blood work
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A "foamy" mastocytosis case

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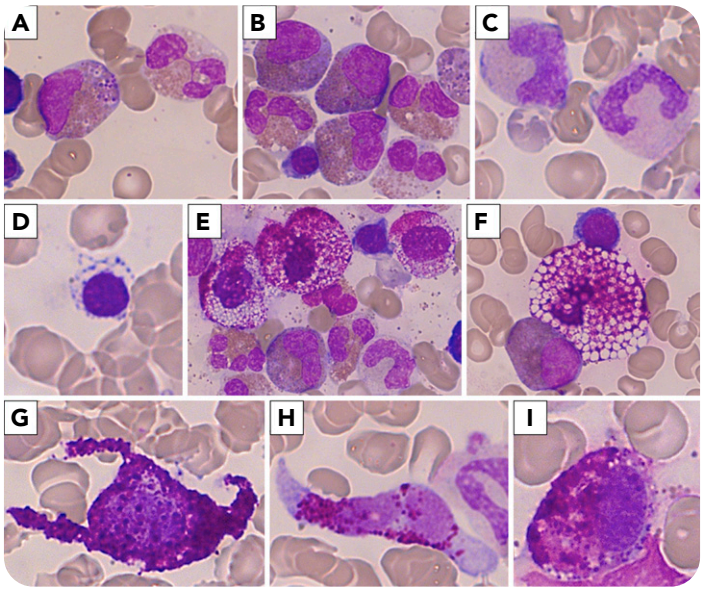

A 50-year-old man presented with flushes, fever, intestinal disorders, urticaria pigmentosa, and splenomegaly. Anemia and thrombocytopenia associated with hypereosinophilia (16.5 g/L) were found. Cytomorphological examination of a bone marrow aspiration revealed dystrophic eosinophilic precursors, erythroid and granular myeloid-associated dysplasia (panels A-D; in all panels, May-Grünwald-Giemsa stain; original magnification ×1000) related to a myelodysplastic syndrome with multilineage dysplasia (MDS-MD). It associated with a high infiltration (8%) of abnormal mast cells (MCs). Most MCs were degranulated, displaying a very atypical foamy aspect with large optically empty vacuoles, and were often grouped within islet-like cell clusters (panels E-F). Flow cytometry analysis showed aberrant expression of CD2 and CD25 on KIT^+/$FceRI^+$ MCs. The serum tryptase level was at 773 ng/mL. Trephine biopsy confirmed the diagnosis of aggressive systemic mastocytosis (ASM) with associated hematological neoplasm (AHN). Molecular biology found neither D816V *KIT* mutation nor any mutation on panexonic sequencing of the *KIT* gene. Screening for *BCR-ABL1* and *FIP1L1-PDGFRA* was negative. The disease evolved 9 months later into MC leukemia refractory to many lines of treatment, and the patient died 2 years after the diagnosis.

This is a very unique case of an ASM-AHN (MDS-MD) with foamy MCs, contrasting with classical cytomorphology of spindle-shaped (panels G-H) or hypogranulated MCs (panel I) also present to a lesser extent.

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