Teaching Video *NeuroImages*: Bent spine syndrome as an early presentation of late-onset Pompe disease

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Bent spine syndrome (BSS), an abnormal trunk ante-flexion of at least 45° that increases during walking and abates in supine position, is described in parkinsonism and myopathies.¹ We report a 56-year-old man with genetically proven late-onset Pompe disease (LOPD) showing BSS (figure 1 and video at Neurology.org). He complained of axial weakness that progressively forced him to walk stretching his arms backward since his 20s. Whole-body muscle MRI revealed severe fatty replacement of lumbar paraspinal (figure 2) and iliopsoas muscles, and spirometry showed restrictive ventilatory defect. Periodic acid-Schiff–positive vacuoles and reduced acid α -glucosidase activity were demonstrated in muscle biopsy. While lumbar hyperlordosis is a typical feature of LOPD, other trunk abnormalities may appear early in the disease. LOPD must be included in the differential diagnosis of BSS.²

AUTHOR CONTRIBUTIONS

Francesca Magrinelli: study concept and design, acquisition, analysis, and interpretation of data, draft of manuscript. Michele Tosi: acquisition of data, critical revision of manuscript for intellectual content. Paola Tonin: study concept and design, analysis and interpretation of data, critical revision of manuscript for intellectual content.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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2. Taisne N, Desnuelle C, Juntas Morales R, et al. Bent spine syndrome as the initial symptom of late-onset Pompe disease. *Muscle Nerve* Epub 2016 Nov 15.

Figure 1 Clinical photographs

Clinical photographs of the patient with bent spine syndrome (A), which is markedly improved on supine position (B).

Supplemental data
at Neurology.org

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Figure 2 Sagittal T1-weighted MRI of the lumbar spine shows severe fatty replacement of lumbar paraspinal muscles

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