Acute myeloid leukemia with NPM1 somatic mutations

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Acute myeloid leukemia with NPM1 somatic mutations. ORPHA:402026

Acute myeloid leukemia with NPM1 somatic mutations is a subtype of acute myeloid leukemia with recurrent genetic abnormalities characterized by clonal proliferation of myeloid blasts harboring mutations of the NPM1 gene in the bone marrow, blood and other tissues. It is associated with multilineage dysplasia, involving the myeloid, monocytic, erythroid, and megakaryocytic cell lineages. Patients usually present with leukocytosis, thrombocytosis and nonspecific symptoms related to ineffective hematopoiesis (fatigue, bleeding and bruising, recurrent infections, bone pain), with frequent extramedullary involvement typically presenting as gingival hyperplasia and lymphadenopathy.