Generalized epilepsy-paroxysmal dyskinesia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Generalized epilepsy-paroxysmal dyskinesia syndrome. ORPHA:79137

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.