Hemepath Case 48: Newborn Female

HISTORY

A female newborn, the product of an uneventful pregnancy, presents with physical features consistent with trisomy 21, and is confirmed to have Down syndrome via karyotype analysis. Upon further examination, the baby is noted to be slightly hypoxic, with mild peripheral edema as well as hepatomegaly. A routine CBC is performed.

CBC

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

DESCRIPTION OF SLIDE

Peripheral Blood Smear

There are many circulating blasts (see circles) and a severe increase in WBC count. Also evident are numerous nucleated erythrocytes, some of which appear dysplastic (see rectangles). Platelets are increased and abnormal (hypogranular and giant; see arrows). These findings are suggestive of a myeloproliferative disorder.

*** 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

Transient myeloproliferative disease of the newborn

DISCUSSION

Transient myeloproliferative disease of the newborn (TMD) is an uncommon condition seen in patients with trisomy 21, and has been shown to be associated with mutations in the GATA1 gene on the X chromosome. GATA1 is a transcription factor involved in normal maturation of RBCs and megakaryocytes. Very rarely, TMP is also seen in non-Down newborns: these infants may have mosaic Down syndrome, with trisomy 21 only in their marrow but not in skin fibroblasts or other cells.

Patients with TMD exhibit marked leukocytosis in their peripheral blood with circulating blast cells of megakaryocytic origin. In severe cases, blast cells may infiltrate and cause failure of organs (e.g. heart, lungs, liver), with lethal consequences. However, for the majority of patients, the hematological abnormalities resolve on their own within a few weeks or months. Some patients go on to develop acute leukemia later in life, particularly acute megakaryoblastic leukemia (AML FAB M7).