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