Opis choroby *

Definicja

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.

Dane

Klasyfikacja
Choroba

Synonimy
Severe congenital encephalopathy due to MECP2 mutation
Ciężka wrodzona encefalopatia z powodu mutacji MECP2

Kod ORPHA
209370

Kod OMIM
300673

Kod ICD10
Q02

Kod ICD11
LD20.2

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*Źródło

orphanet