DISEASE:
Focal epilepsy-intellectual disability-cerebro-cerebellar malformation

NAME:	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation
DESCRIPTION:	Focal epilepsy-intellectual disability-cerebro-cerebellar malformation is a rare, genetic neurological disorder characterized by early infantile-onset of seizures, borderline to moderate intellectual disability, cerebellar features including dysarthria and ataxia and cerebellar atrophy and cortical thickening observed on MRI imaging. Seizures are typically focal (with prominent eye blinking, facial and limb jerking), precipitated by fever and often commence with an oral sensory aura (anesthetized tongue sensation). When not properly controlled by anti-epileptic medication, weekly frequency and persistance into adult life is observed.
ORPHACODE:	352587
SYNONYMS:	Focal epilepsy-intellectual disability-dysarthria-ataxia syndrome
XREF(S):	Orphanet
ANALYTE(S):	TBC1D24
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