Charcot-Marie-Tooth neuropathy misdiagnosed as chronic inflammatory demyelinating polyradiculoneuropathy

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The diagnosis of Charcot-Marie-Tooth (CMT) neuropathy is straightforward when the clinical and neurophysiological features are supported by a positive family history. However in sporadic cases misdiagnosis is common. We describe 6 patients (4 men, 2 women, mean age 53.8 ± 11.7 yrs) affected with genetically confirmed CMT who were initially diagnosed with chronic inflammatory demyelinating polyradiculoneuropathy (CIDP).

Patients and methods

6 patients
4 men, 2 women
mean age 53.8 ± 11.7 yrs

Neurophysiology → demyelinating features in 5/6 pts.

CSF analysis → 2/6 pts showed increased CSF proteins, and one had oligoclonal bands.

Nerve US (in 2 pts with demyelinating neuropathy) → diffuse increased cross-sectional area (CSA) with variable values within the same nerve.
MR-neurography (in one pt with demyelinating neuropathy) → diffuse nerve hyperthrophy

Therapy

Immunomodulatory therapies in 6/6 pts
3/5 pts with demyelinating features → IV immunoglobulins without benefit
2/5 pts were treated with steroids, with only temporary improvement in one of them.

One pt with axonal neuropathy and long progressive history →plasma exchange and IV immunoglobulins, without benefit.

Clinical and neurophysiological features

Lack of benefit from therapies

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Screening for hereditary neuropathies

5 pts with demyelinating neuropathy
Two CMT1A
One CMT1B
One CMT1D
In one pt genetic studies are still ongoing

The patient with axonal neuropathy was diagnosed with CMT2K.

In 2 pts an overlap syndrome (CMT-CIDP) was present.

Conclusions

Several clinical and laboratory features can lead to a misdiagnosis in CMT patients without family history, especially when CIDP-like patterns are present.
Some clinical findings may also be suggestive of overlap syndromes CMT-CIDP, such as acute or subacute deterioration, or proximal involvement. CSF protein elevation has also been described in CMT, but with high levels (>1 g/L) only in overlap syndromes.
Only one patient with features suggestive of an overlap syndrome, showed benefit after steroids.

As already reported in previous studies, refractoriness to immunomodulatory treatment represents a red flag, arising the suspicion of a possible hereditary neuropathy.