Abstract

By sequencing of the FGD4 coding sequence in a cohort of 101 patients affected by autosomal recessive demyelinating Charcot-Marie-Tooth disease (CMT), we have identified two novel missense mutations inFGD4 in two patients from consanguineous descent: p.Arg442His in an Algerian patient and p.Met566Ile in a Lebanese girl. The patients present early onset, slowly progressive CMT, with drastic reduction of nerve conduction velocities. These mutations are the second and third missense mutations characterized in FGD4. They are likely to lead to conformational changes in the PH1 and FYVE domains