DISEASE:
Generalized epilepsy-paroxysmal dyskinesia syndrome

NAME:	Generalized epilepsy-paroxysmal dyskinesia syndrome
DESCRIPTION:	Generalized epilepsy-paroxysmal dyskinesia syndrome is characterised by the association of paroxysmal dyskinesia and generalised epilepsy (usually absence or generalised tonic-clonic seizures) in the same individual or family. The prevalence is unknown. Analysis in one of the reported families led to the identification of a causative mutation in the KCNMA1 gene (chromosome 10q22), encoding the alpha subunit of the BK channel. Transmission is autosomal dominant.
ORPHACODE:	79137
SYNONYMS:	GEPD
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	KCNMA1
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