Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome. ORPHA:313772

Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome is a rare hereditary spastic ataxia disorder characterized by childhood onset of slowly progressive lower limb spastic paraparesis and cerebellar ataxia (with dysarthria, swallowing difficulties, motor degeneration), associated with sensorimotor neuropathy (including muscle weakness and distal amyotrophy in lower extremities) and progressive myoclonic epilepsy. Ocular signs (ptosis, oculomotor apraxia), dysmetria, dysdiadochokinesia, dystonic movements and myoclonus may also be associated.