

Abstract

Autosomal recessive Charcot-Marie-Tooth diseases, relatively common in Algeria due to high prevalence of consanguineous marriages, are clinically and genetically heterogeneous. We report on two consanguineous families with demyelinating autosomal recessive Charcot-Marie-Tooth disease (CMT4) associated with novel homozygous mutations in the MTMR2 gene, c.331dupA (p.Arg111LysfsX24) and PRX gene, c.1090C>T (p.Arg364X) respectively, and peculiar clinical phenotypes. The three patients with MTMR2 mutations (CMT4B1 family) had a typical phenotype of severe early onset motor and sensory neuropathy with typical focally folded myelin on nerve biopsy. Associated clinical features included vocal cord paresis, prominent chest deformities and claw hands. Contrasting with the classical presentation of CMT4F (early-onset Dejerine-Sottas phenotype), the four patients with PRX mutations (CMT4F family) had essentially a late age of onset and a protracted and relatively benign evolution, although they presented marked spine deformities. These observations broaden the spectrum of clinical phenotypes associated with these two CMT4 forms