Opis choroby *

Definicja

A form of constitutional sideroblastic anemia characterized by severe microcytic anemia, B-cell lymphopenia , panhypogammaglobulinemia and variable neurodegeneration. The disease presents in infancy with recurrent febrile illnesses, gastrointestinal disturbances, developmental delay, seizures, ataxia and sensorineural deafness.

Dane

Klasyfikacja
Choroba

Synonimy
SIFD syndrome
Zespół SIFD

Kod ORPHA
369861

Kod OMIM
616084

Kod ICD10
D64.0

Kod ICD11
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*Źródło

orphanet