Opis choroby *

Definicja

A rare lysosomal disease characterized by intermittent vomiting, hypotonia, lethargy, opisthotonos, and fatal outcome in early infancy, associated with deficient acid phosphatase in lysosomes. There have been no further descriptions in the literature since 1971.

Dane

Klasyfikacja
Choroba

Kod ORPHA
35121

Kod OMIM
200950

Kod ICD10
E83.3

Kod ICD11
5C64.3

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*Źródło

orphanet