Charcot-Marie-Tooth syndrome associated with cerebello-extrapyramidal manifestations in a patient with tomaculous neuropathy

Sir,

The association of a Charcot-Marie-Tooth (CMT) syndrome with extrapyramidal symptom has rarely been reported [3]. A true tomaculous neuropathy as the background of a very complex clinical picture is not mentioned in the literature.

We describe a 57 year old male presenting: (1) a *chronic sensorimotor polyneuropathy* suggesting a CMT syndrome with late, painful and apparently monomelic onset; [2] an *extrapyramidal syndrome*: hypomimia, static tremor of the upper limbs of moderate amplitude and rapid rhythm (84/min), bilateral extrapyramidal-type rigidity, marked disability for fine movements in the upper limbs; extrapyramidal signs were: responsiveness to antiparkinsonian therapy (L-DOPA, bromocriptine); [3] a *neocerebellar syndrome*: intention tremor, asynergy in the finger-nose and heel-knee tests, oppositional spasms, illegible writing due to dyskinetic movements, dysarthric cerebellar speech, intermittent dysphagia for liquid and solid food with depression of the pharyngeal reflexes; [4] *depression of the cognitive functions*. EMG (tibial anterior muscle): partial denervation. MCV: left peroneal nerve = 30.0 m/s, right peroneal nerve = 35.2 m/s, left median nerve = 50.0 m/s.

Sural nerve biopsy: tomaculous neuropathy: 24% of the teased fibers bearing tomaculae sized 84-32/20-32 μ m against the background of a demyelinating process (56% of the teased fibers) associated with a mild myelin fiber loss (6353 fb/mm²) and an active regeneration (24% of the teased fibers). *Gastrocnemius muscle biopsy*: neurogenic changes.

The initial clinical supposition of a Guam syndrome was excluded by the peripheral neuropathic type of the involvement, electrophysiologically and morphologically confirmed.

Paretic episodes were absent in our patient and his family history, as in other tomaculous neuropathy cases in the literature [1, 2, 4, 5].

Such cases might support the nonspecificity of tomaculous changes. They might also represent infraclinical cases of recurrent familial neuropathy with liability to pressure palsies with a more or less complex clinical picture.

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