Opis choroby *

Definicja

A rare genetic autoinflammatory syndrome with immune deficiency characterized by a combination of autoinflammation, immunodeficiency, and neutrophil dysfunction, as well as mild bleeding diathesis. Patients present recurrent attacks of abdominal pain, high fever, and systemic inflammation lasting four to five days and occurring every few weeks. Attacks may be accompanied by nailbed, tongue, submandibular, and gluteal abscesses, intra-abdominal granulomas, pyoderma gangrenosum, and buccal ulcerations. Frequent episodes of purulent paronychia, superficial skin and mucosal infections, and purulent upper respiratory tract infections have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
CAIN
CAIN

Kod ORPHA
566067

Kod OMIM
-

Kod ICD10
D89.8

Kod ICD11
-

---

*Źródło

orphanet