Niedobór odporności zwiazany z FADD
Kod Orpha: 306550 Kod OMIM: 613759

Opis choroby *

Definicja
FADD-related immunodeficiency is a rare genetic immunological disease reported in a single consanguineous Pakistani family with several affected members presenting with severe bacterial and viral infections, recurrent hepatopathy (portal inflammation, fibrosis), and recurrent, stereotypical febrile episodes, sometimes lasting several days, with encephalopathy and difficult-to-control seizures. Variable cardiac malformations were also reported. Although there were autoimmune lymphoproliferative syndrome (ALPS)-like biological features, clinical ALPS was not present. A homozygous missense mutation in the FADD gene (11q13.3) was found in the family and the disease is thought to follow an autosomal recessive pattern of inheritance.
Dane

Klasyfikacja
Choroba

Kod ORPHA
306550

Kod OMIM
613759

Kod ICD10
D89.8

Kod ICD11
4A01.21

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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