IMAGEM EM NEUROLOGIA/IMAGE IN NEUROLOGY

Facial Myorhythmia: A Clinical Sign of Whipple's Disease

Miorritmia Facial: Um Sinal Clínico da Doença de Whipple

André Costa [1,*]; Joana Pona-Ferreira [1]; Ana-Graça Velon [1]

1-Neurology Department, Local Health Unit of Trás-os-Montes and Alto Douro, Vila Real, Portugal

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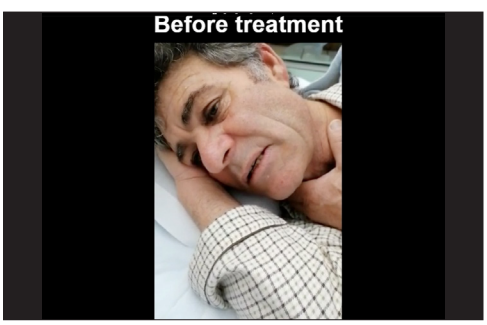
A 60-year-old man with a 6-month history of cognitive dysfunction marked by attentional and executive deficits, along with anhedonia, hypersomnia, and paroxysmal facio-cervical involuntary movements, was admitted to a neurology department for diagnostic investigation. He had a 10-year history of unexplained joint pain and weight loss monitored by an autoimmune clinic. He was on a statin for dyslipidemia and had a 40-pack-year smoking history. Neurologic examination revealed cognitive impairment affecting memory and attention function (Mini-Mental State Examination: 26/30), and a vertical supranuclear gaze palsy. No signs of palatal abnormal movements, ataxia or parkinsonism were observed. The involuntary movements were consistent with myorhythmia involving the left facial and cervical muscles (see video). Laboratory analyses excluded common infectious and nutritional causes of cognitive impairment. A serum analysis was also performed to assess autoimmune conditions, and the results showed no significant findings. Computed tomography (CT) of the chest, abdomen, and pelvis, along with serum onconeuronal antibodies, ruled out paraneoplastic causes. Electroencephalography was normal, and brain magnetic resonance imaging (MRI) showed nonspecific findings of cerebral small vessel disease. Cerebrospinal fluid (CSF) analysis showed no pleocytosis, proteins 0.64 g/L (N: <0.45 g/L) and microbiologic studies, including *Mycobacterium tuberculosis* and *Cryptococcus neoformans*, were negative. CSF neuronal surface antibodies against the NMDA receptor, LGI1, CASPR2, as well as serum IgLON5, were also negative. Given the possibility of rare infectious causes of cognitive impairment and the patient's medical history, CSF PCR testing for *Tropheryma whipplei* DNA was conducted and returned positive. A gastroenterology consult led to endoscopy with duodenal biopsies for histology and PCR, which detected *Tropheryma whipplei* DNA. Suspecting Whipple's disease (WD) with central nervous system (CNS) involvement, intravenous ceftriaxone (2 g daily for 2 weeks) was administered, followed by oral sulfamethoxazole-trimethoprim (960 mg twice daily for 1 year), resulting in a significant improvement in involuntary movements at 1-year follow-up (see video). Neuropsychological assessment at that time showed the presence of amnestic mild cognitive impairment. CSF PCR testing for *Tropheryma whipplei* DNA was repeated 18 months after diagnosis



Video. On admission, the patient exhibits asymmetric, slow, rhythmic facio-cervical involuntary movements consistent with myorhythmia. At 1-year follow-up after antibiotic therapy, both the movements and vertical supranuclear gaze palsy had resolved. (see the video)

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***Autor Correspondente / Corresponding Author:**
André Jorge Assunção Costa
Local Health Unit of Trás-os-Montes and Alto Douro
Avenida da Noruega;
5000-508 Lordelo, Vila Real, Portugal
andre.ja.costa@hotmail.com

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and returned negative, indicating biological cure. The patient attends neurological evaluations every six months, and at the 2-year follow-up, there is no evidence of progressive cognitive decline or additional neurological symptoms. After the gastrointestinal tract, the brain represents the second primary site affected in WD, a rare multisystemic infection. A prodromal stage characterized by joint symptoms often precedes the classic manifestations of WD. Neurological manifestations include cognitive impairment, neuropsychiatric syndromes, oculomotor disturbances and movement disorders. The latter are common, representing approximately half of CNS-WD syndromes, and may be pathognomonic for Whipple's disease.[1,2] In this patient, myorhythmia was unilateral and occurred without ocular involvement, differing from the classical oculo-masticatory myorhythmia. Facial myorhythmia should be differentiated from other rhythmic facial movement disorders, including hemifacial spasm, tic disorders, myoclonus, dystonia and psychogenic movements.[3] It is thought to result from dysfunction of brainstem oscillatory networks, particularly the dentato-rubro-olivary system. This case highlights the importance of recognizing myorhythmia through careful phenomenological assessment of paroxysmal facial involuntary movements, as well as its clinical value in raising suspicion for Whipple's disease, particularly when associated with unexplained cognitive decline, supranuclear gaze palsy, weight loss, and arthralgias, enabling timely diagnosis and treatment.[4] ■

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