Opis choroby *

Definicja

A rare, severe early-onset neurodegenerative encephalopathy characterized mainly by developmental delay (DD) / developmental regression (DR), epilepsy, cortical atrophy, secondary hypomyelination and thin corpus callosum. Additional features include secondary microcephaly, hypotonia, spasticity, optic atrophy and skeletal anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
496641

Kod OMIM
617193

Kod ICD10
G93.4

Kod ICD11
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*Źródło

orphanet