Teaching NeuroImages: MR neurography for the diagnosis of hypertrophic neuropathies

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Figure Magnetic resonance neurography study of the lumbosacral plexus

Neuroimaging study discloses marked enlargement of peripheral nerve, spinal roots, and lumbosacral plexus (white arrow) in coronal gadolinium-enhanced T1-weighted images (A) and short tau inversion recovery sequences (B) and axial T2-weighted (C) and T1-weighted images (D).

A 25-year-old woman presented with a 10-year history of frequent falls and deafness. Her mother had a similar neurologic picture. Examination showed peroneal amyotrophy, pes cavus, and hearing loss. Magnetic resonance (MR) neurography showed diffuse nerve enlargement in the lower limbs (figure). Genetic analysis revealed heterozygous mutation c.82T>C (p.Trp28Arg) in the *PMP22* gene, defining diagnosis of Charcot-Marie-Tooth disease (CMT) type 1A.

The spectrum of disorders with nerve enlargement has widened as imaging studies improved detection of spinal root and plexus enlargement.^{1,2} MR neurography aids in the diagnosis of conditions like CMT, Refsum disease, chronic inflammatory demyelinating polyneuropathy, neurofibromatosis, and neurolymphomatosis.^{1,2}

AUTHOR CONTRIBUTIONS

Dr. de Souza: case report project conception, organization, execution; writing of the first draft. Dr. Bortholin: case report project conception, organization, execution; writing of the first draft. Dr. Naylor: case report project execution; manuscript review and critique. Dr. Pinto: case report project conception; writing of the first draft, manuscript review and critique. Dr. Oliveira: manuscript review and critique.

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