

Opis choroby *

Definicja

A rare genetic neurological disorder characterized by a pregnancy complicated by polyhydramnios, severe intractable epilepsy presenting in infancy, severe hypotonia, decreased muscle mass, global developmental delay, craniofacial dysmorphism (long face, large forehead, peaked eyebrows, broad nasal bridge, hypertelorism, large mouth with thick lips), and macrocephaly due to megalencephaly and hydrocephalus in most patients. Additional features that have been reported include cardiac anomalies like atrial septal defects, diabetes insipidus, and nephrocalcinosis, among others.

Dane

Klasyfikacja

Choroba

Synonimy

PMSE syndrome

PMSE syndrome

Kod ORPHA

500533

Kod OMIM

611087

Kod ICD10

G40.4

Kod ICD11

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*Źródło

orphanet