## 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