TUBULAR AGGREGATE MYOPATHY RESEMBLING A LOWER MOTOR NEURON SYNDROME

Silvia Rota, Fabrizio Rinaldi, Elisa Pari, Alice Todeschini, Irene Volonghi, Paolo Costa, Alessandro Padovani, Massimiliano Filosto

Clinical Neurology, Section for Neuromuscular Diseases and Neuropathies
University Hospital "Spedali Civili", Brescia

Objectives

To describe a tubular aggregate myopathy clinically resembling a lower motor neuron syndrome.

Patient and methods

A 76-year-old man started to experience muscle weakness, intolerance to exercise and rest muscle cramps at lower limbs since his 75 years.

Neurological examination showed atrophy of the right first interosseous and thenar eminence muscles, occasional calf fasciculations and absent ankle jerk reflexes. Electromyography and muscle biopsy were performed.

Results

Electromyography showed diffuse chronic neurogenic abnormalities and isolated fasciculations in the left anterior tibialis muscle.

Muscle biopsy revealed myopathic abnnormalities (i.e. fiber diameter variability, internal nuclei, scattered atrophic fibers), rare nuclear morulae and, mainly, multiple fibers with cytoplasmic areas which were basophilic on hematoxylin-eosin staining, reddish on Modified Gomori's trichrome and intensely blue on NADH, compatible with tubular aggregates. No frank neurogenic atrophy was detected.

Discussion

Tubular aggregates can be observed in primary forms of tubular aggregate myopathy (TAM) as well as in muscle biopsy from various conditions such as myasthenic syndromes, inflammatory myopathies, myotonic syndromes and myopathies induced by alcohol and drugs. Mutations in the STIM1 and ORAI1 genes were detected in some families with TAM.

Clinical presentation of the primary forms is somewhat variable, ranging from mild pictures (i.e. iperCKemia, exercise induced myalgia) to moderate limb-girdle muscle weakness, to more severe phenotypes with early onset, ophthalmoparesis, muscle contractures and respiratory insufficiency.

Our patient exhibits an unusual late-onset phenotype. Beyond exercise intolerance and muscle cramps, he presents mild asymmetrical distal muscle atrophy in upper limbs and diffuse chronic neurogenic changes at the electromyographic study, resembling a slowly progressive lower motor neuron syndrome.

Conclusions

Clinical spectrum of TAM is wide and may include a mild late-onset distal muscle involvement. A strict follow-up is mandatory to assess the evolution of the disease.

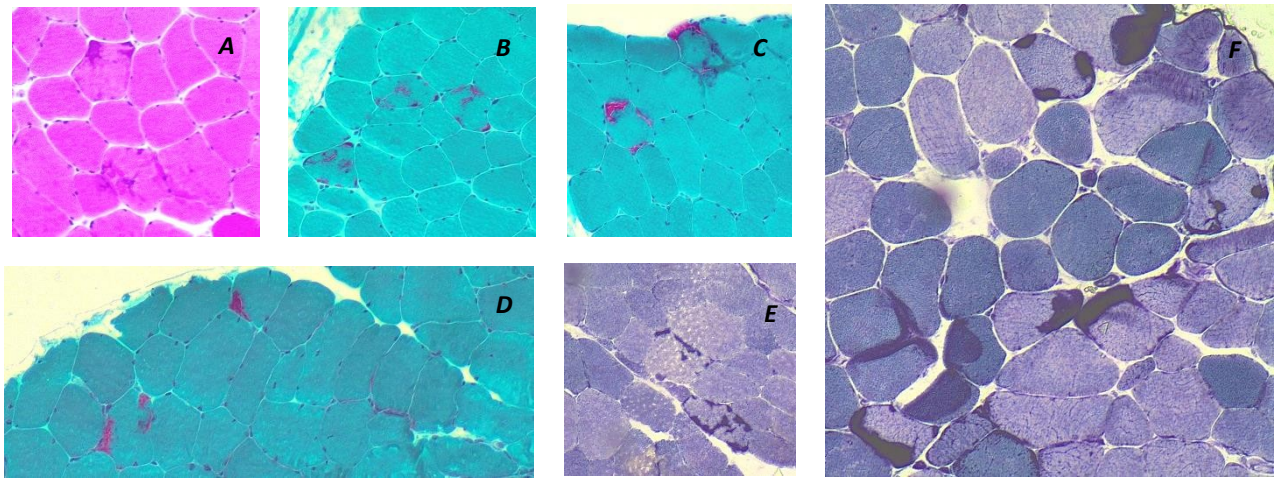

Fig. 1: Tubular aggregates as observed by different histological staining. A: Hematoxylin-Eosin staining; B, C and D:Gömöri trichrome stain; E and F: NADH staining