



Hemepath Case 20: 3-Year-Old Girl

HISTORY

A 3-year-old girl who recently emigrated from Puerto Rico is brought in by her mother over concerns that the child is not gaining adequate weight. She is always tired, and sleeps through most of the day. The girl is not a picky eater and enjoys foods from all food groups. However, she is known to have tropical sprue and has only begun receiving treatment recently.

On physical examination, the girl appears tired, quite thin, and slightly jaundiced. The corners of her mouth are red and cracked, and her tongue is very red and swollen. Mild bruising is observed on her lower legs. No lymphadenopathy is noted. Findings from the neurological examination are normal.

CBC

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

DESCRIPTION OF SLIDE

Bone Marrow Aspirate

There is evidence of erythroid hyperplasia and megaloblastic changes (see circles), such as nuclear-cytoplasmic dyssynchrony and irregularly-shaped nuclei. Megakaryocytes (see arrows) are large and hyperlobulated. Granulocytic maturation shows giant metamyelocytes (see rectangles).

*** 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

Megaloblastic anemia

DISCUSSION

Megaloblastic anemia is a group of anemias in which impaired DNA synthesis leads to marrow erythroblasts with delayed nuclear maturation (relative to the cytoplasm). It is most commonly due to vitamin B12 and/or folate deficiency from reduced dietary intake, poor absorption (e.g. tropical sprue), or impaired utilization.

Vitamin B12 and folate are essential co-factors in purine synthesis; their deficiency prevents maturation of the nucleus and consequently impairs mitosis, not only in blood cells (thus giving rise to pancytopenia), but also in other body cells. This is particularly evident in cells with rapid turnover, such as the GI epithelium, and presents as glossitis and angular cheilosis in our patient. Progressive neuropathy resulting from demyelination of peripheral sensory nerves may also be observed in severe vitamin B12 deficiency, although this is not seen in our case.