhood	De novo	ND	+	ND	-	-	Mild	-	+a	Sagnelli et al.,[3] 2016
H190Y	18 months	De novo	-	+	ND	-	-	Minimal	-	-	Nicita et al.,[4] 2016
H190Y	1–12 months	AD	+	+/-	+	-	+	-	-	+	Kancheva et al.,[5] 2015
R282P	2, 5	De novo	ND	+	-	+	+	Mild	-	+	Pizzino et al.,[6] 2014
Q292K	3	De novo	ND	+	ND	+	+	-	-	-	Pizzino et al.,[6] 2014
V255I	< 2 yr	De novo	+	+	ND	-	+	+	+	-	Pizzino et al.,[6] 2014
E410K	12 months	De novo	+	+	ND	+	-	+	-	+	Blumkin et al.,[7] 2014
F341L*	35	AD	-	+	-	-	+	-	-	+	Our patient
F341L*	23	AD	-	+	-	-	-	-	-	-	Our patient
F341L*	21	AD	-	+	-	-	-	-	-	-	Our patient

'+a' means 'PET indicate hypometabolism in cerebellar region'.
*our case.
MRI, magnetic resonance imaging; ND, not mentioned; AD, autosomal dominant; PET, positron emission tomography.

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