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
Choroba

Synonimy
HO-1 deficiency
HO-1 deficiency

Kod ORPHA
562509

Kod OMIM
614034

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet