

## 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](#)

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