

E-Pearl of the Week

Brought to you by the Resident and Fellow Section of the Journal *Neurology*®.

June 10, 2009

Genetic testing in inherited demyelinating polyneuropathy

Charcot-Marie-Tooth type 1A (CMT1A) accounts for *more than half* of inherited demyelinating polyneuropathies. CMT1A is autosomal dominant, but about one-fourth of patients present with a de novo mutation and thus have no family history. CMT1A typically presents with foot deformity, such as high arches and hammer toes, and distal weakness and sensory loss, without positive neuropathic sensory symptoms. CMT1A is caused by a duplication of the PMP-22 gene. Many experts recommend *first* testing only for the PMP-22 duplication in patients with an inherited demyelinating polyneuropathy. If this test is negative, there are comprehensive diagnostic panels that test for the many other, less common, mutations that can cause inherited polyneuropathy.

Submitted by Ted Burns, MD

Disclosure: Dr. Burns receives a stipend as Podcast Editor for Neurology®, performs EMG studies in his neuromuscular practice (30% effort), and received compensation for a presentation on MG-QOL15, given to study investigators of eculizumab in myasthenia gravis.