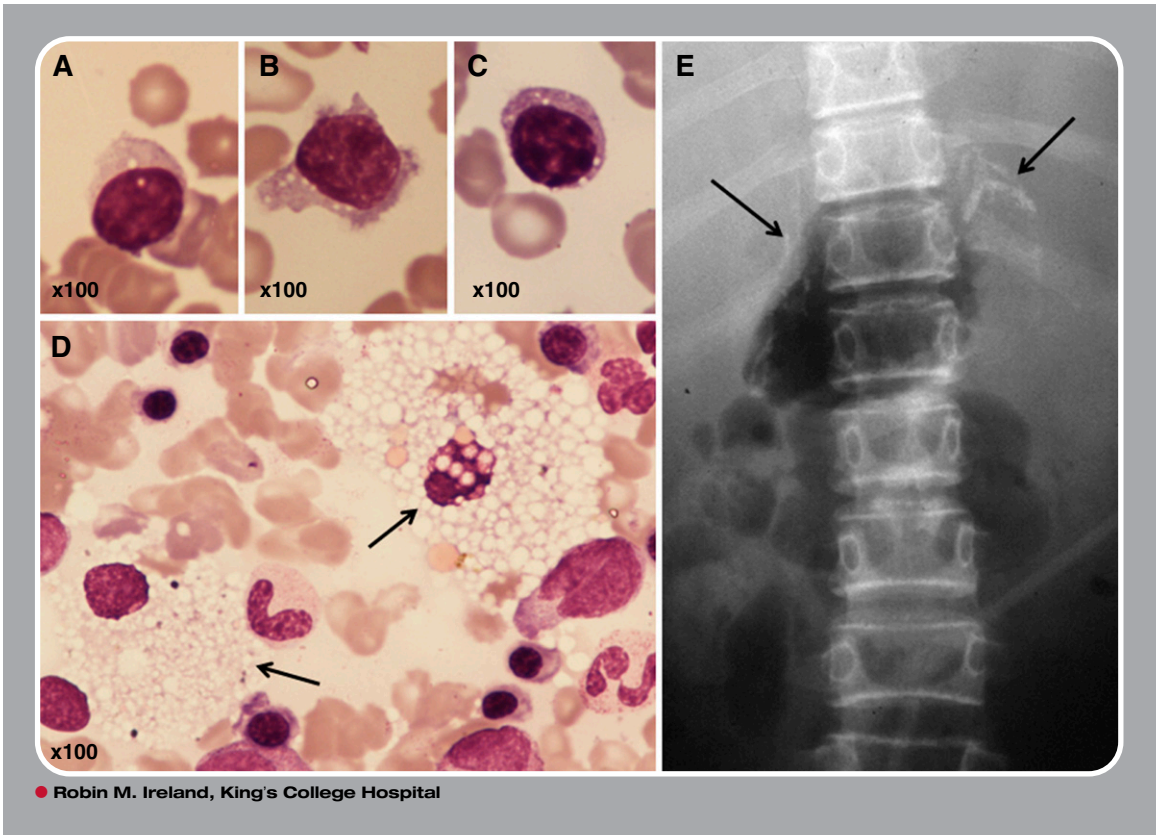
bloodwork
IMAGES IN HEMATOLOGY

Morphology of Wolman cholesteryl ester storage disease



A 21-year-old man had presented at 3 months of age with failure to thrive, malabsorption, diarrhea, weight loss, ascites, and hepatosplenomegaly. A diagnosis of Wolman disease (lysosomal acid esterase deficiency) was made following demonstration of excess cholesterol ester in the bone marrow, liver, and jejunal mucosa. Enzyme studies confirmed severe deficiency of lysosomal acid esterase and heterozygous levels in the parents. Parents were nonconsanguineous; a brother had died of the same disease in infancy, and 1 brother was unaffected. Despite severe malabsorption, the patient unusually survived into adulthood on a low-fat diet, vitamin supplements, exogenous cholesterol, clofibrate, and parenteral nutrition. Peripheral blood showed macrocytic anemia with mild neutropenia, thrombocytopenia, and acanthocytes. Approximately 20% of lymphocytes demonstrated between 1 and 7 cytoplasmic vacuoles (panels A-C; May–Grünwald–Giemsa stain). Marrow aspirate showed numerous abnormal foamy macrophages (panel D, arrows; May–Grünwald–Giemsa stain) that were also positive with oil red O and Cain's Nile blue, confirming cholesteryl ester storage substances. Abdominal radiography demonstrated characteristic bilateral adrenal calcification (panel E, arrows).

Lymphocyte vacuolation may be acquired but is a useful morphological feature to refine the differential diagnosis of abnormal storage cells associated with neurovisceral disease, being absent in Niemann-Pick type C, Gaucher disease, and GM1 gangliosidosis type 2.

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