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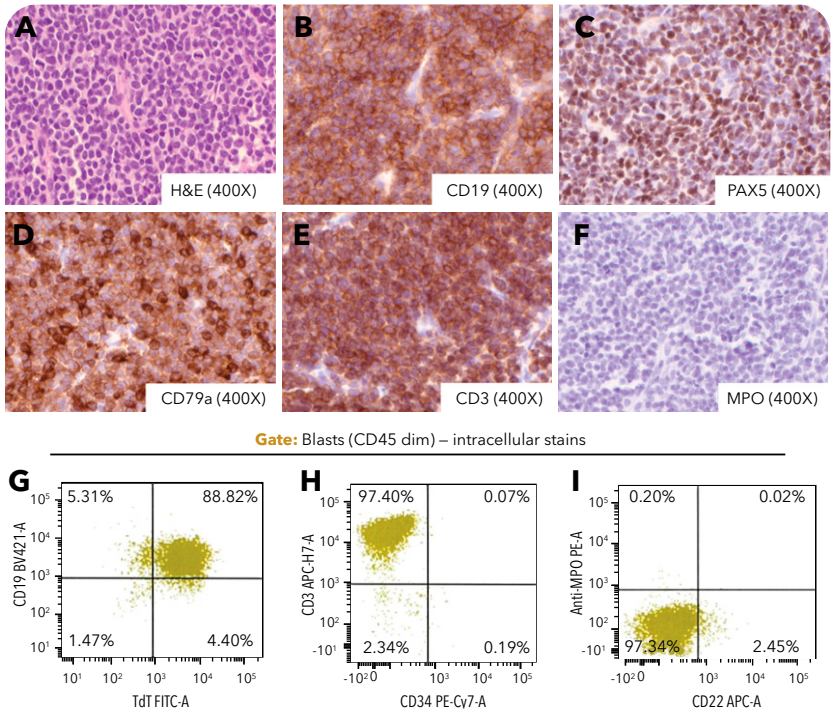
# Nonleukemic T/B mixed phenotype acute leukemia with *PHF6* and *NOTCH1* mutations

**Oscar Silva; Stanford University School of Medicine; and Allison Shaw, Kaiser Permanente Regional Laboratory Northern California**




A 24-year-old woman presented with cervical lymphadenopathy and a mediastinal mass. Complete blood count showed thrombocytosis (533 K/μL) and no other abnormality. Cervical lymph node biopsy showed sheets of immature mononuclear cells expressing TdT, CD19, CD79a, PAX5, CD3 and lacking MPO (panels A-F; hematoxylin and eosin stain [A], immunostains [B]; 40× objective, total magnification ×400). Flow cytometry showed blasts expressing TdT, cytoplasmic-CD3(strong), CD19(strong), cytoplasmic-CD79a and not expressing cytoplasmic-CD22, CD10, or MPO (panels G-I). Bone marrow showed 30% involvement by the same blast population. Cytogenetics showed gain of 9q, but fluorescence in situ hybridization was negative for *BCR-ABL1* and *MLL* rearrangements. Mutational analysis revealed *PHF6 Y105**, *NOTCH1 V1576E*, *NRAS A59D*, and *TP53 Y236D* pathologic mutations. Variants of unknown significance were identified in *JAK3*, *SUZ12*, and *IL7R*. Mutations in *FLT3* and *DNMT3A* were not identified.

This is a case of nonleukemic T/B–mixed phenotype acute leukemia (MPAL) presenting with adenopathy and a mediastinal mass. T/B-MPAL usually presents in young adults as lymphadenopathy, with only rare nonleukemic cases described. This is the second reported case of T/B-MPAL with concurrent inactivating *PHF6* and activating *NOTCH1* mutations, both mutations which promote T-cell differentiation at the expense of B-cell differentiation. Overall, the genetic aberrations of T/B-MPAL are similar to early T-cell precursor acute lymphoblastic leukemia with common mutations in *PHF6* and JAK-STAT pathway genes, however without mutations in *FLT3*, *DNMT3A*, *WT1*, or *EZH2*.

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