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Del(5q) myelodysplastic syndrome combined with pure red cell aplasia

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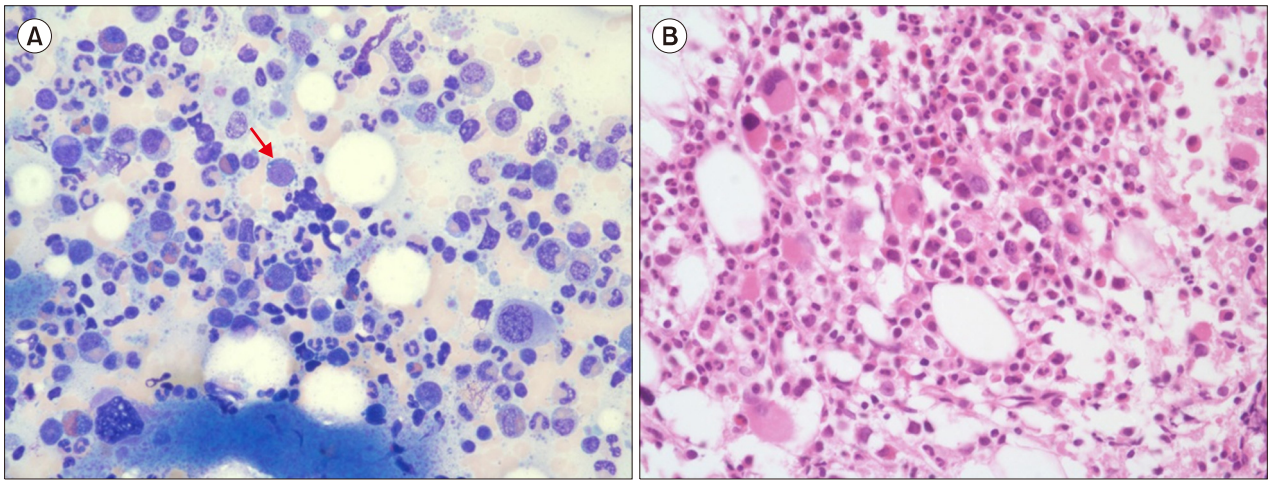

A 76-year-old woman was admitted with dizziness for 2 weeks. No organomegaly or lymphadenopathy was revealed on physical examination. Initial blood cell values were as follows: hemoglobin, 5.4 g/dL; mean corpuscular volume, 105 fL; white blood cell count, 8.1×10^9/L (differential count: neutrophils, 54%; lymphocytes, 37%; monocytes, 6%; eosinophils, 3%); platelet count, 402×10^9/L; and absolute reticulocyte count, 3.9×10^9/L. Biochemical and serologic test results were unremarkable. Direct Coombs test result was negative. Bone marrow (BM) examination showed hypercellular marrow with marked erythroid hypoplasia (1.4% erythroblasts). The estimated myeloid-to-erythroid ratio was 72:1. Most erythroblasts found were early erythroid precursors, and late erythroblasts were rarely observed (**A**, BM aspiration, Wright-Giemsa stain, ×400, arrow). Granulopoiesis was normal; however, dysplastic megakaryocytes with non-lobulated and hypolobulated nuclei were observed in several fields (**B**, BM biopsy, hematoxylin and eosin stain, ×400). No evidence of thymoma was detected on computed tomography scans. Subsequently, chromosome study demonstrated 46,XX,del(5)(q15q33)[18]/46,XX[2]; therefore, the patient was diagnosed with myelodysplastic syndrome (MDS) with isolated del(5q). Erythroid hypoplasia is frequently observed in 5q- syndrome; however, a marked decrease with maturation arrest of erythropoiesis mimicking pure red cell aplasia is very unusual. TP53 activation has been suggested to cause erythroid suppression in MDS with del(5q).