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Atypical Familial Amyotrophic Lateral Sclerosis Secondary to Superoxide Dismutase 1 Gene Mutation With Coexistent Axonal Polyneuropathy: A Challenging Diagnosis

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Abstract : Amyotrophic lateral sclerosis (ALS), also known as Lou Gehrig's disease, is a neurodegenerative disease that involves both the upper and lower motor neurons. Familial ALS, including superoxide dismutase 1 (SOD1) mutation, accounts for 5-10% of all cases of ALS. Typically, the symptoms of ALS are purely motor, though coexistent sensory symptoms have been reported in rare cases. In this report, we describe the case of a 47- year-old man who presented with progressive bilateral lower limb weakness and numbness for the last four years. A nerve conduction study (NCS) showed evidence of coexistent axonal sensorimotor polyneuropathy in addition to the typical findings of ALS in needle electromyography. Genetic testing confirmed the diagnosis of familial ALS secondary to the SOD1 genetic mutation. This report highlights that the presence of sensory symptoms should not exclude the possibility of ALS in an appropriate clinical setting.

Keywords : Saudi Arabia, polyneuropathy, SOD1 gene mutation, familial amyotrophic lateral sclerosis, amyotrophic lateral sclerosis
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