Hereditary Motor Sensory Neuropathy With Pyramidal Signs – Report Of Rare Case Presentation With Review Of Literature

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Abstract

Introduction: Hereditary motor and sensory neuropathy is a chronic degenerative disorder of the peripheral nerves. It has neuropathic pattern of motor and sensory deficits without evidence of Upper motor neuron signs such as spasticity, brisk deep tendon reflexes.
Material and Methods: We report a case of 19 year old male having above mentioned features of neuropathy and pyramidal signs combined. We also have done literature search to find similar case reports.
Results: There were 9 articles having cases presenting with neuropathy and pyramidal tract signs. Genetic mutations were mentioned in 5 articles. Our patient had heterozygous deletions encompassing COX 10 and PMP22 gene mutation. Spasticity was mentioned in three out of nine articles similar to our case. Flexor plantar response was seen in four out of nine articles as in our case.
Conclusion: HMSN may have unusual presentation like spasticity, brisk deep tendon reflexes, extensor plantar response. Neurophysicians should also be aware of the variety of differentials that have such presentation. The clinical clue to diagnosis is the presence of foot deformities, chronic indolent course of illness. Genetic analysis is usually confirmatory.

Keywords: HMSN, CMT, hereditary motor sensory neuropathy with pyramidal signs, Abnormal Deep Tendon Reflex, , Spasticity, Genetic Sequence Database,

Introduction

Hereditary motor sensory neuropathy also called as Charcot Marie Tooth disease[1,2] is a chronic progressive neurodegenerative disorder involving the peripheral nerves. Its clinical features include insidious onset of distal lower limb weakness with or without sensory symptoms, tendency to fall. Signs include distal foot deformities, distal weakness, reduced or absent deep tendon reflexes, impaired sensations of touch, pain, vibration and proprioception and sensory ataxia. Diagnosis[3,4,5,6,7,8] is based on clinical history, involvement of family members, clinical examination findings, investigations [9,10] including nerve conduction studies, electromyography studies and genetic analysis[11]. Imaging can be done to rule out other differential diagnosis. We present a case of hereditary motor sensory neuropathy presenting with pyramidal signs spasticity and brisk deep tendon reflexes.

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Case Report

A 19-year-old male, with no known comorbid illness, presented with history of tripping of toes in right more than left lower limb for the past two years. He also noticed that he could not balance himself while squatting. He also had minimal difficulty in wearing footwear. He had no other upper limb or sensory or autonomic symptoms. He had no significant past history or family history. He denied any addictions and had no known exposure to toxins or chronic medication use.
On examination, he was conscious and oriented with stable vitals and no postural Blood Pressure variation. He had bilateral claw toes and pes cavus foot deformity (fig 1,2).

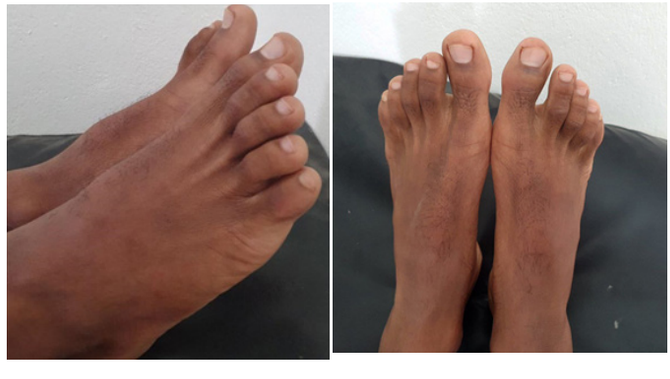

Fig 1,2. Showing prominent Extensor digitorum brevis tendon and claw toes

There were no neurocutaneous markers/trophic ulcers. Higher mental function examination was normal. Cranial nerve examination was normal. Motor system examination showed distal muscle wasting especially in bilateral Extensor Digitorum brevis (EDB) and spasticity in both lower limbs. There was weakness of bilateral ankle dorsiflexion grade 4/5 and eversion grade 3/5. Deep tendon reflexes were exaggerated in bilateral knees and elicitable in both ankles. Plantar was flexor bilaterally and his superficial abdomen reflexes were present. Upper limb motor examination including tone, bulk, power, reflexes were all normal. On palpation, there were non-tender thickened nerves in both common peroneal nerves. Sensory system examination did not show any abnormalities. Spine examination revealed no abnormalities. He had bilateral high stepping gait.

INVESTIGATIONS:

Routine blood investigations including complete blood count, renal and liver functions tests were normal. Workup for inflammation/vasculitis - ESR, CRP, ANA by IFA were negative. NCS (table 1-3) was suggestive of bilateral symmetrical demyelinating motor predominant sensorimotor neuropathy of all limbs with conduction block in bilateral peroneal nerves (fig 3,4). There was prolonged distal motor latency of bilateral median, ulnar, peroneal and tibial nerves. CMAP were not recorded from bilateral peroneal nerves above fibular head. F waves were prolonged in Right ulnar, right peroneal and not recordable from left peroneal. Sensory conduction showed prolonged peak latency of bilateral median and ulnar nerves

Table 1 Motor Nerve Conduction:

Nerve and Site	Latency	Amplitude	Duration	Segment	Lat.diff.	Distance	C.V.
Median.L							
Wrist	5.4 ms	15.18 mV	10.3 ms	Abductor pollicis brevis-Wrist	5.4 ms	mm	m/s
Elbow	10.0 ms	14.40 mV	10.6 ms	Wrist-Elbow	4.6 ms	230 mm	50 m/s
Median.R							
Wrist	6.7 ms	16.08 mV	6.7 ms	Abductor pollicis brevis-Wrist	6.7 ms	mm	m/s
Elbow	10.8 ms	14.34 mV	7.2 ms	Wrist-Elbow	4.1 ms	230 mm	56 m/s
Ulnar.L							
	ms	mV	ms	Abductor digiti minimi (Manus)-Wrist	4.4 ms	mm	m/s
Wrist	4.4 ms	16.63 mV	10.4 ms	Wrist-Below elbow	4.1 ms	240 mm	59 m/s
Below elbow	8.5 ms	15.89 mV	10.4 ms		ms	mm	m/s
Ulnar.R							
Peroneal.L							
Ankle	7.8 ms	8.421 mV	9.5 ms	Extensor digitorum brevis-Ankle	7.8 ms	mm	m/s
Fibula (head)	16.3 ms	7.581 mV	9.7 ms	Ankle-Fibula (head)	8.5 ms	315 mm	37 m/s
P. fossa	……..	……..	……..	Fibula (head)-Fibula (head)+2	ms	mm	m/s
Peroneal.R							
Ankle	9.5 ms	5.238 mV	10.3 ms	Extensor digitorum brevis-Ankle	9.5 ms	mm	m/s
Fibula (head)	17.2 ms	4.443 mV	9.9 ms	Ankle-Fibula (head)	7.7 ms	310 mm	40 m/s
P. fossa	……..	……..	……..	Fibula (head)-Fibula (head)+2	ms	mm	m/s
Tibial.L							
Ankle	7.2 ms	21.06 mV	9.2 ms	Abductor hallucis-Ankle	7.2 ms	mm	m/s
Popliteal fossa	16.9 ms	17.50 mV	10.3 ms	Ankle-Popliteal fossa	9.7 ms	380 mm	39 m/s
Tibial.R							
Ankle	7.6 ms	23.31 mV	8.1 ms	Abductor hallucis-Ankle	7.6 ms	mm	m/s
Popliteal fossa	17.1 ms	21.28 mV	8.9 ms	Ankle-Popliteal fossa	9.5 ms	375 mm	39 m/s
Peroneal.L							
Ankle	8.4 ms	8.616 mV	9.1 ms	Extensor digitorum brevis-Ankle	8.4 ms	mm	m/s
Fibula (head)	16.5 ms	7.702 mV	10.2 ms	Ankle-Fibula (head)	8.1 ms	mm	m/s
Fibula (head)+2	18.4 ms	2.510 mV	9.2 ms	Fibula (head)-Fibula (head)+2	1.9 ms	mm	m/s
Fibula (head)+4	……..	……..	……..		ms	mm	m/s
Fibula (head)+6	……..	……..	……..	Fibula (head)-Fibula (head)+4	ms	mm	m/s
P. fossa	……..	……..	……..		ms	mm	m/s
	……..	……..	……..	Fibula (head)+6-P. fossa	ms	mm	m/s

Peroneal.R							
Ankle	9.5 ms	5.290 mV	11.4 ms	Extensor digitorum brevis-Ankle	9.5 ms	mm	m/s
Fibula (head)	17.0 ms	4.378 mV	10.8 ms	Ankle-Fibula (head)	7.5 ms	mm	m/s
Fibula (head)+2	18.1 ms	4.049 mV	10.2 ms	Fibula (head)-Fibula (head)+2	1.1 ms	mm	m/s
Fibula (head)+4	……..	……..	……..		ms	mm	m/s
Fibula (head)+6	……..	……..	……..	Fibula (head)-Fibula (head)+4	ms	mm	m/s
P. fossa	……..	……..	……..		ms	mm	m/s
	……..	……..	……..	Fibula (head)+6-P. fossa	ms	mm	m/s

Table 2 F-Wave Studies

Nerve	M-Latency	F-Latency
Median.L	5.9	31.1
Median.R	6.9	32.5
Ulnar.L	4.5	31.9
Ulnar.R	4.5	34.3
Peroneal.L	9.0	……
Peroneal.R	9.7	66.0
Tibial.L	7.7	56.6
Tibial.R	7.7	51.9

Table 3: Sensory Nerve Conduction:

Nerve and Site	Onset Latency	Peak latency	Ampli-tude	Segment	Dist.	CV	
Median.L							
Wrist	3.8 ms	5.1 ms	30 µV	Digit II (index finger)-Wrist	150 mm	40 m/s	
Median.R							
Wrist	4.1 ms	5.1 ms	19 µV	Digit II (index finger)-Wrist	145 mm	35 m/s	
Ulnar.L							
Wrist	4.2 ms	5.7 ms	16 µV	Digi v-wrist	130mm	31m/s	
Ulnar.R							
Wrist	4.5 ms	6.0 ms	20 µV	Digi-v-wrist	125mm	28 m/s	
Sural.L							
Lower leg	3.6 ms	4.3 ms	26 µV	Ankle-Lower leg	140 mm	39 m/s	
Sural.R							
Lower leg	3.8 ms	4.7 ms	22 µV	Ankle-Lower leg	140 mm	37 m/s	
Superficial peroneal.L							
Ankle	3.4 ms	4.3 ms	14 µV	Dorsum of foot-Ankle	120 mm	35 m/s	
Superficial peroneal.R							
Ankle	2.9 ms	3.3 ms	13 µV	Dorsum of foot-Ankle	115 mm	40 m/s	

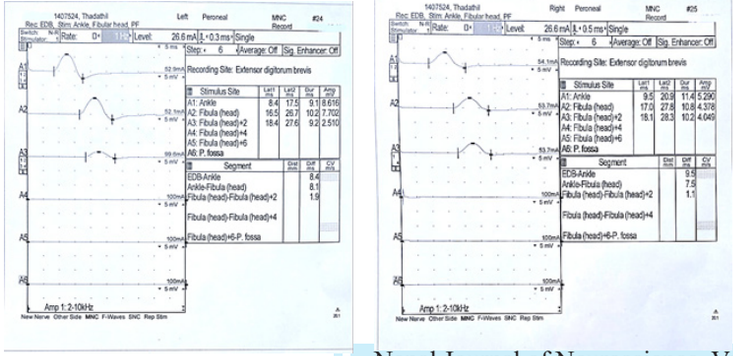

Fig 3,4. Showing Motor nerve conduction study of Left and right peroneal nerve. Conduction block 4cm above both fibular head is seen.

MRI Whole spine and Brain imaging done to rule out pyramidal tract involvement was negative. CSF analysis was acellular with normal protein and sugar levels.

A contiguous heterozygous deletion of size [~1070.45 kb], on chromosome 17 (17p12 microdeletion) encompassing COX10 and PMP22 genes was detected on genetic analysis using whole exome next generation sequencing.

Hence our patient was diagnosed with hereditary motor sensory neuropathy type 1 - demyelinating pattern with NCVS showing demyelination with conduction block in motor conduction and prolonged sensory latencies.

Review of Literature:

We did an extensive literature search (Pubmed, Google Scholar). We included all case reports or articles pertaining to neuropathy and coexisting pyramidal pattern findings similar to our case here. We excluded diagnosis of Leukodystrophies, Subacute Combined degeneration (SACD), Motor neuron diseases and patients with delayed milestones, cerebral palsy.

We found 9 articles having findings similar to what we have described. The information is recorded in table 4 below.

Table 4. Articles with case series or reports of neuropathy with pyramidal tract signs

Author	Age	Sex	Gene mutation	NCS	Deformities	Brisk Reflex	Spasticity	Babinski
Fusco Carlo [12]	16	M	EGR2	Sensorimotor polyneuropathy	Yes	Yes	Yes	Yes
Bienfait HM [13]	Case series of 61 patients from 18 families		MFN2, BSCL2, and RAB7	Neuropathy	Yes	Yes in 36%	No	Yes(n=10)
Eduardo Luis de Aquino Neves [14]	Case series of 35 patients from a large family			Neuropathy	Yes	Yes in 46%(n=14)	No	Yes(n=14)
Angelini [15]	Case series from a family		HMSN V	Neuropathy	Yes	No	Yes (n=1)	No
M F Dohrn [16]	48	M	KIF 5A	Axonal Motor neuropathy in Legs	Yes	Yes, with absent ankle jerk	No	-
Pipis M [17]	Case series of 30 patients from 8 families		NEFH	Motor and sensory neuropathy in Lower limbs	Yes	Yes in 52%(n=13)	No	Yes
Adriana P Rebelo[18]	Case series of 3 families		CADM3	Neuropathy	Yes	Yes in 2 patients	No	no
S Vucic [19]	Case series from 2 families		CMT105 and CMT66 (no association with PMP 22, etc)	Neuropathy	Yes	Yes (n=7/27)	Yes (n=8/27)	Yes(n=13/27)
Biancheri [20]	Two sibling age 16 and 18	Male and Female	GDAP1	Neuropathy	Yes	Yes	Not mentioned	No

There were 5 articles in which the causative genes were EGR2, MFN2, BSCL2, and RAB7, KIF 5A, NEFH and CADM3. The initial two genes mentioned are presumed to be the most common. Our case had heterozygous deletions encompassing COX 10 and PMP22 gene mutation. Neuropathy was present in all cases based on Nerve Conduction and Electromyography finding, but none had presence of conduction block or combined demyelinating and axonal pattern. Foot deformities were present in all including our case. Brisk deep tendon reflexes especially knee jerks were mentioned in all journals with varying incidence. Spasticity was mentioned in three of nine articles. Our case had definite hypertonia of both lower limbs. Extensor plantar response was not seen in 3 articles as in our patient who had flexor plantar response.

DISCUSSION:

Hereditary neuropathies are a complex group of Lower motor neuron diseases[21] that have an indolent course over years to decades. They account for nearly 40% of chronic polyneuropathies. The evidence of family history, associated skeletal abnormalities such as hammer toes, scoliosis, high arched foot, lack of positive sensory symptoms, symmetrical findings, early age of onset, slowly progressive course is strongly in favour of hereditary neuropathies. Even if there is truly negative family history, the possibility of hereditary neuropathy cannot be ruled out[22].

Initially there were two classifications of Charcot-Marie-Tooth disease – CMT 1 and CMT 2 (23,24). CMT 1, Autosomal Dominant disease was due to disorder of peripheral myelination resulting from a mutation in the peripheral myelin protein-22 (PMP22) gene. It usually manifests in the first decade of life with reduced reflexes, distal muscle wasting and sensory deficits. CMT 1 has demyelinating pattern of neuropathy. CMT 2 is also autosomal dominantly inherited which manifests in the second decade of life. It has also similar pattern as CMT 1 with more significant sensory and motor deficits. CMT2 is axonal type of neuropathy mainly due to mutations in the ATP1A1 gene. There is another type-infantile/early childhood onset, CMT type 3 - known as Dejerine-Sottas disease which results in severe demyelination with delayed motor skills and is much more severe than CMT type 1. CMT 3 also includes congenital hypomyelinating neuropathy. It occurs in early infancy with hypotonia, delayed milestones and difficulty in feeding. Both diseases in CMT 3 are genetically heterogenous.

After the advent of genetic testing, many more genes have been implicated in cause of CMT which is now labelled as HMSN 1 with diffusely slow nerve conduction velocity -hypertrophic neuropathy and HMSN 2 with normal or borderline abnormal nerve conduction velocity -neuronal/ axonal type. Other types of CMT/HMSN include type[4 ,5,6,7,] X. There are multiple subtypes within each type with different gene affections and variable presentations including proximal muscle involvement, entrapment neuropathies, vocal cord and diaphragm paralysis[25,26]. HMSN 4 is Refsum's disease [27] with Autosomal recessive pattern with distal sensory motor neuropathy, hypertrophic nerves, skin manifestations like ichthyosis. HMSN type V [28] has similar presentation to our case, i.e., neuropathy with pyramidal tract signs, except that it is characterized by normal upper limbs and the absence of sensory symptoms. HMSN 6 and 7 are associated with vision abnormalities with former having optic atrophy[29] and latter associated with retinitis pigmentosa. CMT X has both dominant and recessive pattern of inheritance. There is distal muscle weakness in usually second decade of life with demyelinating pattern neuropathy.

The mainstay of treatment is supportive therapy. Use of orthosis and tendon corrective surgeries is when deformities are hindering with the daily activities. It requires multidisciplinary care from neurologist, orthopedician and physical medical rehabilitation departments.

The differential diagnosis[5,6,7,30] includes hereditary neuropathy liable to pressure palsies, CIDP, toxin induced neuropathy; myeloneuropathy – B12 deficiency, HIV, neurosyphilis; Motor neuron diseases – SMA, ALS; Amyloidosis, Fabry's disease, Hereditary spastic paraplegia.

CONCLUSION:

This article with review of literature is to highlight the

importance of the variable presentation of hereditary peripheral neuropathy as in this patient where there is associated pyramidal findings in form of spasticity and brisk knee jerks. With such possible presentations, genetic testing can be of great value and thus averting other unnecessary tests, procedures and treatments which are intended for other differential diagnosis.

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