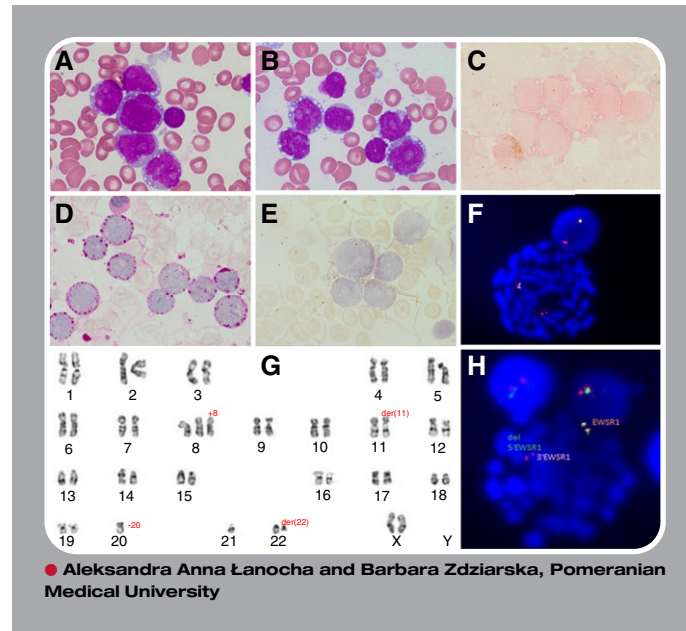


T-cell lymphoblastic leukemia with t(11;22)(q24;q12) and EWSR1 rearrangement



A 35-year-old woman with a medical history of hypothyroidism after stabilization of the thoracic spine because of scoliosis and after Lyme disease was referred for possible leukemia. Laboratory tests showed leukocytosis (white blood cells, $65 \times 10^9/L$), anemia (hemoglobin, 9.2 g/dL), and thrombocytopenia (platelets, $9 \times 10^9/L$), and blasts were noted on a peripheral blood smear. Whole-body computed tomography showed lymphadenopathy (~ 15 mm), splenomegaly (137 mm), and enlarged left ovary ($25 \times 27 \times 30$ mm). Bone marrow aspirate (May-Grünwald-Giemsa stain) revealed hypercellularity with 99.2% blasts (panels A and B; original magnification $\times 1000$ for panels A-E). Cytochemical staining was negative with myeloperoxidase (panel C), positive with periodic acid Schiff (panel D), and negative with alkaline phosphatase and specific esterase (panel E). Flow cytometric analysis demonstrated blasts (89%) with antigens MPO⁻, CD45RA⁺, CD7⁺, cytCD3⁺, TdT⁺, CD99⁺, and sCD3⁻. A molecular study was negative for *BCR-ABL* rearrangement. A cytogenetics study (panel G) revealed 46,XX,+8,der(11)t(11;22)(q24;q12),-20,der(22)t(11;22)(q24;q12) (International System for Human Cytogenetic Nomenclature [ISCN] 2013). Interphase fluorescence in situ hybridization studies demonstrated nuclear in situ hybridization (5'EWSR1x2,3'EWSR1x1)(5'EWSR1con3'EWSR1x1) [196/200]; ish 22q12 (EWSR1+), 22q12(5'EWSR1con3'EWSR1+)(5'EWSR1+,3'EWSR1-) (ISCN 2013) (panels F and H). Clinical staging confirmed a diagnosis of acute T-cell lymphoblastic leukemia. The patient was treated with chemotherapy according to the Polish Adult Leukemia Group ALL6 Ph(-) <55 years protocol.

This is a rare case of coexistence of acute lymphoblastic leukemia and Ewing sarcoma breakpoint region 1 (EWSR1) rearrangement. The EWSR1 rearrangement is present in more than 90% of Ewing sarcomas and in the related group of peripheral primitive neuroectodermal tumor, rhabdomyosarcoma, neuroblastoma, and giant cell tumor of the bone.



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