Abnormal tongue features as a clinical clue for late-onset Pompe’s disease

Wladimir Bocca Vieira de Rezende Pinto,1 Paulo Victor Sgobbi de Souza,1 Thiago Bortholin,1 Fernando George Monteiro Naylor,1 Acary Souza Bulle Oliveira

A 58-year-old woman presented with slowly-progressive lower limb weakness. Medical history disclosed a six-year history of obstructive sleep apnea syndrome (OSAS). Examination disclosed abnormal tongue features (Figure 1) and proximal flaccid tetraparesis. Muscle MRI showed marked compromise of the adductor magnus, and muscle biopsy disclosed vacuolar myopathy with PAS-positive vacuoles (Figure 2). Dried blood spot-based GAA (acid alpha-glucosidase) activity testing and GAA gene sequencing confirmed late-onset Pompe’s disease (LOPD). Clinicians should consider LOPD in cases of limb-girdle weakness with atypical findings1, such as obstructive sleep apnea syndrome, pulmonary hypertension, axial involvement with myotonic or complex repetitive discharges and tongue weakness with fatty infiltration2.

1Universidade Federal de São Paulo, Departamento de Neurologia e Neurocirurgia, Divisão de Doenças Neuromuscular, São Paulo SP, Brasil.

Correspondence: Wladimir Bocca Vieira de Rezende Pinto; Departamento de Neurologia e Neurocirurgia da UNIFESP; Rua Estado de Israel, 899; 04022-002 São Paulo SP, Brasil; E-mail: wladimirbvrpinto@gmail.com

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Figure 2. Muscle MRI and muscle biopsy findings in LOPD. (A) Right thigh muscle MRI showed marked compromise of the adductor magnus and mild involvement of the vastus medialis and vastus intermedius muscles. (B–D) Deltoid muscle biopsy showing vacuolar myopathy (black arrow-head) with PAS-positive vacuoles (black arrow).

References
