Hemepath Case 1: 5-Year-Old Boy

HISTORY

A 5-year-old boy presents with exquisitely painful fingers and toes. The family emigrated from West Africa 6 months ago.

CBC

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

DESCRIPTION OF SLIDE

Peripheral Blood Smear

The peripheral smear shows anemia with polychromasia, poikilocytosis, and anisocytosis. RBCs are sickled (see rectangles) – note both the crescent shape and the "boat"-like form. Hyposplenism is indicated by the presence of target cells (see circles) and Howell-Jolly bodies (see arrows). WBCs 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

Sickle cell disease

DISCUSSION

Sickle cell disease is a hereditary condition characterized by abnormal sickle hemoglobin (HbS) from a point mutation in the β-globin chain. In patients with sickle cell *disease* (also known as sickle cell anemia), both β-globin chains are abnormal; in patients with sickle cell *trait*, only one β globin is affected while the other is normal. At low oxygen tension, HbS polymerizes into insoluble cross-linking fibers, and the erythrocytes become sickled and trapped in the splenic vasculature. These are quickly cleared by reticuloendothelial macrophages, thereby reducing the life span of the erythrocytes. Because the rate of RBC production cannot keep up with the rate of RBC destruction, anemia results.

Sickled erythrocytes can become trapped in the small blood vessels of the hands and feet, preventing oxygen transport to the bones and bone marrow. This gives rise to painful ischemic necrosis of the marrow known clinically as “Hand-Foot” Syndrome.

Sickle cell trait is found in up to 40% of some African populations, as it offers resistance against malaria. RBCs in patients with sickle trait are generally immune to sickling because there is enough HbA in the RBCs to prevent this process.