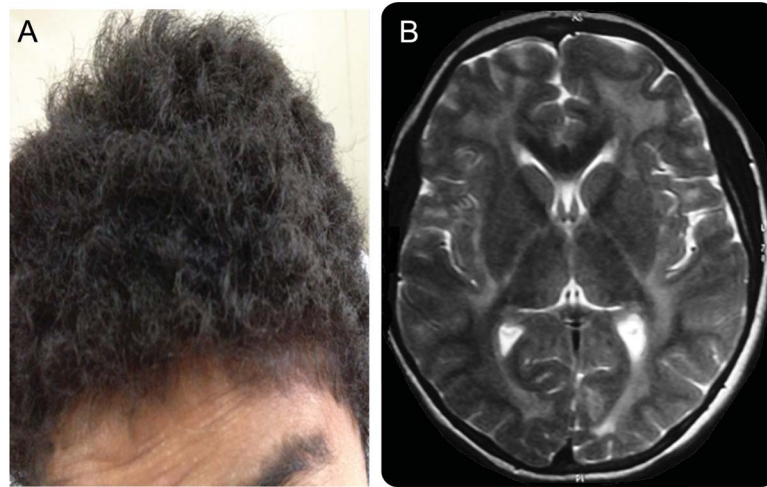


# Giant axonal neuropathy

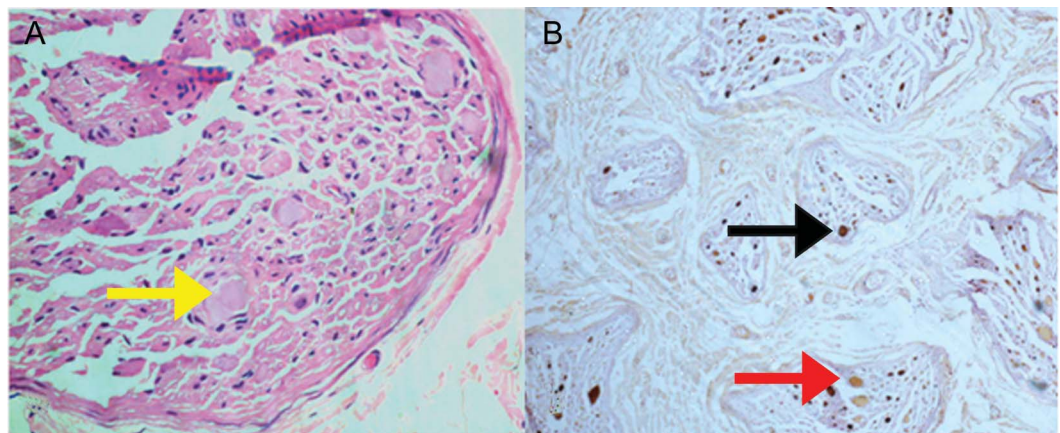
## A clinicoradiopathologic diagnosis

**Figure 1** Hair and MRI brain changes in giant axonal neuropathy



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

**Figure 2** Sural nerve biopsy changes in giant axonal neuropathy



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

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 quadriplegia, 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.<sup>1,2</sup>

Anil Israni, MD, Biswaroop Chakrabarty, DM, Sheffali Gulati, MD, Akbar Mohamed, CH MD, Atin Kumar, MD, Mukund N. Sable, MD, Vaishali Suri, MD

From All India Institute of Medical Sciences, New Delhi, India.

*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.

*Study funding:* No targeted funding reported.

*Disclosure:* The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

*Correspondence to Dr. Gulati:* sheffaligulati@gmail.com

1. Kuhlensbaumer G, Timmerman V, Bomont P. Giant axonal neuropathy. *Gene Rev* 2013 (updated 2012).
2. Mahammad S, Murthy SN, Didonna A, et al. Giant axonal neuropathy-associated gigaxonin mutations impair intermediate filament protein degradation. *J Clin Invest* 2013;123:1964–1975.

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*Neurology* 2014;82;816-817

DOI 10.1212/WNL.0000000000000177

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