Hemepath Case 12: Newborn Male

HISTORY

A newborn male, of African descent on his father's side (mother is Caucasian), is severely jaundiced. The pregnancy and delivery were unremarkable. Family history reveals that the father has G6PD deficiency and hereditary elliptocytosis.

The baby is now 2-day-old with a very high bilirubin level.

CBC

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

DESCRIPTION OF SLIDE

Peripheral Blood Smear

The peripheral blood smear shows moderate anemia with elliptocytosis (see rectangles) and pyropoikilocytosis (note RBC fragments with little "thumbs" projecting out; see circles). 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

Neonatal poikilocytosis elliptocytosis syndrome

DISCUSSION

Neonatal poikilocytosis elliptocytosis syndrome (NPES) is usually found in children of African ancestry, in families which have hereditary elliptocytosis (HE). Patients exhibit moderate to severe hemolytic anemia, with findings of elliptocytosis and pyropoikilocytosis in the peripheral smear. The anemia becomes less severe after the first 6-12 months of life, and the condition gradually evolves into mild HE without pyropoikilocytosis on the peripheral smear.

The pathogenesis for this syndrome appears to be related to the increased level of 2,3-diphosphoglycerate (2,3-DPG) in fetal erythrocytes. 2,3-DPG weakens interaction between spectrin, actin, and protein 4.1, intensifying the self-association defect in spectrin (the defect is a consequence of the genetic mutation found in HE). Resultant red cells are fragile and have an increased susceptibility to fragmentation and hemolysis.

In this case, the father's history of G6PD deficiency led to the infant being misdiagnosed with G6PD deficiency. Genetically this is impossible (barring a new mutation): G6PD is X-linked, so it cannot under normal circumstances be transferred from fathers to sons.