blood work
images in hematology

Concurrent *JAK2* mutation and isolated del(5q) associated with marrow fibrosis and small hypo/monolobated megakaryocytes

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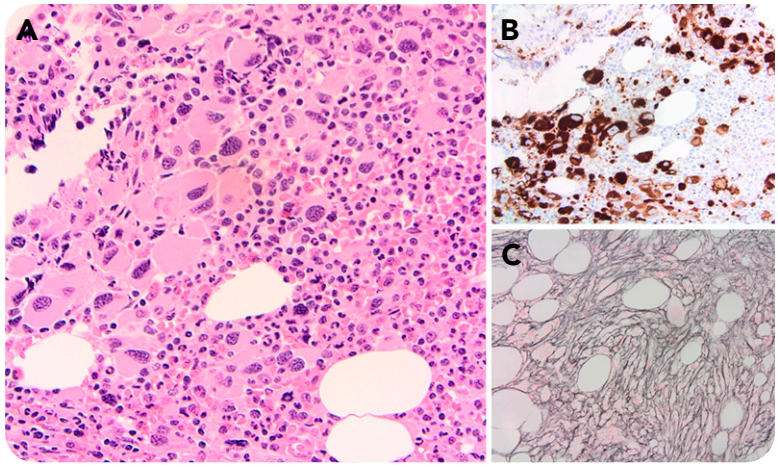

A 68-year-old woman of Asian descent presented with fatigue and anemia requiring blood transfusion. There was no splenomegaly. The blood work revealed hemoglobin, 59 g/L; mean corpuscular volume, 103 fL; absolute neutrophil count, 2.3 × 10^9/L; platelets, 238 × 10^9/L; and lactate dehydrogenase, 246 U/L. Occasional teardrop cells and rare pseudo-Pelger neutrophils were found in the blood smear. The bone marrow aspirate was a dry tap. The biopsy showed a hypercellular marrow with proliferation of small hypolobated/monolobated megakaryocytes forming clusters (panel A; original magnification ×40, hematoxylin and eosin stain) that stained positive for CD61 (panel B; original magnification ×40, CD61 stain). $CD34^+$ blasts were <5%. There was increased reticulin fibrosis (marrow fibrosis [MF] 2/3) (panel C; original magnification ×20, reticulin stain). Molecular studies detected *JAK2* V617F mutation with 29% allele burden; conventional cytogenetics demonstrated an isolated 5q deletion in 4 of 20 metaphases [46,XX, del(5)(q22q35)], confirmed by interphase fluorescence in situ hybridization in 25% of nuclei.

Myelodysplastic syndrome with isolated del(5q) is characterized by distinctive hypo/monolobated megakaryocytes. Concurrent *JAK2* mutation has been described in 5% of cases, but significant MF (≥2) was not reported. Isolated del(5q) in primary myelofibrosis (PMF) is extremely rare. The megakaryocyte morphology typical of PMF was absent in this case. Whether there are 2 independent clones or 1 clone expressing both abnormalities remains to be determined. Nevertheless, this patient could potentially benefit from lenalidomide.

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