DISEASE:
Epileptic encephalopathy with global cerebral demyelination

NAME:	Epileptic encephalopathy with global cerebral demyelination
DESCRIPTION:	Epileptic encephalopathy with global cerebral demyelination is a rare mitochondrial substrate carrier disorder characterized by severe muscular hypotonia, seizures (with or without episodic apnea) beginning in the first year of life, and arrested psychomotor development (affecting mainly motor skills). Severe spasticity with hyperreflexia has also been reported. Global cerebral hypomyelination is a characteristic imaging feature of this disease.
ORPHACODE:	353217
SYNONYMS:	AGC1 deficiency Mitochondrial aspartate-glutamate carrier 1 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	SLC25A12
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