

## **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