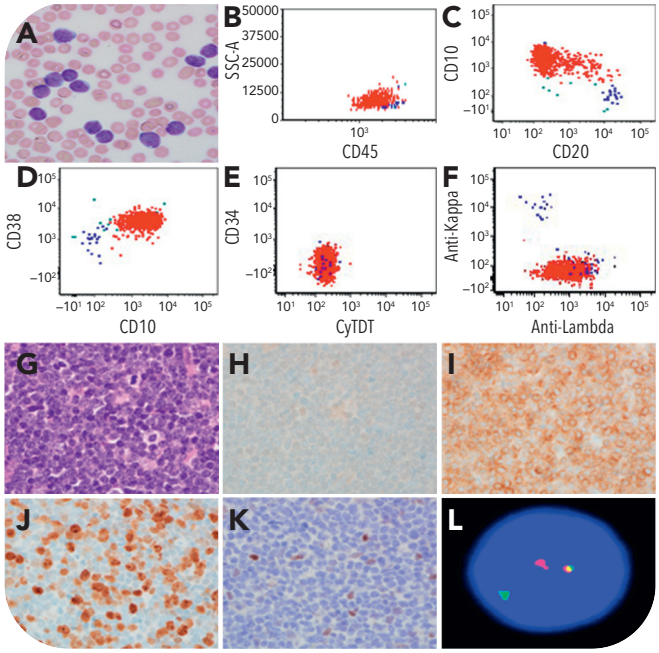
blood work
images in hematology

KMT2A-rearranged B-lymphoblastic leukemia/lymphoma with surface light chain restriction and lacking immature markers

Lucy Fu and Shunyou Gong, Ann & Robert H. Lurie Children's Hospital of Chicago



An 8-month-old boy presented with scalp nodules and diffuse lymphadenopathy. Complete blood cell count showed leukocytosis (white blood cell count, 282 000/μL), anemia, and thrombocytopenia. Peripheral blood smear (panel A; Wright-Giemsa stain, 100× objective) showed numerous medium-sized lymphoid cells with blastic morphology. Flow cytometry (panels B-F) detected surface λ light chain restricted B cells expressing CD19, CD20 (partial), CD10, and CD38; the cells were negative for CD34 and terminal deoxynucleotidyl-transferase (TdT). Biopsy of the left inguinal lymph node (panel G; hematoxylin and eosin stain, 100× objective) showed sheets of monotonous medium-sized cells with multiple small nucleoli and frequent mitoses. The cells were B-cell lymphoma (BCL)6$^-$ (panel H; 100× objective), BCL2$^+$ (panel I; 100× objective), Ki-67 60% (panel J; 100× objective), and c-MYC$^-$ (panel K; 100× objective). Fluorescence in situ hybridization was positive for *KMT2A* rearrangement (panel L) and negative for *MYC* and *BCL2* gene rearrangements. Molecular studies detected the *KMT2A*::*MLLT3* fusion and 8-bp deletion of *KMT2D* gene, leading to a truncated loss-of-function protein. A diagnosis of B-lymphoblastic leukemia/lymphoma (B-ALL) was made.

B-ALLs with light-chain restriction or lacking both CD34 and TdT were each sporadically reported. However, cases with a combination of these 2 "mature" features are extremely rare and difficult to be distinguished from mature B-cell neoplasms. Recognition of these unusual characteristics in B-ALL should facilitate prompt confirmatory/genetic testing and avoid misdiagnosis as mature B-cell lymphoma.

https://doi.org/10.1182/blood.2023021556