Opis choroby *

Definicja

A rare myelodysplastic/myeloproliferative neoplasm characterized by a proliferation primarily of granulocytic and monocytic lineages with infiltration of the liver and spleen, among other organs. Blasts and promonocytes account for less than 20% of white blood cells in peripheral blood and bone marrow. Erythroid and megakaryocytic abnormalities are often present. BCR-ABL1 fusion is absent, while somatic mutations in genes of the RAS pathway or monosomy 7 may be found. The condition may also occur in the context of neurofibromatosis type 1 or Noonan syndrome-like disorder. Children of less than three years are predominantly affected, with a clear male preponderance. Most patients present with constitutional symptoms, signs of infection, and hepatosplenomegaly.

Dane

Klasyfikacja
Choroba

Synonimy
JMML
Młodzieńcza przewlekła białaczka mielomonocytowa
Juvenile chronic myelomonocytic leukemia

Kod ORPHA
86834

Kod OMIM
607785

Kod ICD10
C93.3

Kod ICD11
2A42

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*Źródło

orphanet