X-linked Emery-Dreifuss muscular dystrophy manifesting with adult onset axial weakness, camptocormia, and minimal joint contractures.

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Abstract
Emery-Dreifuss muscular dystrophy is an early-onset, slowly progressive myopathy characterized by the development of multiple contractures, muscle weakness and cardiac dysfunction. We present here the case of a 65-year-old male patient with a 20 year history of slowly progressive camptocormia, bradycardia and shortness of breath. Examination showed severe spine extensor and neck flexor muscle weakness with slight upper limb proximal weakness. Cardiologic assessment revealed slow atrial fibrillation. Whole body MRI demonstrated adipose substitution of the paravertebral, limb girdle and peroneal muscles as well as the tongue. Emerin immunohistochemistry on patient muscle biopsy revealed the absence of nuclear envelope labeling confirmed by Western Blot. Genetic analysis showed a hemizygous duplication of 5 bases in exon 6 of the EMD, emerin, gene on the X chromosome. This is an unusual presentation of X-linked Emery-Dreifuss muscular dystrophy with adult onset, predominant axial muscles involvement and minimal joint contractures. Diagnosis was prompted by the analysis of emerin on muscle biopsy.

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KEYWORDS: EMD gene; Emerin; Emerin immunohistochemistry; Emery–Dreifuss muscular dystrophy; Muscle biopsy; Muscular dystrophy; Whole body MRI

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