Opis choroby *

Definicja

Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome is a rare, genetic, neurometabolic disorder characterized by severe, progressive microcephaly, severe to profound global development delay, intellectual disability, seizures (typically tonic and/or myoclonic and frequently intractable), hyperekplexia, and axial hypotonia with appendicular spasticity, as well as hyperreflexia, dyskinetic quadriplegia, and abnormal brain morphology (cerebral atrophy with variable additional features including ventriculomeglay, pons and/or cerebellar hypoplasia, simplified gyral pattern and delayed myelination). Cortical blindness, feeding difficulties and respiratory insufficiency may also be associated.

Dane

Klasyfikacja
Choroba

Synonimy
Asparagine synthetase deficiency
Niedobór syntetazy kwasu asparaginianowego

Kod ORPHA
391376

Kod OMIM
615574

Kod ICD10
E72.8

Kod ICD11
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*Źródło

orphanet