

Opis choroby *

Definicja

A rare syndromic agammaglobulinemia characterized by profound B-cell depletion (with normal T-cell numbers) resulting in agammaglobulinemia, associated with severe developmental delay, microcephaly, craniosynostosis, cleft palate, narrowing of the choanae, blepharophimosis, and severe dermatitis. Additional reported features include distal joint contractures, renal/genitourinary anomalies, and mild cerebral atrophy, among others.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

83617

Kod OMIM

610483

Kod ICD10

Q87.0

Kod ICD11

4A01.00

*Źródło

orphanet