DISEASE:
Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome

NAME:	Early-onset spastic ataxia-myoclonic epilepsy-neuropathy syndrome
DESCRIPTION:	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.
ORPHACODE:	313772
SYNONYMS:	AFG3L2-related spastic ataxia-myoclonic epilepsy-neuropathy syndrome Autosomal recessive spastic ataxia type 5 SPAX5
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	AFG3L2
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