



## THE ANSWER

### **Diagnosis: Gaucher's Disease**

Suspected by morphology and confirmed with genetic testing. The marked expansion of red pulp by a large number of histiocytes with a finely fibrillary cytoplasm appearing “crinkled or wrinkled paper-like” is morphologically classic for a diagnosis of Gaucher's disease. Genetic testing confirmed the diagnosis with a markedly reduced activity of beta-glucosidase, supporting the diagnosis. Followup bone biopsy to evaluate the extent of the disease unfortunately confirmed involvement of the active bone marrow.

Flow cytometry testing was normal on the bone marrow specimen. Classic representation of Bone Marrow involvement of Gaucher's can be seen in the figure below, with abundant, finely fibrillary, pale blue-gray cytoplasm that is crinkled or wrinkled paper-like.



