

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 autoinflammatoryjnej 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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