Opis choroby *

Definicja

A rare inborn error of metabolism characterized by congenital asplenia and childhood or adolescent onset of generalized inflammation, persistent intravascular hemolysis and anemia, severe endothelial injury with abnormal coagulation, bleeding diathesis, and nephropathy. Additional reported manifestations include growth retardation, mild facial dysmorphism, and hepatomegaly.

Dane

Klasyfikacja Synonimy

Choroba HO-1 deficiency

HO-1 deficiency

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 562509
 614034
 E88.8

Kod ICD11

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*Źródło

orphanet