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Myelodysplastic/myeloproliferative neoplasm with ring sideroblasts, thrombocytosis, and mutated *JAK2/SF3B1* without anemia

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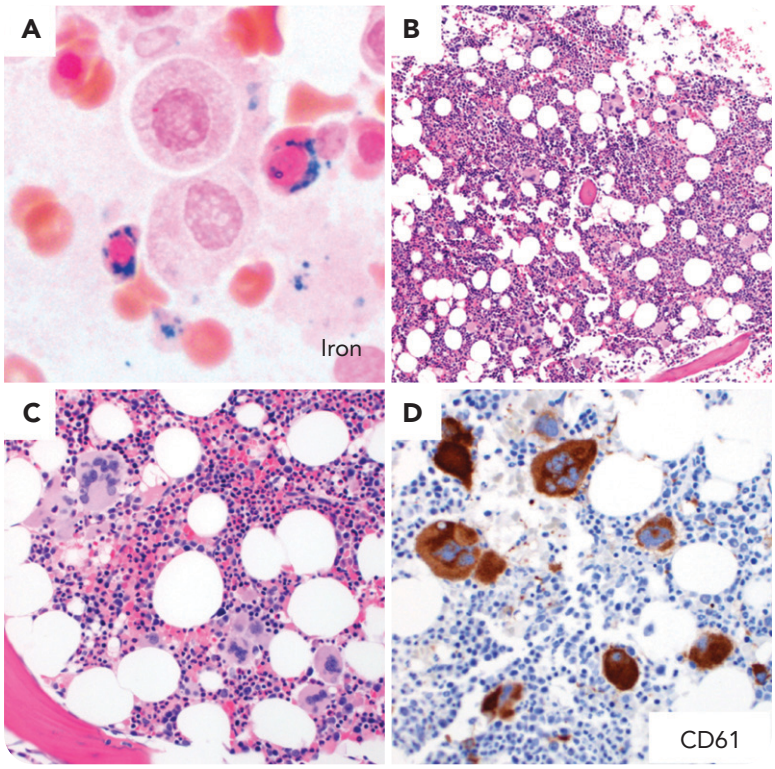

A 76-year-old woman with metastatic lung squamous cell carcinoma after chemoradiation presented for follow-up and was found to have leukocytosis, thrombocytosis, and normal hemoglobin with macrocytosis (white blood cell count, 21.2 × 10^9/L; platelets, 825 × 10^9/L; hemoglobin, 12.1 g/dL [normal reference, 11.7-15.5 g/dL]; mean corpuscular volume, 108 fL). Prior blood counts showed similar hemoglobin levels 14 months before (13.0 g/dL) and 2 months after marrow biopsy (12.6 g/dL). Bone marrow evaluation revealed 36% ring sideroblasts (prussian blue iron stain in panel A; 100× objective, original magnification ×1000), hypercellular marrow for age (40% to 50%; hematoxylin and eosin stain in panel B; 4× objective, original magnification ×40) with clustering of atypical, hyperlobated megakaryocytes (hematoxylin and eosin stain in panel C and CD61 immunohistochemical stain in panel D; 20× objective, original magnification ×200), and <2% blasts. Cytogenetics showed 46,XX[20]. Next-generation sequencing revealed *SF3B1* K700E (21%) and *JAK2* V617F (14%) mutations.

This case is a rare example of myelodysplastic syndrome/myeloproliferative neoplasm with ring sideroblasts and thrombocytosis (MDS/MPN-RS-T) in which no anemia is observed. The 2016 World Health Organization defines the entity as anemia associated with erythroid or multilineage dysplasia, ≥15% ring sideroblasts, <5% marrow blasts, persistent thrombocytosis (≥450 × 10^9/L), *SF3B1* mutation, and exclusion of other MPN, MDS, or certain gene mutations/rearrangements. These patients may have additional alterations, including *JAK2* (≤70%), *CALR*, *MPL*, *ASXL1*, *DNMT3A*, *SETBP1*, and *TET2*. The co-occurrence of mutations likely contributes to its complex presentation. The absence of anemia, in the presence of disease-specific mutations along with other major criteria, should not exclude the diagnosis of MDS/MPN-RS-T.

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