Opis choroby *

Definicja

A rare functional neutrophil defect characterized by infantile onset of increased susceptibility to pyogenic infections, especially of the skin, ears, lung, and lymph nodes, with neutrophils lacking specific granules and exhibiting bilobed nuclei on peripheral blood smear. Bone marrow biopsy shows hypercellularity, paucity of neutrophil granulocytes, and progressive myelodysplasia. Additional manifestations may include mild to moderate developmental delay, mild facial dysmorphic features (such as dysplastic ears), and anomalies of bones, teeth, and nails.

Dane

Klasyfikacja
Choroba

Synonimy
Neutrophil-specific granule deficiency
Niedobór specyficznych ziarnistości neutrofilowych

Kod ORPHA
169142

Kod OMIM
245480

Kod ICD10
D71

Kod ICD11
4A00.0Y

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*Źródło

orphanet