

## Opis choroby \*

### Definicja

A rare pervasive developmental disorder characterized by microcephaly, profound developmental delay, intellectual disability, bilateral cataracts, severe epilepsy including infantile spasms, hypotonia, irritability, feeding difficulties leading to failure to thrive, and stereotypic hand movements. The disease manifests in infancy. Brain imaging reveals delay in myelination and cerebral atrophy.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

500545

#### Kod OMIM

617393

#### Kod ICD10

F84.8

#### Kod ICD11

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### \*Źródło

orphanet