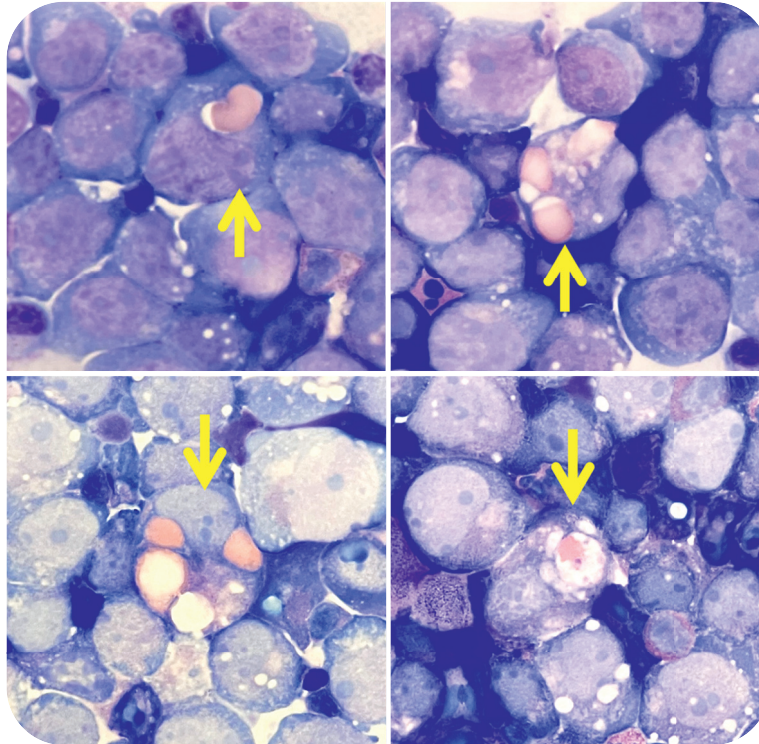


Leukemic myeloblasts with erythrophagocytosis

Huifei Liu, Esoterix Pathology Practice Group



An 80-year-old woman was diagnosed with acute myeloid leukemia (AML). Bone marrow aspirate revealed increased large-sized myeloblasts with scant cytoplasm, irregular nuclei, and fine chromatin. Some blasts contained cytoplasmic vacuoles with engulfed red blood cells, consistent with erythrophagocytosis (see yellow arrows; Wright-Giemsa stain, original magnification $\times 100$ for all panels). The myeloblasts had high side scatter, and were positive for CD4, CD11b(dim), CD11c, CD13 (negative to dim), CD14 (minor subset), CD15, CD33, CD56(partial), CD64, HLA-DR, and myeloperoxidase and negative for CD34 and CD117. Next-generation sequencing identified 3 different tier 1 mutations of TET2 with variant frequencies of 37%, 22%, and 6%. Chromosome analysis showed a complex karyotype: 45~47,XX,add(3)(q29),+5,i(5)(p10),t(8;22)(p11.2;q13),der(10;15)(q10;q10),der(10;18)(q10;q10),der(10;21)

(q10;q10),der(10;22)(q10;q10),+13,der(19)t(11;19)(q13;p13.3)del(11)(q23q23),+mar[cp19]/46,XX[1]. Translocations t(8;22)(p11;q13) involving *KAT6A* (lysine histone acetyltransferase 6A) and *EP300* have been reported in rare cases of acute monocytic leukemia or acute myelomonocytic leukemia.

KAT6A is a member of the MYST family of histone acetyltransferases. AML with *KAT6A* translocation is very rare (<0.5% of AML), with *CREBBP* the most common partner, followed by *TIF2* and *EP300*. The majority of AML cases with *KAT6A* translocations are therapy-related, with poor prognosis. Our case demonstrates the unique features of myeloblasts with *KAT6A* translocation, including monocytic differentiation without immature marker expression (CD34 and CD117) and erythrophagocytosis.