
DISEASE:
KCNQ2-related epileptic encephalopathy

NAME:	KCNQ2-related epileptic encephalopathy
DESCRIPTION:	KCNQ2-related epileptic encephalopathy is a severe form of neonatal epilepsy that usually manifests in newborns during the first week of life with seizures (that affect alternatively both sides of the body), often accompanied by clonic jerking or more complex motor behavior, as well as signs of encephalopathy such as diffuse hypotonia, limb spasticity, lack of visual fixation and tracking and mild to moderate intellectual deficiency. The severity can range from controlled to intractable seizures and mild/moderate to severe intellectual disability.
ORPHACODE:	439218
SYNONYMS:	KCNQ2-NEE KCNQ2-related neonatal epileptic encephalopathy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	KCNQ2
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