

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

Achado anormal na língua como dica clínica para doença de Pompe de início tardio

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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.

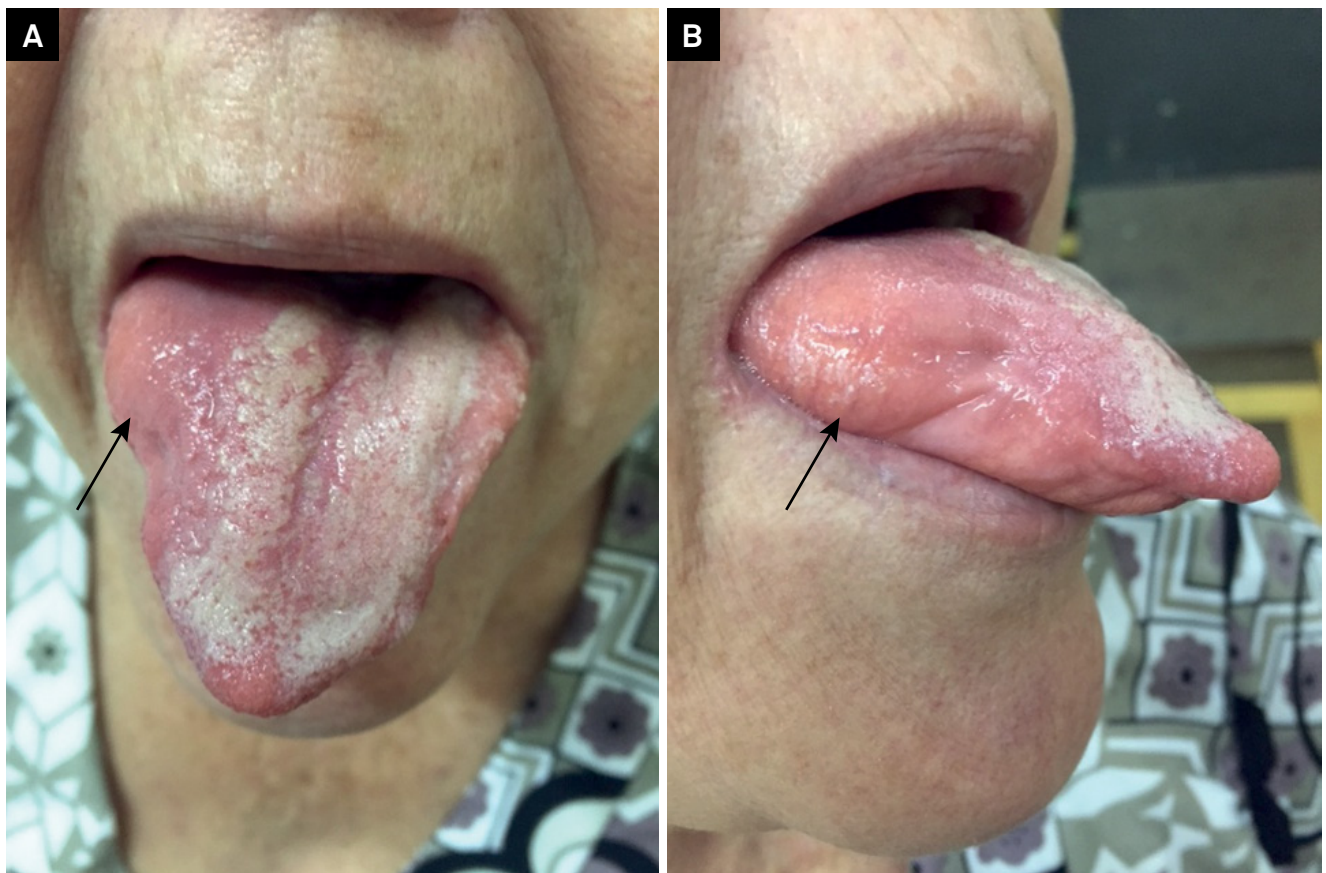


Figure 1. Abnormal tongue morphology in LOPD. (A–B) Diffuse tongue atrophy and abnormal fatty replacement of the tongue musculature resembling a “tumor-like” structure in the right side of the tongue (white arrow).

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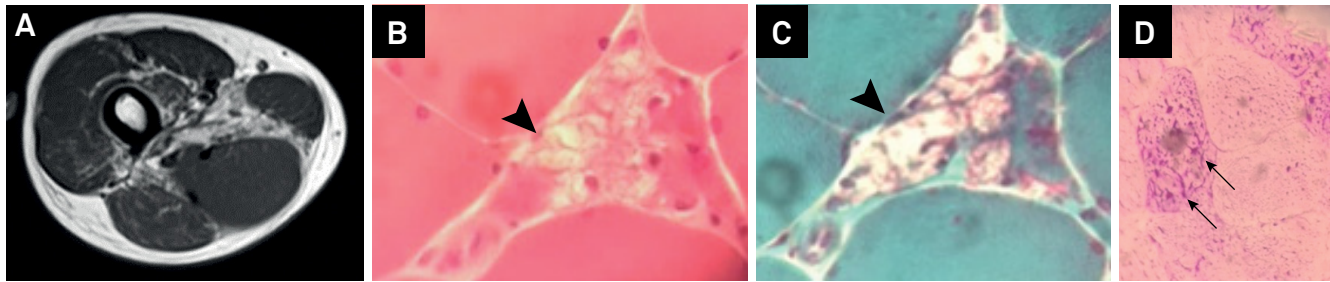


Figure 2. Muscle MRI and muscle biopsy findings in LOPD. (A) Right thigh muscle MRI showed marked compromise of the adductor magnus and mild involvement of the vastus medialis and vastus intermedius muscles. (B–D) Deltoid muscle biopsy showing vacuolar myopathy (black arrow-head) with PAS-positive vacuoles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

References

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