

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

[*Źródło](#)

orphanet