Opis choroby *

Definicja

A rare autosomal dominant autoinflammatory syndrome characterized by early onset systemic inflammation with autoimmune manifestations and more rarely, humoral immune deficiency and increased production of circulating proinflammatory cytokines, variably manifesting with recurrent oral aphthous ulcers, genital ulcers, arthralgia or arthritis, periodic fever, uveitis, and severe gastrointestinal involvement (pain, diarrhea, vomiting, rectal bleeding).

Dane

Klasyfikacja
Choroba

Synonimy
Behçet-like disease due to HA20
Choroba podobna do choroby Behçeta spowodowana HA20
Choroba podobna do choroby Behçeta spowodowana haploinsuficjencją A20
Behçet-like disease due to haploinsufficiency of A20

Kod ORPHA
476102

Kod OMIM
301074

Kod ICD10
D89.8

Kod ICD11
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*Źródło

orphanet