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

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*Źródło

orphanet