

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

*Źródło

orphanet