Bent spine syndrome as an early presentation of late-onset Pompe disease

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Title character count: 72

Number of references: 2

Number of figures: 2

Number of videos: 1

Word count paper: 120

Supplemental Data: Figure 1, Figure 2, Video, Video Legend, Teaching Slides, and Patient Consent Form.

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Search Terms:

[185] Muscle disease; [16] Clinical neurology examination; [91] All Genetics; [120] MRI

Author Contributions:

Francesca Magrinelli (Corresponding Author): 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.

Acknowledgements:

None.

Disclosures Statement:

Dr. Francesca Magrinelli reports no disclosures. Dr. Michele Tosi reports no disclosures. Dr. Paola Tonin reports no disclosures.

Bent spine syndrome (BSS), an abnormal trunk anteroflexion of at least 45° which 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 1). 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 alpha-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]

References

1. Azher SN, Jankovic J. Camptocormia: pathogenesis, classification, and response to therapy. Neurology 2005;65(3):355-359.
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 Legends

Figure 1: Clinical photographs of the patient with BSS (A), which is markedly improved on supine position (B).

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