Opis choroby *

Definicja

A rare hematologic disease characterized by clinical and morphological findings indistinguishable from those of acute myeloid leukemia, typically occurring in newborns with Down syndrome. Peripheral blood and bone marrow blasts display features suggestive of megakaryoblasts. In addition to trisomy 21, acquired GATA1 mutations are present in blast cells. Patients may be asymptomatic or present with thrombocytopenia, less commonly other cytopenias, leukocytosis, hepatosplenomegaly, jaundice, ascites, respiratory distress, bleeding, and pericardial or pleural effusions. Most patients undergo spontaneous remission within the first three months of life, although some may develop life-threatening hepatic, renal, or cardiac complications.

Dane

Klasyfikacja
Choroba

Synonimy
TMD
Przejściowa choroba mieloproliferacyjna
Przejściowa nieprawidłowa mielopoeza
TMD
Transient abnormal myelopoiesis
Transient myeloproliferative disease

Kod ORPHA
420611

Kod OMIM
159595

Kod ICD10
D47.7

Kod ICD11
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*Źródło

orphanet