Hereditary Neuropathy with Liability to Pressure Palsies

Genetics

-Gene: PMP22 (Peripheral myelin protein 22, 17p.11.2); PMP22 del vs. PMP22 dup in CMT1a

Clinical findings/Dysmorphic features

-Repeated focal pressure neuropathies (i.e. carpal tunnel syndrome, peroneal palsy with foot drop); recovery from acute neuropathy often complete; if not complete, disability usually mild

-Some affected individuals also have signs of a mild to moderate peripheral neuropathy

-Mild to moderate pes cavus deformity

-First attack usually in the second or third decade

Etiology

-2-5 per 100,000

Pathogenesis

-Decreased PMP22 mRNA and decreased peripheral myelin protein 22 in peripheral nerve cells

Genetic testing/diagnosis

-PMP22 sequencing (20%), 1.5-Mb PMP22 deletion (80%)