Opis choroby *

Definicja

A rare genetic hematologic disease characterized by decreased or undetectable serum L-ferritin with otherwise normal laboratory parameters. Clinical signs and symptoms include generalized seizures, atypical restless leg syndrome, mild neuropsychologic impairment, and progressive hair loss. Asymptomatic cases have also been reported.

Dane

Klasyfikacja
Wada biologiczna

Kod ORPHA
440731

Kod OMIM
615604

Kod ICD10
E88.0

Kod ICD11
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*Źródło

orphanet