Hemepath Case 21: 9-Year-Old Boy

HISTORY

A 9-year-old boy of Ashkenazi Jewish ancestry complains of pain in his left arm. There has been no history of trauma or injury.

For the past year, the patient has been taking increasingly longer naps in the afternoons as he always feels tired. He has stopped playing sports with his friends as he feels out of breath when he runs. About 8 months ago, he began having regular nosebleeds (about 1 episode per month) and acquires frequent bruises on his legs, although he doesn't remember injuring himself.

On physical examination, the patient appears pale and listless. Localized tenderness and swelling are noted on his left forearm, and he winces with pain during gentle palpation. There is no skin breakage. Additionally, several ecchymoses are noted on the anterior part of his shins bilaterally. The spleen is found to be severely enlarged, with the tip protruding into the pelvic cavity.

CBC

Hgb (g/L)	Low
MCV	N
Reticulocyte Count	N
WBC	Low
Plt	Low

DESCRIPTION OF SLIDES

Bone Marrow Aspirate (Slide 21a)

There are numerous large, foamy macrophages (see circles), particularly at the feather edge of the slide. Some of these foamy macrophages have the classic "tissue paper cytoplasm".

Bone Marrow Biopsy (Slide 21b)

There are numerous eosinophilic macrophages (see circles) interspersed throughout the marrow space. They are morphologically benign and show variable foamy cytoplasm. Normal tri-lineage hematopoiesis is present in the background.

*** To see the slide annotations in Imagescope, click on VIEW, then ANNOTATIONS, and then on the "eye" icon adjacent to the word "Layers". In the "Layer Attributes" box, a brief description of the annotations is provided. You may also click on individual layer region (e.g. region 1) in the "Layer Regions" box to locate each annotation – this is especially helpful in identifying annotations when the slide is not zoomed in. ***

MORPHOLOGICAL DIAGNOSIS

Gaucher's disease

DISCUSSION

Gaucher's disease is a rare autosomal recessive condition in which deficient function of the lysosomal hydrolase, β-glucocerebrosidase, results in buildup of glucocerebroside in cells of the monocyte-macrophage system. It is most frequently found in patients of Ashkenazi Jewish descent, and can be further classified based on clinical symptoms and signs. Type I is the most common form of this condition and can present with anemia, thrombocytopenia, splenomegaly, and skeletal disease, as is evident in our patient.

Histologically, Gaucher's disease is characterized by lipid-laden macrophages with a crumpled cytoplasmic appearance and a displaced nucleus (Gaucher cells). Glycolipids can also accumulate in the bone marrow (resulting in pancytopenia and giving rise to bony infarcts and pathologic fractures) and in other organs, such as the liver and the spleen (producing hepatosplenomegaly).