Occurrence of Andermann Syndrome out of French Canada – Agenesis of the Corpus Callosum with Neuropathy

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In 1987, we described (2) a 6-year-old male patient suffering from spastic tetraparesis, striking muscular hypotonia, hypotonia and distal hypotrophy of all four limbs, absence of tendon reflexes, strabismus convergens, bilateral ptosis, scoliosis, moderate mental retardation (IQ = 67 on the Stanford Binet Scale) and total absence of the corpus callosum (CT). Neurophysiological examination revealed signs of denervation (electromyogram), reduced motor nerve conduction velocity (external sciatic-popliteal nerve 24 m/s, median nerve 41.5 m/s) with normal brainstem auditory evoked potentials and visual evoked potentials. Neuropathological findings consisted of reduced myelinated fibres (peroneal nerve) and signs of chronic denervation and reinnervation (quadriceps femoris muscle). The parents were healthy and non-consanguineous; they came from the Veneto region of Italy. The clinical, neurophysiological and neuroradiological findings in our case report are consistent with the conditions described by Erratum

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