Opis choroby *

Definicja

Epileptic encephalopathy with global cerebral demyelination is a rare mitochondrial substrate carrier disorder characterized by severe muscular hypotonia, seizures (with or without episodic apnea) beginning in the first year of life, and arrested psychomotor development (affecting mainly motor skills). Severe spasticity with hyperreflexia has also been reported. Global cerebral hypomyelination is a characteristic imaging feature of this disease.

Dane

Klasyfikacja
Choroba

Synonimy
AGC1 deficiency
Niedobór mitochondialnego nośnika 1 asparaginianu-glutaminianu
Mitochondrial aspartate-glutamate carrier 1 deficiency

Kod ORPHA
353217

Kod OMIM
612949

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet