Opis choroby *

Definicja

A rare genetic neurological disorder characterized by infantile or childhood onset of recurrent acute encephalopathic episodes with cerebellar and extrapyramidal involvement following febrile illnesses. During the episodes, patients typically show sudden onset of truncal ataxia, occasionally accompanied by lethargy and impairment of speech, as well as choreic and athetoid movements, seizures, loss of deep tendon reflexes, and presence of pathological reflexes. Episodes last from day to weeks and may leave residual symptoms such as speech impairment and poor coordination. There have been no further descriptions in the literature since 1983.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Recurrent encephalophathy of childhood
Nawracająca encefalopatia dziecięca

Kod ORPHA
2672

Kod OMIM
130950

Kod ICD10
G93.4

Kod ICD11
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*Źródło

orphanet