Opis choroby *

Definicja

Immunodeficiency with factor I anomaly is a rare, genetic, primary immunodeficiency disease characterized by increased susceptibility to recurrent, usually severe, infections (particularly by Neisseria meningitidis, Haemophilus influenzae and Streptococcus pneumoniae), typically manifesting as otitis, sinusitis, bronchitis, pneumonia, and/or meningitis. Autoimmune disease (e.g. systemic lupus erythematosus, glomerulonephritis) and atypical hemolytic uremic syndrome may be associated. Laboratory serum analysis reveals, in addition to diminished or undetectable complement factor I, variably decreased complement C3, complement factor B and complement factor H.

Dane

Klasyfikacja
Choroba

Synonimy
Complete factor I deficiency

Kod ORPHA
200418

Kod OMIM
610984

Kod ICD10
D84.1

Kod ICD11
4A00.1Y

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*Źródło

orphanet