Giant axonal neuropathy
A clinicoradiopathologic diagnosis

A 10-year-old girl without consanguinity or neurologic disease in the family presented with an 8- to 9-year history of progressive gait disturbance, incoordination, and impaired hearing and cognition; antenatal and neonatal histories were unremarkable. Salient examination findings were tightly curled scalp hair (figure 1A), impaired cognition and hearing, flaccid quadriparesis, and pancerebellar dysfunction. Salient investigative findings were leukodystrophy on MRI brain (figure 1B), sensorimotor polyneuropathy, and giant axons with aberrant neurofilament immunostaining on sural nerve biopsy (figure 2, A and B). Giant axonal neuropathy is a rare (worldwide 50 families reported) autosomal recessive disorder characterized by gigaxonin gene mutations and disorganization of intermediate filaments.1,2

(A) Hair changes. (B) T2-weighted axial MRI brain shows bilateral posterior limbs of internal capsules and deep white matter hyperintensities.

Sural nerve biopsy shows (A) giant axons (yellow arrow) on hematoxylin & eosin staining (×200), (B) neural intermediate filament immunostaining (arrows; black [normal], red [abnormal], ×200).

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Author contributions: Dr. Israni, Dr. Chakrabarty, and Dr. Akbar worked up the patient and prepared the manuscript under the guidance of Dr. Gulati. Dr. Kumar provided the radiologic inputs. Dr. Sable and Dr. Suri provided the pathologic inputs.

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