Lesion on Forearm

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A 42-year-old woman presents to the office with a tender lesion on her forearm of one month's duration.

Examination reveals multiple, soft, purple-blue papulonodules on the volar aspect of the left forearm (Figure 1). Inquiry reveals that she has had these lesions since early childhood. Functional inquiry does not reveal any history of neurologic complaints, fatigue, melena nor hematochezia.

The symptomatic lesion was excised and pathology reveals a cavernous hemangioma with thrombosis (Figure 2).

Complete blood count was within normal limits. Stools for occult blood was negative. Colonoscopy was unremarkable.

Figure 1: Lesion on patient's forearm

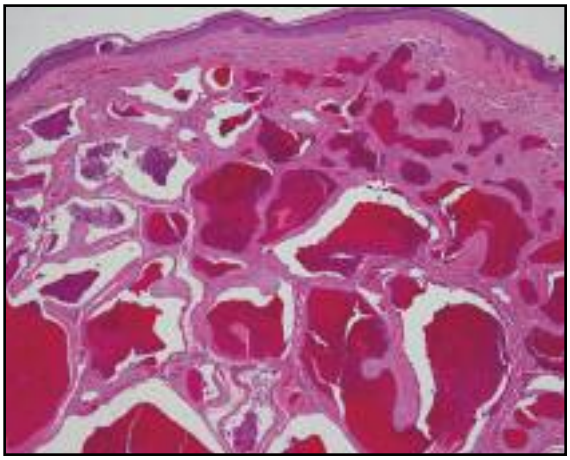

Figure 2: Histology of the lesion

What is your diagnosis?

a. Epidermal nevus syndrome
b. Blue rubber bleb nevus syndrome (BRBNS)
c. Nevoid basal cell syndrome
d. Cowden disease – multiple hamartomas

Answer: Blue Rubber Bleb Nevus Syndrome

About Blue Rubber Bleb Nevus Syndrome (BRBNS)

The blue rubber bleb nevus syndrome (BRBNS) is a rare (less than 160 cases reported), idiopathic disorder characterized by multiple venous malformations of the skin and viscera, most commonly the gastrointestinal tract.

BRBNS was first described by Gascoyen in 1890, although Bean published a treatise on the entity and coined the present name in 1950. The majority of cases are sporadic in nature, although familial cases have been reported. There is no sexual predilection.

These cutaneous vascular malformations typically present in infancy and early childhood. They present as highly characteristic, purple-red to dark-blue, easily compressible papulonodules, which give the syndrome its colourful name (Figure 1). The size of each " bleb" ranges from several millimeters to several centimeters in diameter. Histology reveals blood filled, ectatic channels lined by a single layer of endothelium (Figure 2). Some lesions may also be associated with localized hyperhidrosis. Patients may have several to over one hundred lesions. The hemangiomas tend to increase in number and size over time. Larger cavernous hemangiomas may cause disfigurement and compression of vital organ systems.

The most common extracutaneous involvement is the gastrointestinal tract, particularly the small bowel. Patients typically present with symptoms of iron deficiency secondary to chronic, insidious GI bleeding. Rarely, spontaneous rupture of the lesions can result in severe, and occasionally fatal, hemorrhage. The astute clinician will perform a thorough skin exam of a patient who presents to the emergency room with life-threatening GI bleeding.

The musculoskeletal system may also be affected, resulting in bone deformities, articular derangement and even spinal compression. BRBNS has also been reported to affect the CNS, solid organs (*i.e.*, liver, kidney, spleen and thyroid), and genitourinary tract.

Fortunately, most patients have a normal life expectancy, although surveillance and monitoring of affected systems are required. Basic investigations include CBC, ferritin level, fecal occult blood tests and urinalysis. Imaging studies for bone involvement may demonstrate bony overgrowth or fractures. Although radiographic contrast studies may delineate bowel involvement, GI endoscopy is the favoured approach. MRI may be used as a noninvasive screening and evaluative tool.

Symptomatic cutaneous lesions may be treated with excision, curettage, cryotherapy and laser ablation. Medical treatment of GI involvement includes iron supplementation and transfusions. Endoscopic removal and coagulation of symptomatic lesions may be required. Segmental bowel resection may be indicated for extensive involvement by lesions, although there is still a risk of recurrence. The prognosis is thus determined by the extent of systemic involvement.

Take Home Message

Although BRBNS is a rare condition, it is nonetheless an important entity to recognize because of the risk of serious and fatal hemorrhage. Any patient with unexplained, acute GI bleeding presenting to the ER should have a thorough cutaneous exam to rule out this syndrome. Dx

Resource
1. Oranje AP: Blue Rubber Bleb Nevus Syndrome. Pediatr Dermatol 1986;3(4):304-10.

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