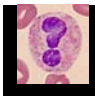

Hemepath Case 17: 5-Year-Old Boy

HISTORY

A 5-year-old Chinese-Canadian boy presents with recurring abdominal pain localized to the right upper quadrant. The discomfort is not associated with meals. Past medical history is unremarkable except that the boy's skin and sclera have "always" been slightly jaundiced.

Physical exam reveals a mildly jaundiced boy in no apparent distress. On abdominal exam, an enlarged, firm, non-tender spleen is palpated 7 cm below the left costal margin. A positive Murphy's sign is also elicited.

CBC

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

OTHER LABORATORY FINDINGS

Osmotic Fragility Test	N
Coombs Test	(-)

DESCRIPTION OF SLIDE

Peripheral Blood Smear

The peripheral smear shows mild anemia, polychromasia, and moderate poikilocytosis, mostly of small, shrunken acanthocytes (sputnik cells – see circles). There is also evidence of hyposplenism, such as Howell-Jolly bodies (see rectangles) and Pappenheimer bodies (see arrow). Leukocytes and platelets are unremarkable.

*** 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

Pyruvate kinase deficiency

DISCUSSION

Pyruvate kinase (PK) deficiency is an autosomal recessive disorder more commonly found in patients of northern European and Chinese ancestry. Normal RBCs lack mitochondria, and are therefore dependent on the anaerobic glycolytic (Embden-Meyerhof) pathway for ATP generation. ATP, in turn, provides an energy source to maintain ion gradients across the cell membrane. Erythrocytes with a defective PK enzyme have reduced ATP levels, and thus become dehydrated, shrunken, and prematurely destroyed. In this boy, increased RBC destruction is evident via jaundice, splenomegaly, and findings suggestive of cholelithiasis (a positive Murphy's sign indicates cholecystitis).

PK deficiency is proven by an enzyme spot test.

In addition to low ATP levels, intermediate products in the glycolytic pathway can also accumulate: one such product is 2,3-diphosphoglycerate (2,3-DPG). 2,3-DPG can shift the oxygen dissociation curve to the right, allowing RBCs to release oxygen more readily. This is why patients with PK deficiency may have markedly low Hb levels, but only mild symptoms of anemia.