Opis choroby *

Definicja

A rare genetic systemic or rheumatologic disease characterized by neonatal or infantile onset of enterocolitis (which resolves with age), periodic fever, and episodes of severe systemic inflammation, which may be precipitated by infections, stress, or fatigue. Signs and symptoms include splenomegaly, urticaria-like rashes, arthralgia, and myalgia. Associated laboratory findings are elevated inflammatory markers (such as ferritin, C-reactive protein), pancytopenia, and elevated transaminases. If left untreated, flares can progress to coagulopathy, organ failure, and death.

Dane

Klasyfikacja
Choroba

Synonimy
NLRC4-related MAS
Zespół aktywacji makrofagów zależny od NLRC4
Zespół dziecięcego zapalenia jelit i autozapalenia zależny od NLRC4
Zespół autozapalenia z zespołem aktywacji makrofagów zależny od NLRC4
Zespół dziecięcego zapalenia jelit i choroby autoinflamatoryjnej zależny od NLRC4
NLRC4-related autoinflammatory syndrome with MAS
NLRC4-related autoinflammatory syndrome with macrophage activation syndrome
NLRC4-related infantile enterocolitis-autoinflammatory syndrome
NLRC4-related macrophage activation syndrome

Kod ORPHA
436166

Kod OMIM
616050

Kod ICD10
E85.0

Kod ICD11
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*Źródło

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