Abstract "SPG 30 - eine neue autosomal rezessive Form der hereditären spastischen Spinalparalyse"

SPG30

The hereditary spastic paraplegias (HSPs) are a clinically and genetically heterogeneous group of neurodegenerative diseases characterised by progressive spasticity in the lower limbs.
Twenty nine different loci (SPG) have been mapped so far, and 11 responsible genes have been identified. Clinically one distinguishes between pure and complex HSP forms which are variably associated with numerous combinations of neurological and extra-neurological signs. Less is known about autosomal recessive forms (ARHSP) since the mapped loci have been identified in few, often single, families and account for only few percent of patients.
We report a new ARHSP locus (SPG30) on chromosome 2q37.3 in a consanguineous family of Algerian origin living in Eastern France with a significant multipoint lod score of 3.8. Ten other families were not linked to the newly identified locus demonstrating further genetic heterogeneity. The phenotype of the linked family consists of spastic paraparesis and peripheral neuropathy associated with slight cerebellar signs confirmed by cerebellar atrophy on one CT scan.