**Case report 3**

A follow-up of a 20-year-old female previously reported when she was eight years old (4). She started to walk at the age of 20 months. Some progressive walking difficulties were observed. At the time of her first report, she presented with generalised flaccid paresis of her limbs, scoliosis, absent knee and ankle reflexes, as well as waddling gait. Her nerve conduction studies revealed sensory-motor demyelinating neuropathy with conduction velocity (cv) = 20 m/s in the motor median nerve, as well as pronounced reinnervation in a needle EMG. Some mild clinical symptoms (absent knee and ankle reflex, pes cavus) and sensory-motor demyelinating neuropathy with cv= 20.5 m/s in the motor median nerve were presented in the patient’s father as well. The SMN1 gene mutation and the PMP22 duplication were confirmed in the family (Pedigree 3). Our proband lost ambulation when she was eight years old. She underwent a feet deformity surgery; scoliosis was managed with growing rods. Currently, she presents with significant weakness of her lower limbs and proximal part of her upper limbs. The patient wheels and transfers herself independently. An effective use of the distal part of her arms is preserved with mild degree of decrease in her muscle strength. Her reflexes are absent and some significant contractures in her lower limbs are observed. Mild tremors of fingers and tongue are noticeable. The patient’s most recent nerve conduction studies results are comparable with her previous evaluation of 2006 (Fig. 5).