



“UNMASKING THE UNCOMMON: A NEUROFASCIN-144 POSITIVE NEUROPATHY”

Dr Swarup Hange¹ Dr Sangeeta Rawat¹ Dr Neeraj Jain¹ Dr.Dnyaneshwar Asole¹
Department of Neurology KEM Hospital, Mumbai

CASE HISTORY

61 Year, righted handed male with history of acute onset severe pain in both calves
Followed by tingling sensation in both lower limb and upper limb finger tips.



Next day patient had lower limb distal weakness followed by semi distal followed by proximal weakness



No cranial nerve , Bowel bladder involvement
No history of prior episode of loose motion or Upper Respiratory tract infection.

GENERAL EXAMINATION

Pulse – 140 /min, Blood pressure– 160/90 mmhg

NEUROLOGICAL EXAMINATION

	BOTH UL	BOTH LL
TONE	NORMAL	NORMAL

	ACTION	POWER BOTH LL
HIP JOINT	EXTENSION	4
KNEE JOINT	FLEXION	4
ANKLE JOINT	DORSI & PLANTAR FLEXION	4

DTR	RIGHT	LEFT
UL-BICEP/TRICEP	+2	+2
LL-KNEE	-	-
LL-ANKLE	-	-
PLANATAR	M	M

INVESTIGATIONS

TEST	RESULT
Nerve Conduction test	Sensorimotor Axonal Polyneuropathy affecting both lower limbs
CSF studies	P-90 mg/dl, S-110 mg/dl, Cells-45(100% l)
MRI spine with root contrast	Normal
CT pulmo angio	Normal

TREATMENT

- IVIG given but no response to it

- In view of predominant autonomic dysfunction, neuropathic pain and sensorimotor axonal neuropathy on NCS and no response to IVIG

Nodopathy Panel was sent
Neurofascin- 265 (0 to 233 ng/ml)

- Patient was given 5 days of inj. methylprednisolone f/b inj Rituximab two dosage 2 week apart-
- Autonomic symptoms and neuropathic pain improved after 1 month

DISCUSSION

- **Neurofascin 140 IgG** is associated with pan neurofascin disease spectrum characterized by fulminant disease course resulting in profound sensorimotor tetraplegia, severe cranial nerve involvement, and autonomic dysfunction.
- Patients presenting as GBS and CIDP with additional atypical neurological and systemic features and responding poorly to IVIG should be evaluated for Nodopathies.

References-Khadilkar SV, Kamat S, Patel R. Nodo-paranodopathies: Concepts, Clinical Implications, and Management. Ann Indian Acad Neurol. 2022 Nov-Dec;25(6):1001-1008. doi: 10.4103/aian.aian_382_22. Epub 2022 Aug 4. PMID: 36911467; PMCID: PMC9996523.