DISEASE:
Severe neonatal-onset encephalopathy with microcephaly

NAME:	Severe neonatal-onset encephalopathy with microcephaly
DESCRIPTION:	Severe neonatal-onset encephalopathy with microcephaly is a rare monogenic disease with epilepsy characterized by neonatal-onset encephalopathy, microcephaly, severe developmental delay or absent development, breathing abnormalities (including central hypoventilation and/or respiratory insufficiency), intractable seizures, abnormal muscle tone and involuntary movements. Early death is usual.
ORPHACODE:	209370
SYNONYMS:	Severe congenital encephalopathy due to MECP2 mutation
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MECP2
CREATED:	13 May 2019 - 01:02
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