DISEASE:
Spectrin-associated autosomal recessive cerebellar ataxia

NAME:	Spectrin-associated autosomal recessive cerebellar ataxia
DESCRIPTION:	Spectrin-associated autosomal recessive cerebellar ataxia is a rare, genetic neurological disease, due to SPTBN2 mutations, characterized by global development delay in infancy, followed by childhood-onset gait ataxia with limb dysmetria and dysdiadochokinesia, mild to severe intellectual disability, development of cerebellar atrophy, and abnormal eye movements (including a convergent squint, hypometric saccades, jerky pursuit movements and incomplete range of movement).
ORPHACODE:	352403
SYNONYMS:	Autosomal recessive spinocerebellar ataxia type 14 Infantile-onset spinocerebellar ataxia-psychomotor delay syndrome SCAR14 SPARCA SPARCA1 Spectrin-associated autosomal recessive cerebellar ataxia type 1
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SPTBN2
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