Opis choroby *

Definicja

A rare neurometabolic disease characterized by infantile onset of repeated episodes of developmental regression and neurodegeneration, often triggered by febrile illnesses. Patients present with lethargy, hypotonia, irritability, gait ataxia, loss of speech, movement disorder, seizures, ophthalmoplegia, and hearing loss. Brain imaging shows generalized cerebral atrophy and bilateral basal ganglia abnormalities. Extensive skin lesions, cardiomyopathy, and pancytopenia have been reported in association. The condition is fatal in the first years of life.

Dane

Klasyfikacja
Choroba

Synonimy
CARKD deficiency
CARKD deficiency

Kod ORPHA
555402

Kod OMIM
618321

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet