Opis choroby *

Definicja

A rare, genetic neurometabolic disease characterized by encephalomyopathy (including developmental delay, nystagmus, progressive ataxia, dystonia, amyotrophy, visual loss, sensorineural deafness, seizures) and bilateral, symmetrical lesions in the basal ganglia or brainstem on imaging, associated with nephrotic syndrome.

Dane

Klasyfikacja
Choroba

Synonimy
Infantile subacute necrotizing encephalopathy with nephrotic syndrome
Choroba Leigha z zespołem nerczycowym
Dziecięca podostra encefalopatia martwicza z zespołem nerczycowym
Leigh disease with nephrotic syndrome

Kod ORPHA
255249

Kod OMIM
614652

Kod ICD10
G31.8

Kod ICD11
5C53.24

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*Źródło

orphanet