Supplementary Online Content

Paradas C, Akman HO, Ionete C, et al. Branching enzyme deficiency: expanding the clinical spectrum. *JAMA Neurol*. Published online November 18, 2013. doi:10.1001/jamaneurol.2013.4888

eTable 1. Nerve Conduction Study Values in Patient 1

eTable 2. Nerve Conduction Study Values in Patient 2

eFigure. Brain Magnetic Resonance Imaging in Patient 1

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eTable 1. Nerve Conduction Study Values in Patient 1

Patient 1																
Motor study *																
Stimulation site	**Median**				**Ulnar**				**Common Peroneal**				**Tibial**			
	L	***A***	***D***	***V***	***L***	***A***	***D***	***V***	***L***	***A***	***D***	***V***	***L***	***A***	***D***	***V***
Distal	3.6	7.2	-		2.8	7.8	-		4.4	0.4	-		4.6	1.8	-	
Proximal		6.2	-	50		6.6	-	53		0.4	-	37		1.0	-	36
Sensory study**																
	Median				**Ulnar**				**Radial**				**Sural**			
	L	***A***	***V***		***L***	***A***	***V***		***L***	***A***	***V***		***L***	***A***	***V***	
Distal	3.6	9	53		2.7	10	53		2.5	17	53		ND	ND	ND	

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eTable 2. Nerve Conduction Study Values in Patient 2

Patient 2																
Motor study *																
Stimulation site	**Median**				**Ulnar**				**Common Peroneal**				**Tibial**			
	L	***A***	***D***	***V***	***L***	***A***	***D***	***V***	***L***	***A***	***D***	***V***	***L***	***A***	***D***	***V***
Distal	3.5	6.3	6.0		3.4	8.0	7.1		3.6	9.6	12		4.5	3.4	5.5	
Proximal		5.6	6.5	47		8.5	7.5	56		9.3	12.1	45		3.0	6.0	40
Sensory study**																
	Median				**Ulnar**				**Superficial Peroneal**				**Sural**			
	L	***A***	***V***		***L***	***A***	***V***		***L***	***A***	***V***		***L***	***A***	***V***	
Distal	2.6	6.0	55		2.1	3.0	54		3.0	2.0	40		3.0	3	35	

L= distal motor latency (ms); A= compound muscle action potential amplitude (mV); D= motor action potential duration (ms); V= motor conduction velocity (m/s); ND= not detected; - = value non available. *Motor amplitude, lower normal limits: median-ulnar 5 mV; common peroneal- tibial 2 mV. Distal motor latency, upper normal limits: median-ulnar 3.9 ms; common peroneal-tibial 5 ms. Motor conduction velocity, lower normal limits: median-ulnar 50 m/s; common peroneal -tibial 40 m/s. **Sensory amplitude, lower normal limits: median-ulnar 10 mV; superficial peroneal- sural 5 mV. Distal sensory latency, upper normal limits: median-ulnar 3.4 ms; superficial peroneal-sural 3.5 ms. Motor conduction velocity, lower normal limits: median-ulnar 50 m/s; superficial peroneal - sural 40 m/s

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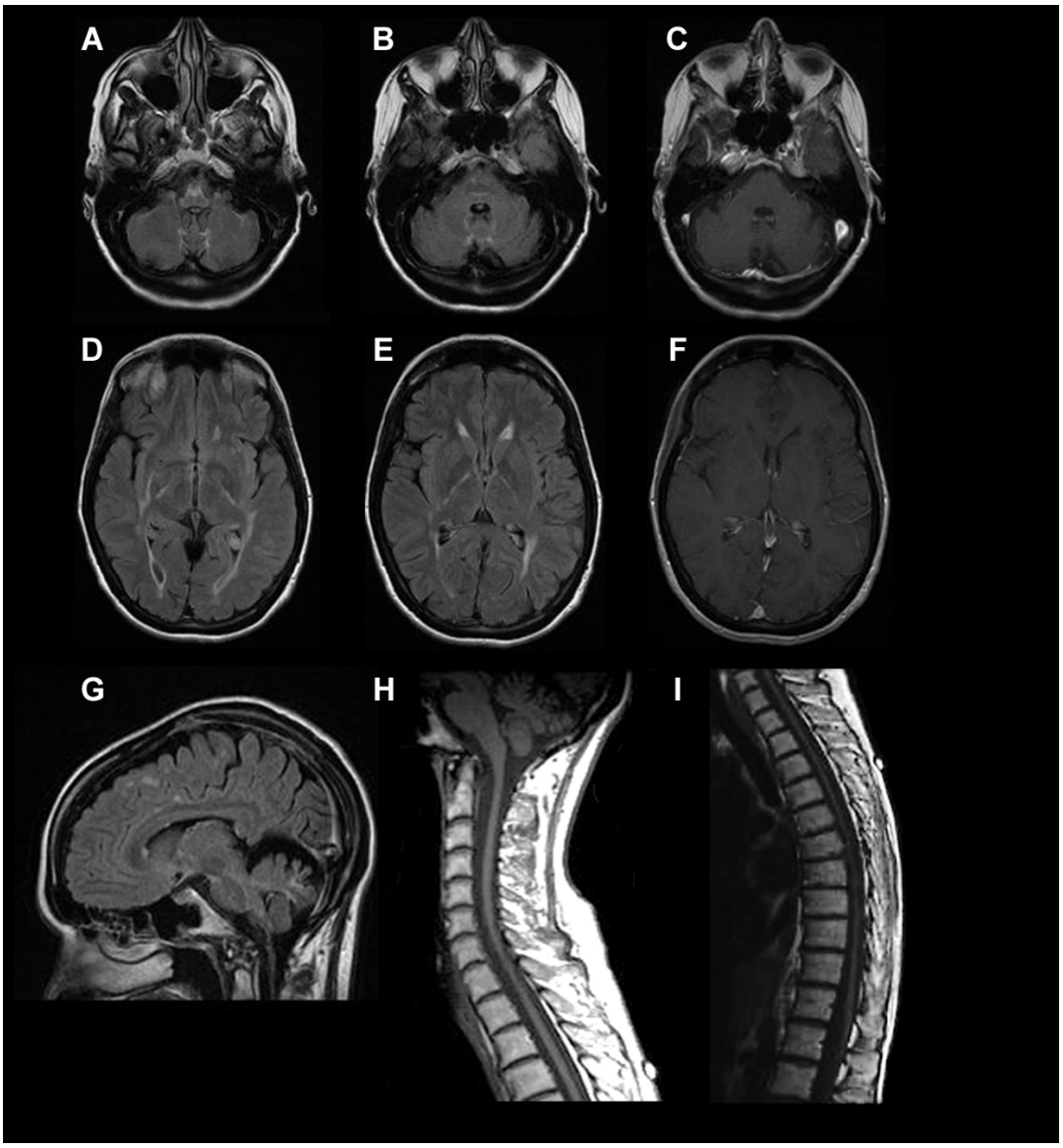

eFigure. Brain Magnetic Resonance Imaging in Patient 1. FLAIR axial images show bilaterally increased signals at the level of brainstem in the medulla, anterior pons, and cerebellum (A, B). FLAIR also showed symmetric hyperintensities in the internal and external capsules, and in the periventricular, callosal, and deep white matter (D, E, F). Scattered subcortical lesions were visible in sagittal scans (G). There were no areas of abnormal enhancement (C, F). Sagittal scans of the spinal cord (T1) showed diffusely small caliber at both cervical and dorsal levels (H, I).

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