DISEASE:
Autosomal recessive cerebellar ataxia due to STUB1 deficiency

NAME:	Autosomal recessive cerebellar ataxia due to STUB1 deficiency
DESCRIPTION:	A rare hereditary ataxia characterized by progressive truncal and limb ataxia resulting in gait instability. Dysarthria, dysphagia, nystagmus, spasticity of the lower limbs, mild peripheral sensory neuropathy, cognitive impairment and accelerated ageing have also been associated.
ORPHACODE:	412057
SYNONYMS:	SCAR16 Spinocerebellar ataxia autosomal recessive type 16
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	STUB1
CREATED:	13 May 2019 - 01:02
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