Hemepath Case 10: 2-Month-Old Girl

HISTORY

A 2-month-old girl is brought in by her mother. The baby has been lethargic, irritable, and has not gained adequate weight. There is no history of illnesses or any episodes of fever, diarrhea, or vomiting. The girl was the product of a healthy pregnancy and full-term home delivery; however, an atrial septal defect was diagnosed at birth.

On physical exam, the patient appears pale and listless. She is noted to have wide-spaced eyes, a short, upturned nose, and a thick upper lip. Both thumbs are subluxed.

CBC

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

DESCRIPTION OF SLIDES

Peripheral Blood Smear (Slide 10a)

There is anemia with numerous round and oval macrocytes (see rectangles). A mild, non-specific poikilocytosis is also noted, with occasional teardrop cells (see arrows) and rare target cells (see circles). There is no polychromasia. Leukocytes and platelets are unremarkable.

Bone Marrow Aspirate (Slide 10b)

There is a relatively normal production of myeloid cells. However, erythropoiesis is extremely limited.

Bone Marrow Biopsy (Slide 10c)

The biopsy is very cellular, with plentiful megakaryocytes and numerous mature lymphoid cells. Granulopoiesis is also evident. However, there is virtually no erythropoiesis – this is known as uni-lineage marrow aplasia.

*** 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

Diamond-Blackfan syndrome

DISCUSSION

Diamond-Blackfan syndrome (DBS) is a rare congenital disorder in which abnormal differentiation of erythroid progenitor cells results in severe hypoplastic anemia. Children exhibit signs of anemia (e.g. fatigue, irritability) and may also have craniofacial dysmorphism, limb/radial malformations, and/or cardiac defects. Most patients are diagnosed before reaching age 1. If a bone marrow biopsy is performed, a few early erythroblasts with maturation arrest may be observed.

The other major diagnostic consideration is Transient Erythroblastopenia of Childhood (TEC). This patient's physical abnormalities, as well as her young age, make DBS more likely. Patients with TEC will invariably improve with only supportive care, whereas DBS does not correct on its own.