

Gastrointestinal and Liver Pathology at Rush

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Case of the Month Answer- April 2009

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Diagnosis: Blue rubber bleb nevus syndrome

Blue rubber bleb nevus syndrome (BRBNS) is a rare vascular anomaly syndrome consisting of multifocal venous malformations (VM). The malformations are most prominent in the skin, soft tissues, and gastrointestinal (GI) tract, but can occur in any tissue. This association of “hemangiomas” of the skin and GI tract was first reported in 1860, and characterized by William Bean in 1958, giving rise to the name “Bean syndrome.” The cutaneous lesions of BRBNS are small, usually measuring less than 1–2 cm, and blue to purple in color. Bean was the first to describe the unique quality of these compressible cutaneous lesions that he called “blue rubber-bleb nevi.” A patient may have from several to hundreds of cutaneous lesions. Nonetheless, the visceral organ system most commonly affected is the GI tract and these lesions are more clinically relevant than the skin and soft tissue lesions. Vascular malformations may occur anywhere from oral to anal mucosa but predominantly occur in the small bowel.

Histopathologic examination of small bowel lesions reveals blood-filled ectatic vessels, lined by a single layer of endothelial cells, with surrounding thin connective tissue (Figures 1 and 2).

In contrast to the skin lesions, the GI lesions often bleed. They may spontaneously rupture causing acute hemorrhage and death. However, most bleeding from the GI tract is slow, minor, chronic, and occult, resulting in iron deficiency anemia from ongoing loss. A case of thrombocytopenia and disseminated intravascular coagulation has been reported in association with BRBNS. Other complications include intussusception, volvulus, and bowel infarction. These diagnoses should be considered in patients with BRBNS and abdominal pain.

References:

Odze R., Goldblum J. and Crawford J. Surgical Pathology of the GI tract, Liver, Biliary Tract and Pancreas. 2004. p.74.

McKee P. Pathology of the Skin. Second edition. Mosby-Wolfe, 1996, pp. 16.63.