

Opis choroby *

Definicja

Immunodeficiency due to MASP-2 deficiency is a rare, genetic immunodeficiency due to a complement cascade protein anomaly characterized by low serum levels of MASP-2 and a variable susceptibility to bacterial infections (e.g. pulmonary tuberculosis, pneumococcal pneumonia, skin abscesses and sepsis), and autoimmune diseases (e.g. inflammatory lung disease, cystic fibrosis, systemic lupus erythematosus). In many cases it remains asymptomatic.

Dane

Klasyfikacja

Choroba

Kod ORPHA

331187

Kod OMIM

613791

Kod ICD10

D84.1

Kod ICD11

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*Źródło

orphanet