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Relapsed *NPM1* mutated acute myeloid leukemia with *PDGFRA* rearrangement

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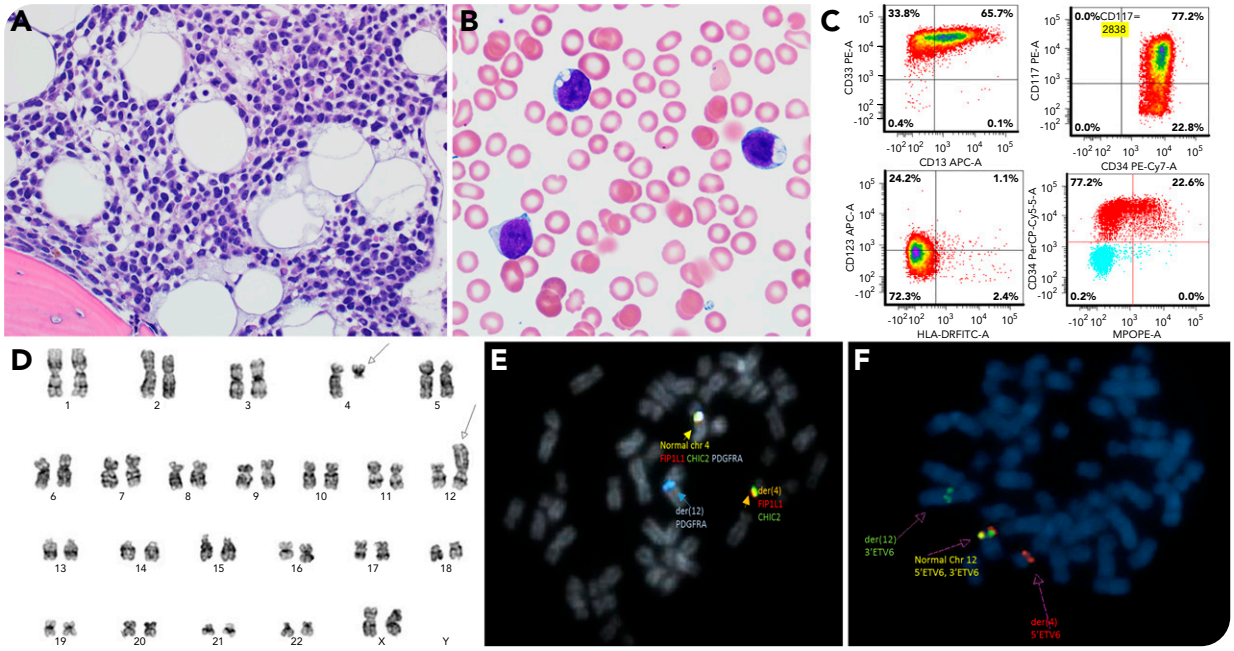

A 40-year-old woman was diagnosed with acute myeloid leukemia (AML) with diploid cytogenetics. Molecular studies were positive for *NPM1*p.W288fs, *FLT-3*p.N676K, and *FLT-3*p.D835V mutations (variant allele frequency [VAF], 37%, 5%, and 17%, respectively). Postchemotherapy, she achieved complete remission and underwent allogeneic stem cell transplant. Unfortunately, her disease relapsed 6 months posttransplant with 45% aberrant myeloblasts (panel A, hematoxylin and eosin stain, original magnification ×20; and panel B, Wright-Giemsa stain, original magnification ×100), with immunophenotype similar to blasts at the time of diagnosis: $CD34^+/CD117^+/CD13^+/CD33^+$/CD123dec/$HLA^-DR^-$/$MPO^+$ (panel C). Molecular studies were positive for *NPM1*p.W288fs (VAF, 10%), *FLT-3*p.N676K (VAF, 4%), and additional mutations in *TET2*p.L1231P, *TET2*p.K1299, and *WT1*p.R458* genes (VAF, 30%, 4%, and 5%, respectively). Chimerism study showed 61% XX and 39% XY. Cytogenetic study showed an abnormal karyotype: 46,XX,t(4;12)(q12;p13[19]/46,XY[1]) (panel D). Fluorescence in situ hybridization with FIP1L1/CHIC2/PDGFRA (panel E) and ETV6 break-apart (panel F) probes on metaphases revealed *PDGFRA* and *ETV6* gene rearrangements, respectively, confirming karyotype and indicating PDGFRA-ETV6 fusion. No eosinophilia was present. FISH was negative for *PDGFRA* rearrangement at initial diagnosis of AML.

NPM1 mutated AML usually has a diploid karyotype. To our knowledge, this is the first case of *NPM1* mutated AML that relapsed after allogeneic stem cell transplant with *PDGFRA* rearrangement, resulting in PDGFRA-ETV6 fusion, likely a result of a $NPM1^+$ subclone that persisted and showed posttransplant clonal evolution.

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