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