DISEASE:
Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome

NAME:	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome
DESCRIPTION:	Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome is a rare, genetic, neurologic disease characterized by congenital microcephaly, severe, early-onset epileptic encephalopathy (manifesting as intractable, myoclonic and/or tonic-clonic seizures), permanent, neonatal, insulin-dependent diabetes mellitus, and severe global developmental delay. Muscular hypotonia, skeletal abnormalities, feeding difficulties, and dysmorphic facial features (including narrow forehead, anteverted nares, small mouth with deep philtrum, tented upper lip vermilion) are frequently associated. Brain MRI reveals cerebral atrophy with cortical gyral simplification and aplasia/hypoplasia of the corpus callosum.
ORPHACODE:	306558
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	IER3IP1 YIPF5
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