Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by early-onset progressive bone marrow failure with anemia, leukopenia, mild thrombopenia, and myelodysplastic features, as well as non-hematologic manifestations, such as developmental delay, cataracts, facial dysmorphism, short stature, and skeletal anomalies. Immunodeficiency primarily affects B-cells and may lead to increased susceptibility to infections. Additional reported features include dry skin and eczema, cardiac anomalies, hearing loss, and reduction of cerebral volume on brain imaging.

Dane

Klasyfikacja
Choroba

Synonimy
MYSM1 deficiency
Niedobór MYSM1

Kod ORPHA
508542

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet