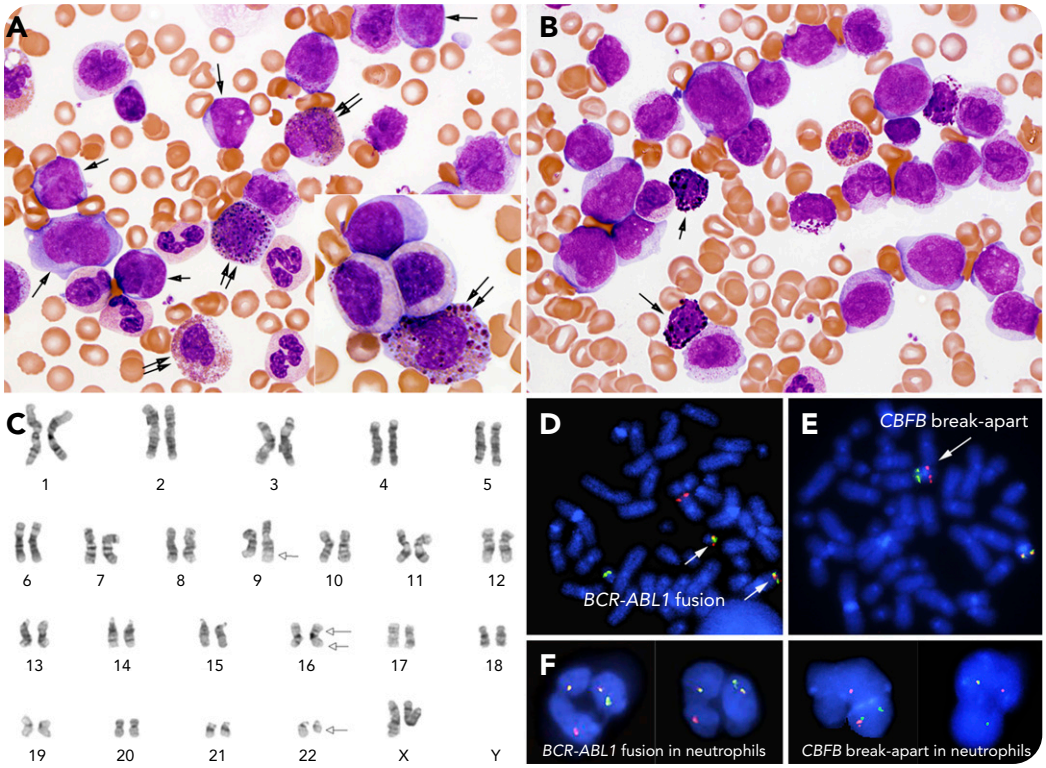
blood work
images in hematology

Acute myeloid leukemia with coexistence of t(9;22) and inv(16)

Sory Ruiz and Yi-Hua Chen, Northwestern Memorial Hospital



A young adult woman with no significant past medical history presented with marked leukocytosis. The blood smear showed 50% blasts (panel A, single arrow [original magnification ×1000; Wright-Giemsa stain]) in a background of neutrophilia with myelocytes and promyelocytes, eosinophilia (panel A, double arrows), and basophilia (panel B, arrow [original magnification ×1000; Wright-Giemsa stain]). The morphologic findings raised the possibility of a blast phase of chronic myelogenous leukemia (CML). However, atypical large purple granules were noted in the eosinophils (panel A, inset [original magnification ×1000]), suggesting an acute myeloid leukemia (AML) with inv(16). Cytogenetic analysis (panel C) did reveal coexistence of t(9;22)(q24;q11.2) and inv(16)(p13.1q22) in all 20 cells analyzed, and fluorescence in situ hybridization was positive for *BCR-ABL1* fusion in 44% of the cells (panel D [original magnification ×1000; DAPI [4′,6-diamidino-2-phenylindole] counterstain]) and *CBFB* rearrangements in 19.5% of the cells (panel E [original magnification ×1000; DAPI counterstain]). These 2 genetic abnormalities were also identified in the neutrophils (panel F [original magnification ×1000; DAPI counterstain]).

De novo AML with coexistence of t(9;22) and inv(16) is rare, and limited data suggest that these cases have as favorable a prognosis as AML with inv(16) alone. However, coexistence of inv(16) in CML seems to have an unfavorable prognosis with rapid disease progression to accelerated or blast phase. The presence of *BCR-ABL1* and *CBFB* rearrangements in the neutrophils in this case indicates that these abnormalities likely precede AML, most suggesting a blast phase of CML. The patient received chemotherapy and dasatinib, underwent a bone marrow transplant, and has been in molecular remission.

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