HEREDITARY NEUROPATHY MISDIAGNOSED AS CHRONIC INFLAMMATORY DEMYELINATING POLYRADICULONEUROPATHY: A CASE SERIES.

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Introduction.

Hereditary and inflammatory neuropathies usually display different clinical and neurophysiological features, that allow to avoid misdiagnosis and administration of unnecessary therapies. However, the diagnosis of hereditary neuropathies may be overlooked especially in case of atypical features or when a family history is lacking. We will present a case series of patients with hereditary neuropathies misdiagnosed with CIDP, and discuss the most common pitfalls that led to the incorrect diagnosis.

Methods.

In the table below we describe the clinical, neurophysiological and genetic characteristics of our 10 patients (8 men, 2 women, mean age 46±13.7 years, mean disease duration 10.2 ± 10.9 years) and their response to immunomodulatory therapies.

PT	Symptoms and signs	Neurophysiology	CSF	Therapy	Response to tp	Diagnosis	Genetic
1 M 42y	distal hands paresthesias, difficulty in fine motor tasks, UL hyporeflexia, LL areflexia, pes cavus	demyelinating SM neuropathy	proteins 0.70 g/L; 2.8 WB/μL	IgIV	None	CMT1B	Mutation c.449-1g>cin (MPZ)
2 M 54y	distal hypoesthesia , stepping gait, distal hyposthenia, hypotrophy at 4 limbs, UL hyporeflexia, LL areflexia	demyelinating SM neuropathy Nerve US: mild increased CSA in median nerves and right tibial nerve	Negative	Oral prednisone	Transient benefit	CMT1A	Duplication PMP22
3 F 27y	UL distal paresthesias and hyposthenia	demyelinating SM neuropathy	Negative	IgIV	None	CMT1D	Mutation Arg381Cys (EGR2)
4 F 48y	distal severe sensory impairment, LL burning pain, wheel-chair bound, diffuse hyposthenia, areflexia	diffuse marked symmetrical demyelinating SM neuropathy Nerve US: diffuse increased CSA	Proteins 1.5 g/L	IgIV and duloxetine	Partial benefit	CMT1A	Duplication PMP22
5 M 50y	progressive hyposthenia at 4 limbs with bilateral stepping-ataxic gait, hypoesthesia below the knees, hyposthenia of intrinsic hand muscles, diffuse areflexia	severe demyelinating SM neuropathy Nerve US: diffuse and symmetric increase of CSA	Proteins 0.57 g/L oligoclonal IgG bands	Parenteral methyl-prednisolone	Moderate transient benefit	-	**Ongoing:** negative for PMP22, MPZ, EGR2, NEFL, GJB1
6 M 50y	slowly progressive distal hyposthenia at 4 limbs and gait instability, difficult in fine motor tasks and bilateral foot drop, distal hand hypotrophy, diffuse hyporeflexia, pes cavus	demyelinating SM polyneuropathy Nerve US: moderate increase of CSA in median nerves	Negative	Steroid therapy	Transient benefit	-	**Ongoing:** Negative for PMP22 duplication and connexin 32
7 M 50y	sub-acute tetraparesis: severe distal and proximal hyposthenia at 4 limbs, hands paresthesias, LL marked distal weakness, UP reduced reflexes, LL areflexia; bilateral pes cavus	diffuse severe demyelinating SM polyneuropathy. Nerve US: diffuse and symmetric increase of CSA, increased fascicules in cervical spinal roots and in brachial plexus	Proteins 3.37 g/L	IV high dose methyl-prednisolone (1 g/day for 5 days)	Moderate benefit	-	**Ongoing:** negative for PMP22 mutation/deletion/duplication
8 M 70y	bilateral stepping gait, inability to maintain the standing position on narrow basis, distal hypoesthesia, severe diffuse hypotrophy, LL areflexia, bilateral carpal tunnel syndrome, fasciculations at thigh and tongue	SM axonal-demyelinating neuropathy Nerve US: diffuse increased CSA in all nerve	Proteins 0.72 g/L	IgIV	Transient benefit	TTR amyloidosis	Val30Met mutation of the TTR gene
9 M 54y	history positive for foot drop, distal hyposthenia at the left leg, LL diffuse hypotrophy, LL hyporeflexia	diffuse demyelinating SM polyneuropathy with more severe involvement at entrapment sites Nerve US: increased CSA of ulnar nerves, left sciatic nerve, right tibial nerve and bilateral brachial plexus in the supraclavicular space	Proteins 0.70 g/L	IV steroids and PE	Partial benefit	HNPP	Deletion in the PMP22 gene
10 M 20y	stepping gait, left LL distal hyposthenia, hyporeflexia at the left leg, mild distal hypotrophy, pes cavus	LL motor axonal neuropathy, signs of chronic neurogenic damage, more severe distally at both legs	Proteins 1.15 g/L WBC 13.5	IgIV	Subjective benefit	-	**Ongoing:** negative for CMT and Kennedy disease; tests for dHMNP are still ongoing

Legend to the figure. SM = sensory-motor; UP= upper limbs; LL= lower limbs; US= ultrasound; IgIV: Intravenous Immunoglobulins.

Discussion and conclusions.

Several clinical and laboratory features can lead to a misdiagnosis in patients with sensory-motor progressive symptoms at 4 limbs, and the most common initial diagnosis is CIDP. The majority of mistakes came from misinterpretation of **neurophysiological data**, such as conduction velocities slowing degree and atypical distribution, or the presence of conduction blocks.

Another common diagnostic pitfall derived from **CSF analysis**, that in some cases showed elevated CSF proteins or the presence of oligoclonal bands. An absent **response to immunomodulatory therapies** should also represent a red flag, and lead to reconsider the diagnosis of CIDP. Response to immunomodulatory therapies, although incomplete may be more likely in patients with an overlap syndrome, as showed by 2 of our patients. In patients with atypical patterns, the use of more recent diagnostic tools, such as **nerve ultrasound and MR-neurography** is advisable in order to allow a correct diagnosis.

References.

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