



May 1998 EMG Case-of-the-Month

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HISTORY

A 30-year-old right hand dominant male presents with an eight-year history of painless progressive distal weakness. Due to a strong positive, autosomal dominant family history, he was previously given the diagnosis of hereditary motor and sensory neuropathy, type I (HMSN I). Over the past several years, he has noted significant worsening of his distal weakness. However, over the past several months he has noted even more prominent weakness in extension of the left wrist and fingers which he assumes was part of the disease process. He has been employed as a commercial fisherman for six years, involving heavy manual labor using the upper limbs.

- **Prior to continuing, please develop a differential diagnosis, and list each diagnosis in order of likelihood.**
- **Is there any additional information from the clinical history that might be helpful in clarifying your differential list or changing its order of priority?**

COMMENTARY I

This man has a family history of hereditary motor and sensory neuropathy, type I (HMSN I). This is a slowly progressive disorder characterized by diffuse muscle weakness predominating distally with prominent distal atrophy. Thus, it might be assumed that the disease might be the source of his recent, asymmetric, upper limb weakness.

Previous work has shown that, on average, HMSN I subjects produce 20% to 40% less force than normal controls using quantitative isometric and isokinetic strength measures, even though manual muscle test (MMT) scores may be normal. However, there is usually no significant side-to-side difference in strength. Given his history of heavy work, one must consider common causes of unilateral upper extremity weakness in this setting, including cervical radiculopathy and entrapment neuropathy.

HISTORY, continued

His work history includes multiple musculoskeletal injuries, with several blunt traumas to the wrists and forearms. He had a dorsal ganglion cyst removed from his left wrist two years prior to the onset of his recent symptoms. Otherwise, the remainder of his past medical history is unremarkable.

- **If necessary, revise your differential diagnosis based on the additional clinical history.**



- **On what details of the physical examination do you think you should focus at this point?**

COMMENTARY II

The possibility of asymmetric weakness associated with HMSN remains, but would be atypical. Focal neuropathy must be considered. The history of repeated blunt trauma to the forearms is important since this puts the patient at higher risk for traumatic neuropathy. Entrapment neuropathy is also a possibility.

PHYSICAL EXAMINATION

Isolated to the left upper limb, there is radial deviation of the wrist on extension and marked weakness of digital extension at the metacarpophalangeal joints, including the thumb. No other weakness is detected in the upper limbs including the intrinsic muscles of the hands. Muscle stretch reflexes are present though difficult to elicit throughout the upper and lower limbs. Sensation to light touch and pinprick is grossly intact including the radial nerve distribution.

- **At this point, review your differential diagnosis, and revise as appropriate.**
- **Are there additional observations on physical examination that might be helpful in narrowing your differential list?**

COMMENTARY III

The extent of the weakness in the left upper limb and its limitation of distribution to the posterior interosseus nerve make a strong case for a mononeuropathy superimposed upon the HMSN. The various causes of mononeuropathy and the possibility of mononeuropathy multiples must be considered.

PHYSICAL EXAMINATION, continued

In the lower limbs, there is mild distal weakness, and the patient has high arched feet. Diminished vibratory and proprioceptive sensation in the feet are the only sensory deficits detected. No cafe au lait spots are noted. There is no palpable enlargement of the greater auricular, median, or ulnar nerves.

COMMENTARY IV

The previously noted point that, for the most part, hereditary neuropathies are symmetrical, is critical. The findings clearly demonstrate focal, asymmetrical weakness, which should raise a red flag for a secondary, localized problem which needs to be addressed in the electrodiagnostic study. This problem is localized to the posterior interosseus nerve, and there are no other findings of focal neuropathy elsewhere. Also, there is no evidence of generalized hypertrophic neuropathy.



ELECTROPHYSIOLOGIC DATA

SENSORY NERVE CONDUCTION									
nr = no response									
NERVE	LATENCY (ms)			AMPLITUDE (µV)			CONduc VEL (m/s)		
	R	L	Norm	R	L	Norm	R	L	Norm
median (at 14 cm)	4.7	4.9	<3.5	16	19	>10			-
radial (at 10 cm)	2.8	2.9	<2.5	18	20	>10			-
ulnar (at 14 cm)	4.9	5.0	<3.5	17	15	>10			-
sural	nr	nr	-			-			-

MOTOR NERVE CONDUCTION									
nr = no response									
NERVE	LATENCY (ms)			AMPLITUDE (mV)			CONduc VEL (m/s)		
	R	L	Norm	R	L	Norm	R	L	Norm
median	-	-	-	-	-	-	-	-	-
wrist to thenar	5.7	-	<4.5	10.0	-	>6.0			-
elbow to thenar	11.9	-	-	6.0	-	-	34	-	>50
ulnar	-	-	-	-	-	-	-	-	-
wrist to hypothenar	-	5.1	<4.5	-	10.5	>6.0			-
BE to hypothenar	-	11.5	-	-	7.0	-	-	40	>50
radial	-	-	-	-	-	-	-	-	-
BE to EDC	-	5.5	<3.5	-	2.0	>4.0			-
AE to EDC	-	7.7	-	-	1.4	-	-	36	>50
peroneal	-	-	-	-	-	-	-	-	-
ankle to EDB	8.0	-	<6.0	7.2	-	>5.0	-	-	>40
below FH to EDB	17.4	-	-	4.5	-	-	29	-	-
tibial	-	-	-	-	-	-	-	-	-
ankle to medial foot	-	7.2	<6.0	-	5.5	>4.0			-
pop fossa to med. foot	-	21.5	-	-	4.5	-	-	30	>40

- **On the basis of both the clinical and electrodiagnostic evaluations, formulate your final impression. List the most likely diagnosis followed by**



other possibilities that are not excluded by the data. Eliminate those diagnoses not supported by the data.

- **Are there additional observations on physical examination that might be helpful in narrowing your differential list?**

ELECTROPHYSIOLOGIC DATA, continued

ELECTROMYOGRAPHY										
n = normal incr = increased decr = decreased 0 = absent 1+ = minimal 4+ = maximal crd = complex repetitive discharge fasc = fasciculation potential myk = myokymic discharge myt = myotonic discharge nmt = neuromyotonic discharge p wave = positive sharp waves fib = fibrillation potentials recr = recruitment amp = amplitude dur = duration poly = polyphasic potential										
R/L	MUSCLE	INSERTION		SPONTAN		VOLUNTARY				
		activ	p wave	fib	other	recr	amp	dur	poly	effort
L	cervical paraspinal	N	0	0	-	-	-	-	-	-
L	infraspinal	N	0	0	-	N	N	N	N	-
L	deltoid	N	0	0	-	N	N	N	N	-
L	biceps	N	0	0	-	N	N	N	N	-
L	triceps (lat)	N	0	0	-	N	N	N	N	-
L	brachioradialis	N	0	0	-	N	N	N	N	-
L	extensor carpi rad longus	N	0	0	-	N	N	N	N	-
L	extensor carpi ulnaris	incr	3+	3+	-	2+decr	2+incr	2+incr	1+incr	-
L	extensor digitorum	incr	3+	3+	-	2+decr	2+incr	2+incr	1+incr	-
L	flexor carpi radialis	N	0	0	-	N	N	N	N	-
L	supinator	N	0	0	-	N	N	N	N	-
L	extensor indicis proprius	incr	3+	3+	-	2+decr	2+incr	2+incr	1+incr	-
L	1st dors interos	N	0	0	-	N	N	N	N	-
L	abd pol brev	N	0	0	-	N	N	N	N	-
R	cervical paraspinal	N	0	0	-	N	N	N	N	-
R	upper extremity screening EMG	N	0	0	-	N	N	N	N	-

- **Make the final revisions of your diagnostic impression(s).**



- **What other diagnostic procedures (laboratory tests, etc.), if any, are needed?**
- **What treatment would you recommend?**

DIAGNOSTIC IMPRESSION

There is an isolated focal lesion of the left posterior interosseus nerve. The clinical and electrodiagnostic data are consistent with the previous diagnosis of hereditary motor and sensory neuropathy, type I.

COMMENTARY

Radiographs of the forearm and elbow were normal.

The patient was referred for surgical exploration. The radial nerve was isolated proximally between the brachioradialis and the extensor carpi radialis longus muscles. The PIN was identified distally between the tendons of the extensor carpi radialis brevis and the extensor digitorum communis. The distal branches of the PIN were visualized exiting just distal to the superficial head of the supinator muscle as well as the entire supinator. The superficial head of the supinator was incised throughout its entire length, exposing the PIN completely. The five possible sites of compression in the radial tunnel were examined: fibrous bands proximally, the vascular leash from the radial artery, the leading edge of the extensor carpi radialis brevis, the proximal edge of the supinator (Arcade of Frohse), and the distal edge of the supinator.

Just proximal to the distal edge of the superficial head of the supinator, there was fusiform swelling of the PIN. This was the only pathological finding. The nerve was of normal caliber and appearance distal and proximal to the enlargement. All potential sites of compression were released and external neurolysis and epineurotomy of the PIN was performed.

At three-month follow-up, the patient did not have significant clinical improvement in function. Tendon transfer to augment digital extensor function is being considered if no recovery occurs in the ensuing 6-12 months.

This case highlights an unusual situation of a pre-existing hereditary motor and sensory polyneuropathy with a superimposed, focal hypertrophic mononeuropathy. Although HMSN is often described as a generalized hypertrophic neuropathy (GHN), only about half of the cases have palpable enlargement of the nerves, with a specific histological "onion bulb" pattern described on microscopic dissection. Our patient did not have clinical or surgical evidence of this pattern. This "onion bulb" formation represents focal myelin reduplication (redundant layers of Schwann cells) and is not specific to HMSN. Similar histopathological findings are also seen in Refsum's syndrome, diabetic neuropathy, relapsing hypertrophic neuropathy, and, infrequently, multiple sclerosis.

Although multifocal onion bulb hypertrophy is not uncommon, isolated focal nerve hypertrophy is quite unusual. A very rare and poorly understood entity, localized hypertrophic neuropathy (LHN), may present with focal hypertrophy confined to a single nerve segment. Nerve conduction studies and needle EMG in LHN demonstrate evidence of axonal loss, rather than primary demyelination. There is debate as to whether this entity represents perineural cell hyperplasia versus focal myelin reduplication. Although superficially similar to GHN on microscopic examination, electron microscopy in LHN reveals



whorls of thinned, markedly elongated structures resembling perineural cells. It is debated as to whether there is a distinction between reactive perineural cell proliferation due to focal damage and a perineurioma, a benign nerve tumor. Thus, histologically LHN appears to be a distinct entity, and not simply a localized form of GHN. Further, unlike GHN, LHN appears to affect primarily major nerve trunks in the upper extremities with a predilection for the PIN, a nerve known to be susceptible to compression or trauma. Previous reports in patients without generalized neuropathies have documented PIN compression from a variety of masses, as well as the fibrous edge of the Arcade of Frohse, fibrous bands proximal or distal to the Arcade, or by tightness of the passage within the supinator.

The exact etiology of LHN is unknown, although many authors suggest local trauma as a possible cause. This is supported by similar histologic findings after experimental disruption of the perineural barrier. Although LHN has a predilection for the PIN, other case reports have described LHN in nerves not prone to compression.

Surgical exploration of a focal neuropathy in the presence of a generalized neuropathy is controversial, but was felt to be warranted in this case due to the profound loss of function in the PIN musculature, markedly out of proportion to the remainder of the upper limbs. Although external neurolysis and decompression were performed, internal neurolysis and biopsy were not done due to the potential for additional nerve injury. Thus, this lesion is not histologically defined. Other authors have previously advised that internal neurolysis is not advisable in LHN, which may cause painful neuroma formation.

FINAL DIAGNOSTIC IMPRESSION

This patient most likely had chronic, repetitive, focal trauma to the posterior interosseous nerve resulting in localized hypertrophic neuropathy with distal axonal degeneration.

Since the patient reported frequent blunt trauma to the forearm, and since there is electrophysical evidence of severe axonal injury to the PIN, and since, at surgery, the PIN appeared to have normal caliber both proximal and distal to the lesion, it is likely that traumatic disruption of the perineural barrier is the cause of the problem. It may also be possible that such lesions occur more readily in nerves already compromised by demyelination (i.e., HMSN I).

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