A 21-year-old man undergoing workup for surgical treatment of scoliosis was referred for evaluation of possible neurofibromatosis due to abnormal imaging studies of the spine. No evidence of neurofibromatosis was found on examination; rather, the patient had prominent foot deformities, hypertrophic peripheral nerves, and signs consistent with a hereditary motor and sensory polyneuropathy. MRI of the spine obtained as part of his surgical workup revealed diffusely enlarged nerve roots (figure, A and B). Electrodagnostic studies showed profound slowing of conduction velocities. DNA analysis identified duplication of the PMP22 gene confirming the diagnosis of Charcot-Marie-Tooth disease (CMT) type 1A, which is a hereditary form of demyelinating polyneuropathy. Our case illustrates that severe nerve root hypertrophy may be associated with CMT type 1A.1,2

Nerve root hypertrophy in CMT type 1A
Jenny P. Liao and Andrew J. Waclawik
Neurology 2004;62:783
DOI 10.1212/01.WNL.0000103234.32489.CA

This information is current as of March 8, 2004