

## Editorial

# Acute Basophilic Leukemia, a Rare Subset of De Novo AML with an Abnormal Tetraploid Karyotype

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Rare de novo AML and MDS in male patients has been reported in a few cases with massive hyperdiploid and tetraploid karyotype [1-3]. A low remission rate and short survival are characteristic of this prognostically unfavorable cytogenetic group in de novo AML.

We report a 50-year-old male with a history of diabetes who presented with pancytopenia, neutropenia, and worsening hypoxemia, ultimately requiring intubation and transfer to the MICU in March 2013. He was diagnosed with AML on the basis of increased blasts in his peripheral blood smear (sample collected 03/09/13). His bone marrow biopsy (sample collected 03/11/13) was markedly hypercellular with 80-90% cellularity and was involved by AML by morphology (48.5% blasts) and immunophenotype. The blasts were remarkable for large, basophilic granules, and a range of immature basophils were seen in the background. Blasts had an immunophenotype suggestive of basophilic differentiation (CD13+, CD33 moderate, CD34+, CD117-, and CD123+).

Molecular genetic studies were negative for KIT Asp816Val mutation, FLT3 ITD, or mutations in CEBPA or NPM1. Both peripheral blood and bone marrow samples revealed an abnormal karyotype with tetraploid chromosomal complement and structural anomalies of chromosome 1 and 12. His karyotype was 92,XXYY,dup(1)(q21q32)x2,del(12)(q24.1)[10]/46,XY[10]. Fluorescence in situ hybridization (FISH) for AML and MDS (5,7,8,20,t(8;21), t(15;17), inv(16) and MLL) probes on the bone

marrow sample (03/11/13) revealed 4 copies for all the target regions tested.

His bone marrow on 03/27/13 on day 14 of induction with HiDAC and etoposide revealed residual acute myeloid leukemia, with persistent blasts that demonstrated more prominent basophilic differentiation and increased background basophilia, as well as persistence of the abnormal tetraploid clone. There was a decrease in the percentage of cells with the abnormal karyotype, as well as a decreased percentage of blasts, compared to the previous bone marrow biopsy. Reinduction chemotherapy was administered and most recent follow up bone marrows were negative for leukemia by morphology and cytogenetics.

Acute basophilic leukemia is an extremely rare subset of AML, and tetraploid karyotype is a rare cytogenetic finding; the combination of these two findings is unique in the literature.

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