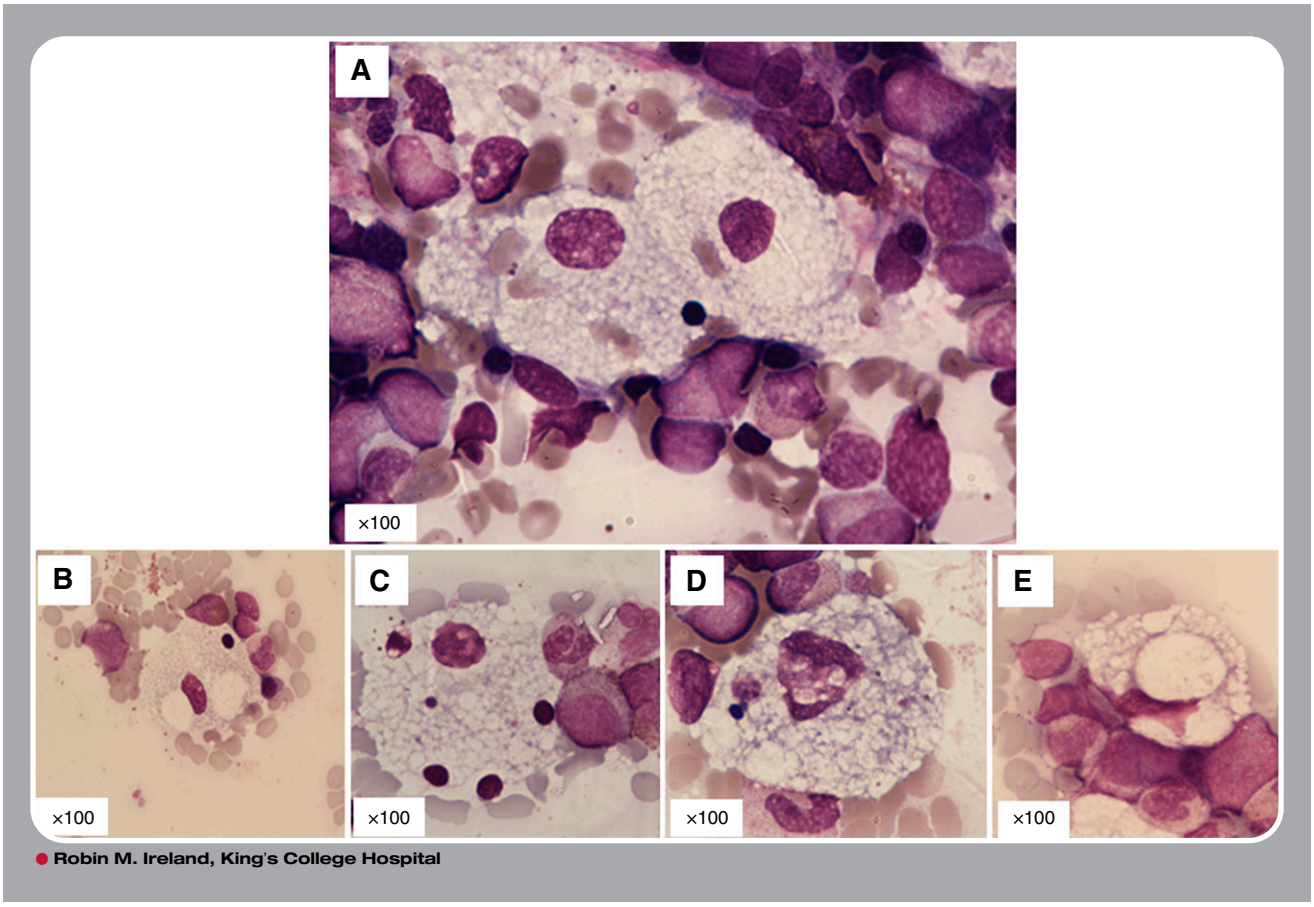
blood work
IMAGES IN HEMATOLOGY

Morphology of Niemann-Pick type C metabolic storage disorder



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A white girl, born of nonconsanguineous parents at term by spontaneous vaginal delivery, developed neonatal hepatitis with jaundice at 2 months. Jaundice resolved, but by 3 years of age she had developed hepatosplenomegaly. There were no neurological or dysmorphic features. Peripheral blood parameters were normal and there were no vacuolated peripheral blood lymphocytes. In the bone marrow aspirate, scattered large foamy macrophages were visible either around the edges of particles (panel A) or free in the trails (panels B-E). They contained variable-sized vacuoles, some with nuclear debris and inclusions. The features were most consistent with Niemann-Pick type C (NP-C), a neurovisceral metabolic disorder. Abnormal storage cells are only present in about 50% of cases and require observer expertise to identify. There were no "sea-blue" histiocytes as sometimes seen in this disease.

Though NP-C is clinically, biochemically, and genetically distinct from NP-A, differentiating between them by morphology is difficult. Storage cells in NP-C usually demonstrate greater variability in vacuole size, are more likely to contain nuclear debris or inclusions, and are absent of peripheral blood lymphocyte vacuolation. Definitive diagnosis of NP-C requires oxysterol measurement, filipin staining, or confirmation of *NPC1*, or more rarely *NPC2*, gene mutations.

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