Opis choroby *

Definicja

A rare, genetic, mixed autoinflammatory and autoimmune syndrome characterized by chronic systemic autoinflammation (presenting as recurrent fever in the neonatal or infantile period) and combined immunodeficiency (manifesting as recurrent viral and invasive bacterial infections). Muscular amylopectinosis may be subclinical or be complicated by myopathy/cardiomyopathy.

Dane

Klasyfikacja
Choroba

Kod ORPHA
329173

Kod OMIM
615895

Kod ICD10
D89.8

Kod ICD11
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*Źródło

orphanet