bloodwork
images in hematology

NUT midline carcinoma with leukemic presentation mimicking CD34-positive acute leukemia

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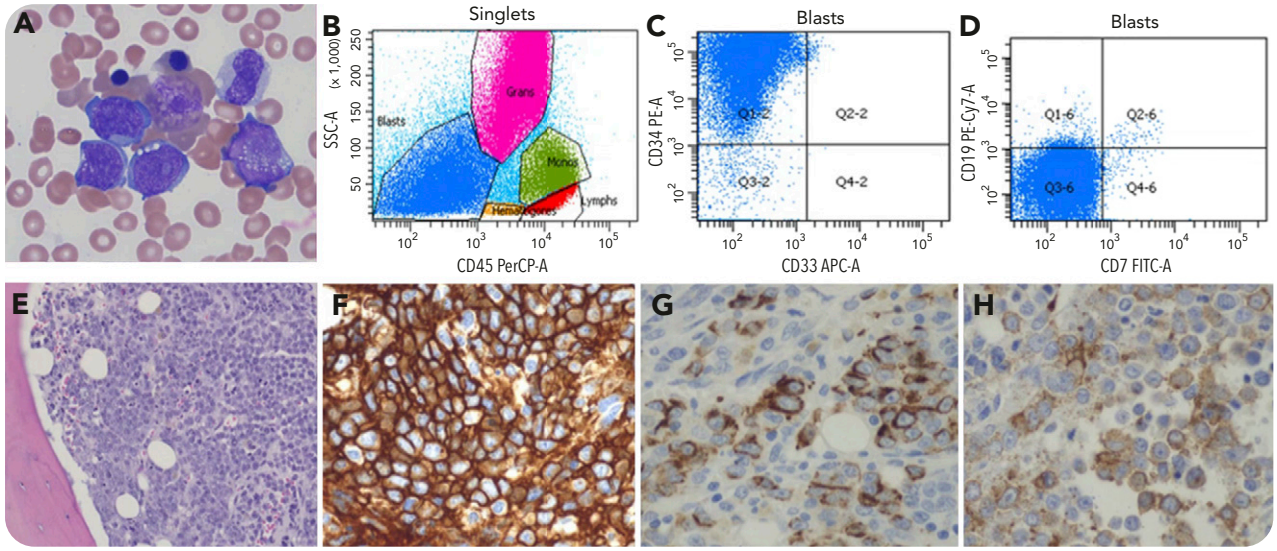

A 17-year-old boy presented with cough, chest pain for 3 weeks, and weight loss for several months. He was treated with multiple rounds of antibiotics with no improvement. A right lower lobe lung lesion with extension into the mediastinum and innumerable bone metastases were found by computed tomography and positron emission tomography scan. Bone marrow (BM) aspirate smear showed frequent medium- to large-sized mononuclear cells with blast morphology (panel A; Wright-Giemsa stain, original magnification ×1000). Flow cytometry of BM aspirate showed a CD45-negative abnormal cell population (in blue) strongly positive for CD34 and negative for myeloid or lymphoid markers (panels B-D). BM biopsy sections showed diffuse infiltrate by abnormal mononuclear cells (panel E; hematoxylin and eosin stain, original magnification ×200). The abnormal cells were strongly positive for CD34 (panel F; original magnification ×400) and focally positive for cytokeratin CAM5.2 and epithelial membrane antigen (panels G and H; original magnification ×400). Fluorescence in situ hybridization analysis showed *NUTM1* (15q14) rearrangement. NUT midline carcinoma (NMC) was diagnosed. The patient had a brief response to the Scandinavian Sarcoma Group (SSG IX) protocol and died approximately 5 months after initial presentation.

NMC is a rare, very aggressive, poorly differentiated tumor that commonly affects mediastinum, head/neck, or other organs of the midline region. It can express CD34. Without cytogenetic study or immunostaining for NUT protein, the diagnosis is often difficult. When it diffusely infiltrates BM, it may mimic acute undifferentiated leukemia.

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