

Supplementary material

Supplementary table 1. Neuroradiological findings in patients with *ITPA* variants in the literature and the present study.

Patient No. (Reference)*	1 (Present study)	2 (Present study)	3 (2)	4 (2)	5 (2)	6 (2)	7 (2)	8 (2)	9 (2)	10 (4)	11 (5)	12 (5)	13 (7)
Age at MRI (months)	5	4	8	6	4.5	4	4.5	8	2	5	5	5	4
Myelination	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed	Delayed
Diffusion-restriction	[†] PLIC, red nucleus	PLIC, dentate nuclei, cerebellar white matter	Not done	PLIC, midbrain	PLIC, optic radiation, midbrain, cerebellar white matter	PLIC, optic radiation, cerebellar white matter	PLIC, optic radiation	PLIC, optic radiation, decussation of superior cerebellar peduncles in midbrain	PLIC-pyramidal tracts	Not known	PLIC-pyramidal tracts	PLIC, optic radiation	PLIC, optic radiation, globus pallidus, cerebral peduncles, red nuclei
T2 Hyperintensity	PLIC	PLIC, midbrain, hilus of dentate nucleus	PLIC, midbrain, cerebellar white matter, cerebellar peduncles	PLIC, midbrain, hilus of dentate nucleus	PLIC, midbrain, cerebellar white matter, cerebellar peduncles	PLIC, cerebral peduncle, cerebellar white matter, cerebellar peduncles	PLIC	PLIC-pyramidal tracts	cerebral white matter, hilus of dentate nucleus, pyramidal tracts in midbrain, Medullary pyramids	PLIC	PLIC-pyramidal tracts	PLIC, midbrain	PLIC, cerebellar white matter
Corpus callosum	Thin	Thin	Thin	Thin	Thin	Thin	Thin	Thin	Normal	Thin	Thin	Thin	Not Known
Cerebral atrophy	+	--	+	+	--	+	--	+	--	+	--	+	+
MR spectroscopy	Normal	Not done	Not done	Not done	Normal	Not done	Not done	Not done	Not done	Not known	Not known	Not known	Not known

*Reference number in parentheses
 PLIC: posterior limb of the internal capsule.