DISEASE:
Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency

NAME:	Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency
DESCRIPTION:	A rare autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome characterized by early-childhood onset of cerebellar ataxia associated with generalized tonic-clonic epilepsy and psychomotor development delay, dysarthria, gaze-evoked nystagmus and learning disability. Other features in some patients include upper motor neuron signs with leg spasticity and extensor plantar responses, and mild cerebellar atrophy on brain MRI.
ORPHACODE:	284282
SYNONYMS:	Autosomal recessive spinocerebellar ataxia type 12 SCAR12
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	WWOX
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