Perforating palmar disease in TTR-related familial amyloid polyneuropathy
Mal perfurante palmar na polineuropatia amiloidótica familiar ligada à TTR

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A 60-year-old retired woman presented with upper and lower limb paresthesias since age 40 and chronic diarrhea and weakness. She worked as a teacher. Her deceased mother had experienced similar symptoms. Neurological examination disclosed dysautonomic features (Figure 1), severe sensory loss of vibration and proprioceptive senses and a moderate decrease of pain and temperature sensation with stocking-glove distribution, and perforating palmar disease (Figure 2). Neurophysiological studies disclosed severe axonal sensorimotor polyneuropathy.

Laboratory tests were unremarkable. The pathogenic heterozygous variant Val50Met in the TTR gene defined TTR-related familial amyloid polyneuropathy. This finding highlights perforating palmar disease as a rare complication of familial amyloid polyneuropathy. Neurogenic perforating palmar ulcers may occur in neuropathies due to: (i) severe distal sensory loss and high risk of multiple local microtrauma; (ii) vasomotor disturbances to peripheral dysautonomia and sympathetic dysfunction with chronic hypoperfusion¹.

References


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Conflict of interests: There is no conflict of interest to declare.
Received 03 January 2018; Received in final form 09 April 2018; Accepted 11 April 2018.