Hemepath Case 42: 13-Year-Old Boy

HISTORY

A 13-year-old boy presents with a history of “easy bruising”. You complete a full history, and learn that there is blood in the patient’s urine from time to time; additionally, the boy has been previously diagnosed with sensorineural deafness. The patient’s mother and grandfather have similar problems.

CBC

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

DESCRIPTION OF SLIDE

Peripheral Blood Smear
RBCs and WBCs are normal. Platelets are low in number. Platelet size is considerably enlarged – many giant platelets (see circles) are visible, although occasional normal-sized platelets can also be seen. Döhle bodies (see arrow) are present, but these are difficult to visualize on the digitized slide.

*** 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

Fechtner syndrome

DISCUSSION

Fechtner syndrome is one of the giant platelet syndromes (GPS). As with the other GPS, patients with Fechtner syndrome have large and giant platelets as well as thrombocytopenia.

Fechtner is one of a group of disorders (along with the more common May-Hegglin anomaly, Sebastian syndrome, and Epstein syndrome) caused by defects in the MYH9 gene. These all share the findings of giant platelets and Döhle-like inclusions within neutrophils (although Döhle-like bodies are not observed in every case). Fechtner

cases generally also present with cataracts and sensorineural deafness. Fechtner and Epstein patients often present with nephritis.

Given the common genetic basis of this group of GPS, differentiation may be made clinically.