Rodzinna izolowana wrodzona asplenia
Kod Orpha: 101351 Kod OMIM: 271400

Opis choroby *

Definicja
Familial isolated congenital asplenia is a rare, non-syndromic, potentially life-threatening visceral malformation characterized by the absence of normal spleen function, resulting in a primary immunodeficiency. Typically, the condition manifests with severe, recurrent, overwhelming infections (especially pneumococcal sepsis) in otherwise apparently healthy infants. In adults with no history of severe sepsis in infancy, thrombocytosis may be the presenting sign. Howell-Jolly bodies on blood smears and an absent spleen on abdominal ultrasound examination are highly suggestive associated findings.
Dane

Klasyfikacja
Wada morfologiczna

Kod ORPHA
101351

Kod OMIM
271400

Kod ICD10
Q89.0

Kod ICD11
LB22.0

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.