Niedobór epimerazy NAD(P)HX
Kod Orpha: 555407 Kod OMIM: 617186

Opis choroby *

Definicja
A rare neurometabolic disease characterized by infantile onset of rapidly progressive neurological deterioration, typically precipitated by a febrile illness. Patients present with hypotonia, loss of previously acquired motor milestones and cognitive skills, ataxia, nystagmus, tremor, seizures, tetraparesis, and respiratory failure, eventually resulting in a vegetative state. Imaging of the brain and spinal cord may show white matter abnormalities, cerebral atrophy, cerebellar edema, and spinal myelopathy. Subacute development of extensive bullous skin lesions within weeks of onset of neurological symptoms has also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Apolipoprotein A-I binding protein deficiency
Apolipoprotein A-I binding protein deficiency

Kod ORPHA
555407

Kod OMIM
617186

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.