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Niemann-Pick disease with isolated leukemic nonnodal mantle cell lymphoma of the spleen

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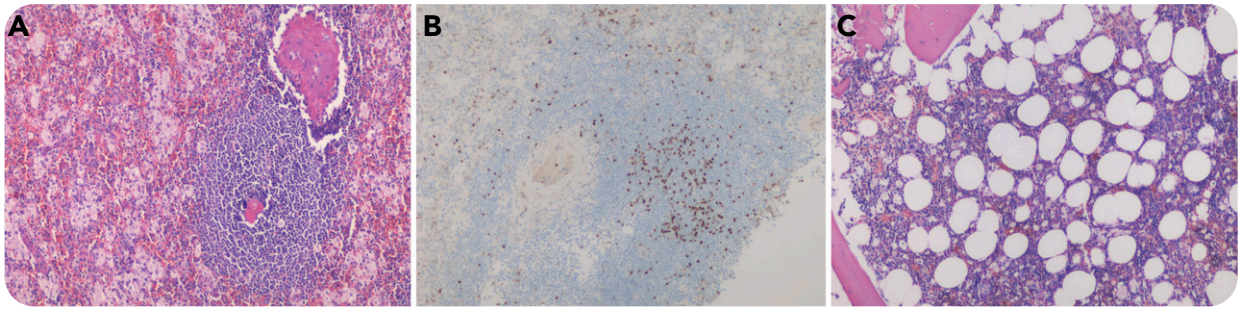

A 54-year-old man with Niemann-Pick disease type B with chronic transaminitis, thrombocytopenia, and splenomegaly for 21 years presented with abdominal discomfort. Magnetic resonance imaging revealed splenomegaly of 23.6 cm with splenic nodules. Splenectomy was performed to resolve discomfort. Laboratory test results were as follows: leukocytes, $6 \times 10^3/\mu L$; hemoglobin, 15 g/dL; platelets, $94 \times 10^9/L$; aspartate aminotransferase, 48 u/L; alanine aminotransferase, 71 u/L; total bilirubin, 1.1 mg/d; and alkaline phosphatase, 39 u/L. The splenectomy specimen was 1116 g. Microscopic evaluation of the spleen showed extensive red pulp infiltration by foamy macrophages and preserved white pulp (panel A; hematoxylin and eosin [H&E] stain; original magnification, ×100). Flow cytometry identified a monotypic $CD5^+$ B-cell population coexpressing CD5, CD11c, CD19, CD20, CD22, CD23 (dim), FMC-7, CD200 (subset), CD45, and monotypic κ surface immunoglobulin light chains. Cytogenetics revealed t(11;14)(q13;q32). Findings were diagnostic of mantle cell lymphoma. Immunostaining with cyclin-D1 demonstrated very subtle white pulp infiltration by neoplastic cells (panel B; cyclin-D1 stain; original magnification, ×100) with Sox 11^-. The bone marrow biopsy had foamy macrophages with cyclin-D1 immunostaining negative with dry aspiration (panel C; H&E stain; original magnification, ×100). Peripheral blood flow cytometry revealed no lymphomatous involvement. Positron emission tomography/computed tomography showed no evidence of FDG-avid neoplasm. Serum paraproteinemia was not evident.

This case illustrates the unusual coexistence of Niemann-Pick disease, a lysosomal storage disorder, with an indolent leukemic nonnodal variant of mantle cell lymphoma. The patient is being followed expectantly.

DOI 10.1182/blood.2019003479