Hemepath Case 14: 7-Year-Old Girl

HISTORY

A 7-year-old girl presents with an upper respiratory tract infection. Her mother is concerned that the girl's eyes have turned "yellow". This has happened before several times, and always occurred when the girl was sick with a viral infection. The mother also comments that her child is paler than the rest of the family members, and tires easily when playing outdoors. Birth history is unremarkable except for neonatal jaundice that lasted for 1 week. Family history reveals that the family migrated from Norway 5 years ago, and the father had a splenectomy in his 20s.

Splenomegaly is noted on physical examination.

CBC

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

OTHER LABORATORY FINDINGS

Osmotic Fragility Test	Increased fragility
Flow for Eosin-5-maleimide	Reduced fluorescence

DESCRIPTION OF SLIDE

Peripheral Blood Smear

There is moderate anemia with severe spherocytosis (see circles) and moderate polychromasia. Leukocytes and platelets are normal.

*** 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

Hereditary spherocytosis

DISCUSSION

Hereditary spherocytosis (HS) is the most common genetic cause of hemolytic anemia in patients of northern European origin. Proteins involved in anchoring the RBC cytoskeleton to the lipid membrane, such as spectrin, ankyrin, band 3, and protein 4.2, are defective in this condition. Unsupported portions of the membrane are easily lost, and the biconcave erythrocytes gradually become spherical. These spherocytes are less deformable, and become trapped and prematurely destroyed in the splenic microvasculature.

Depending on the severity of the condition, patients may be asymptomatic, or have signs of anemia, splenomegaly, and intermittent jaundice (especially following viral infections in pediatric patients). Those with a severe form of HS may present with cholelithiasis or a hemolytic crisis.

This condition used to be diagnosed with the aid of osmotic fragility testing, but more recently a flow cytometry test (eosin-5-maleimide) is used.