

# Peroneal Muscular Atrophy with Pyramidal Signs: Where do we Stand?

## Introduction

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### Introduction

Inherited neuropathies are a clinically and genetically heterogeneous group of disorders. The identification of more than 30 causative genes for the inherited neuropathies has raised important questions regarding the approach to their diagnosis [1]. Inherited neuropathies can be broadly classified into two groups: those in which neuropathy is the sole or primary part of the disease (e.g., Charcot-Marie-Tooth disease, CMT) and those in which the neuropathy is a part of more generalised neurological or multisystem disorder (e.g. familial amyloidotic neuropathy) [1].

Historically, CMT was more commonly called hereditary motor and sensory neuropathy (HMSN). Dyck and Lambert classified hereditary motor and sensory neuropathies into seven subgroups in 1968 on the basis of clinical, electrophysiological, pathological and genetic characteristics [2]. Peroneal muscular atrophy with pyramidal features are rare, but should be identified as a distinct disorder [3]. This disorder has been referred to as hereditary motor and sensory neuropathy (HMSN) type V by Dyck [2].

Since the original description of peroneal muscular atrophy by Charcot, Marie, and Tooth in 1886, it has been well recognised that occasional patients with the syndrome have pyramidal features. This is rare and occurs in less than 5% of cases [4].

The HSMN V classification has largely been overshadowed by work in the hereditary spastic paraplegias, specifically those designated "complicated" spastic paraplegia by having both spasticity and other features, including possibly neuropathy [2]. Nevertheless, the HSMN V classification may be appropriate in individuals in whom the peroneal

atrophy and lower motor neuron involvement is predominant over spasticity [2]. It is relatively benign condition which does not appear to shorten life expectancy or lead to severe disability [3].

### Case Report

A 18-year old male, born of non consanguineous marriage and uneventful antenatal period presented with history of delayed motor milestones noticed since the age of 6 months and difficulty in getting up from sitting and squatting position as well as difficulty in walking since early childhood. By the age of 10 year he had stiff gait with tendency to trip. His parents noticed symmetrical wasting of all extremities especially distal, more in lower limbs at around age of 10 year. His symptoms progressed slowly, was ambulatory and able to do activity of daily living (ADL) independently. His siblings and other family members were reportedly normal. General physical examination revealed Marfanoid habitus (pectus carniatum, pes planus, genu varus), contracture of Achillis tendon and tight hamstrings. There was no nerve thickening. All cranial nerves including fundus and visual acuity were normal.

He had symmetrical diffuse atrophy below mid thigh and mid forearm (Figure 1). He had spasticity in all extremities at proximal joints but decreased tone at distal joints. He had symmetrical weakness of all extremities distal more than proximal without any weakness of neck and trunk muscles. Superficial reflexes were present with bilateral extensor plantar response. His supinator and ankle reflexes were absent bilaterally, where as biceps, triceps and knee reflexes were brisk bilaterally. He had spastic gait. He had no sensory, cerebellar or extra pyramidal features.

His routine haematological and biochemical investigations were normal. Nerve conduction studies revealed significantly prolonged distal latencies, decreased CMAPs and uniform slowing of conduction velocities with non-recordable F waves in all tested nerves suggestive of demyelinating and axonal affection of lower limbs more than upper limbs. SNAPs were nonrecordable in all tested nerves. VEP study was normal. MRI brain was normal. MRI spine showed diffuse atrophy of thoracic spinal cord (Figure 2).

Nerve biopsy sections showed disorganised nerve bundles with endoneurial edema and fibrosis. No significant inflammation or granuloma was seen.



**Figure 1:** Photograph of patient showing symmetrical distal wasting of extremities.

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Figure 2: MRI dorsal spine T2weighted image showing spinal cord atrophy.

## Discussion

Our case reported here superficially resembles those with HMSN types I and II (peroneal muscular atrophy). The range of age of onset with distribution of muscle atrophy and weakness were similar. However important clinical finding in our case was presence of significant UMN features in the form of brisk reflexes, extensor plantar responses and spastic gait. In terms of classification this case of peroneal muscular atrophy with pyramidal features can be allocated to HMSN V, or can also be classified amongst the complicated forms of HSP. Hereditary spastic paraplegias are more commonly designated “uncomplicated” by not having neuropathy or syndromic multiple neurologic system or systemic illness compared to their autosomal recessive and X linked counterparts, in which these features are more common [2].

Peroneal muscular atrophy with pyramidal features is a rare disorder. This disorder has been referred to as hereditary motor and sensory neuropathy (HMSN) type V by Dyck [2]. The HSMN V classification has largely been overshadowed by the work in hereditary spastic paraplegias, specifically those designated as “complicated” spastic paraplegia by having both spasticity and other features, including possibly neuropathy [2]. Nevertheless, the HMSN V classification may be appropriate in individuals in whom the peroneal atrophy and lower motor neuron involvement is predominant over spasticity [2]. Our patient had both LMN and UMN features, with LMN features dominating distally in extremities.

The original descriptions of HMSN V had onset in second decade, with autosomal dominant inheritance [2]. The course is typically insidiously progressive over many years. Although walking may be difficult owing to spasticity, many persons are never confined to

wheelchair. Some become wheelchair bound in the fourth decade or later [2]. Our patient had onset in early childhood which was slowly progressive without any significant family history. He was ambulatory at the age of 18 years.

Pathologic descriptions and electrophysiologic studies of nerve suggested an axonal peripheral nerve process in cases designated as HMSN V or complicated HSP [2]. However, Gemignani et al. [5] reported entire spectrum of pathological changes (Hypertrophic type, neuronal type, Spinal type) in four cases of peroneal muscular atrophy with hereditary spastic paraparesis. In our case nerve biopsy finding was consistent with axonal pathology, but nerve conduction studies revealed findings consistent with both axonal and demyelinating affection.

To conclude, our case had many interesting features. He had spastic quadripareisis with bilateral symmetrical distal wasting which fits into HMSN type V and this association is seen in only 5% of all cases of hereditary sensory motor neuropathy. Our patient can also be classified under “complicated HSP” in view of having spastic paraplegia with amyotrophy with evidence of peripheral neuropathy by NCS. The hereditary neuropathies and spastic paraplegias are groups of genetically diverse disorders classified according to their predominant clinical phenotype. This case further extends the considerable overlap between these clinical syndromes. Lastly our case under discussion also had marfanoid habitus. To the best of our knowledge association of marfanoid features with HMSN V or complicated HSP has not been mentioned in the literature so far.

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