Opis choroby *

Definicja

A rare genetic immune disease characterized by recurrent sinopulmonary infections and autoimmune enterocolopathy, manifesting as frequent episodes of intractable diarrhea with abdominal pain and fever, accompanied by eczematous rashes, due to deficits in components of innate and adaptive immunity. Immunologic abnormalities include IgG subclass deficiency, impaired antigen-induced lymphocyte proliferation, reduced cytokine production by CD8+ T lymphocytes, and decreased numbers of natural killer cells.

Dane

Klasyfikacja
Choroba

Synonimy
NFAT5 haploinsufficiency
NFAT5 haploinsufficiency

Kod ORPHA
529980

Kod OMIM
-

Kod ICD10
D89.8

Kod ICD11
-

---

*Źródło

orphanet