WASHINGTON POSTER FINALIST - CLINICAL VIGNETTE Sarah Anne Buckley, MD

Pernicious Anemia Mimicking Acute Erythroblastic Leukemia

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INTRODUCTION: Vitamin B12 deficiency causes characteristic hematologic alterations such as increased MCV, hypersegmented neutrophils, hemolysis, and cytopenias. These resolve with replacement of vitamin B12. The following case describes a patient with pernicious anemia whose vitamin B12 deficiency was so severe that he developed hematologic abnormalities initially concerning for acute erythroblastic leukemia or myelodysplastic syndrome. These abnormalities, including the presence of peripheral blasts and severe marrow dyserethropoeisis, corrected with vitamin B12 administration.

CASE PRESENTATION: A 26-year-old previously healthy African American man presented to the hospital with a one-month history of fatigue and a witnessed syncopal episode. On presentation, the patient was alert with normal vital signs. A non-contrast head CT ruled out any acute intracranial process. Laboratory studies revealed a white blood cell count of 2400 cells/mm2 with a lymphocytic predominance, a hemoglobin of 5.6 g/dl with an MCV of 107 fl and a reticulocyte index of 0.2%, a platelet count of 125,000 cells/ul, a haptoglobin <10 mg/dl, an LDH of >4000 U/L, an undetectable level of vitamin B12 (<50 pg/dL), and normal folate (10.5 ng/mL) and ferritin (364 ng/mL). The peripheral blood smear showed rare nucleated red blood cells with polychromatophils, dacrocytes, and schistocytes. There were no hypersegmented neutrophils. Peripheral blood flow cytometry revealed 1.8% myeloid blasts. A bone marrow biopsy was hypercellular with maturing trilineage hematopoiesis, erythroid hyperplasia, and diserythropoiesis concerning for erythroblastic leukemia. Subsequent flow cytometry demonstrated 6.7% myeloid blasts with no definite abnormal blast population. Cytogenetics were normal 46XY and iFISH was negative for abnormalities common in myelodysplastic syndrome. The patient was later found to have serum antibodies against intrinsic factor and parietal cells; a diagnosis of pernicious anemia was made, and the patient's hematologic abnormalities resolved with replacement of vitamin B12.

DISCUSSION: Though the correlation between vitamin B12 deficiency and myelodysplasia is well described, the nature of this relationship remains uncertain. Population-based studies reveal that a diagnosis of pernicious anemia confers an elevated risk of developing AML and MDS, though it is not clear whether there is a causative relationship. There are multiple case reports of the co-occurrence of vitamin B12 deficiency and AML/MDS; in each case, vitamin correction was attempted but was unable to prevent dysplastic or malignant progression. In the above case, however, vitamin B12 supplementation was able to reverse all evidence of evidence of leukemia and dysplasia.