Hemepath Case 22: 1-Year-Old Boy

HISTORY

A 1-year-old boy is brought in by his mother. He has not gained adequate weight and is noticeably shorter than the other children of his age. He also does not respond when his name is called, and still has not spoken his first words. Over the past few months, she has noticed a “hump” in his lower back which has gradually increased in size. She also remarks that his facial features seem to be changing and he no longer resembles his parents. The boy was born with an umbilical hernia and has had chronic rhinitis “his entire life”. He also had an ear infection 3 months ago.

On physical examination, the boy is noted to have a prominent forehead, large eyes with marked corneal clouding, a flattened nasal bridge, and a large tongue. He is unresponsive to both auditory and visual stimuli. The rest of the exam reveals hepatosplenomegaly and mild deformation of the lower spine and pelvis.

CBC

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

DESCRIPTION OF SLIDE

Peripheral Blood Smear

The peripheral blood smear shows vacuolated lymphocytes (see circles). The degree of vacuolation is suggestive of a storage disease. RBCs are normal. Severe thrombocytopenia is also seen.

*** 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

Storage disease (special testing confirms Hurler’s syndrome)

DISCUSSION

This degree of lymphocyte vacuolation is more than would be expected in a reactive state (such as a viral infection). Instead, it suggests a storage disease, in which abnormal lipids, for example, accumulate within lymphocyte cytoplasm. Lymphocyte vacuolation is not specific for a single storage disease.

This case happens to be Hurler's syndrome (mucopolysaccharidosis type IH), an autosomal recessive disorder with deficient α-L-iduronidase, an enzyme which normally degrades the glycosaminoglycans dermatan sulfate (DS) and heparin sulfate (HS). The glycosaminoglycans subsequently accumulate in lysosomes of various organs, giving rise to characteristic facial features (classically described as gargoyle-like) and physical anomalies (e.g. hepatosplenomegaly, skeletal deformities), sensory loss (e.g. visual defects, hearing loss), and mental retardation. Histologically, numerous cytoplasmic granules of glycosaminoglycan are seen in lymphocytes.