Hemepath Case 18: 13-Year-Old Girl

HISTORY

A 13-year-old girl presents with jaundice, marked hepatosplenomegaly, and prominent frontal bossing. Both parents are immigrants from Thailand.

CBC

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

OTHER LABORATORY FINDINGS

Bilirubin	High
Serum Fe	N
Serum Ferritin	N

DESCRIPTION OF SLIDES

Peripheral Blood Smear (Slide 18a)

The peripheral blood smear shows mild anemia with microcytosis (see circles), polychromasia, and RBC targeting (see rectangles). Note that there is no significant elliptocytosis, anisochromasia, or anisocytosis – findings that would suggest iron-deficiency anemia.

HbH Preparation (Slide 18b)

HbH preparation demonstrates the presence of H bodies (see circles).

*** 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

Alpha thalassemia

DISCUSSION

α-thalassemia is a group of hereditary disorders characterized by reduced synthesis of α-globin chains, and most frequently affect patients of Asian, African, and Mediterranean ancestry. Two α-globin chains are located on each chromosome 16; therefore, in a normal, healthy individual, 4 α-globin genes are expressed (αα/αα). In α-thalassemia, the severity of symptoms depends on the number of α-globin genes deleted, and ranges from asymptomatic in silent carriers (1 gene deletion, αα/α-) to intrauterine death in Hb Bart's/hydrops fetalis (all 4 genes deleted, --/--).

In hemoglobin H (HbH) disease, loss of 3 α-globin genes (--/α-) leads to an unbalanced ratio of α chains and β chains. The excess β-globin chains aggregate and form a homotetramer (HbH, β_4), which can precipitate and form inclusion bodies within RBCs. These abnormal erythrocytes are unstable and have an increased susceptibility to hemolysis when under oxidative stress. They also have poor oxygen carrying capacity.

Frontal bossing (prominent forehead) is seen in this patient as a sign of bone marrow hyperplasia, similar to that seen in Case 13.