Blue rubber bleb nevus syndrome (BRBNS) can present with vascular malformations throughout the body, especially in the gastrointestinal tract. Spinal cord compression from these lesions is rare, particularly in the pediatric population. The authors report a case of BRBNS involving an 18-year-old female patient who presented with back pain and an epidural thoracic mass with cord compression. She underwent an uncomplicated thoracic laminectomy and decompression, with removal of what appeared to be a venous malformation. Postoperatively her pain improved, and imaging revealed resolution of cord compression. Pathological analysis highlighted dilated venous channels with myxoid degeneration in the wall with clot, characteristic of BRBNS. The early age of presentation and location are unique based on the literature search of BRBNS. The present report highlights the multiplicity of venous malformations in BRBNS, and the management of this case.

**Key Words** • blue rubber bleb nevus syndrome • spinal cord compression • vascular malformation • spine • oncology • vascular disorders

### Case Report

**History and Presentation.** This 18-year-old Caucasian female with BRBNS diagnosed in early childhood presented with the chief complaint of pain between her shoulders, as well as left shoulder and arm pain. The pain between her shoulders was midline, with no radiation, and was aggravated with supine position. The patient also had a large venous malformation of the face and had undergone prior gastrointestinal surgeries for intussusception secondary to vascular malformations.

Her physical examination was remarkable for multiple painless bluish colored nevi throughout the trunk and extremities (Fig. 1). These lesions were firm and rubbery to palpation. Neurological examination revealed good symmetric motor strength, with mild hyperreflexia in the lower extremities, and decreased sensation in the upper thoracic dermatomes.

**Imaging.** MRI revealed a posterior epidural thoracic spinal lesion spanning T1–T3. It appeared T1 iso- to hypointense and T2 hyperintense, with minimal contrast enhancement. The spinal cord was displaced to the right, with no evidence of a syrinx (Fig. 2). The lesion was more eccentric to the left side and displaced the left T-2 nerve root anteriorly. Signs of extramedullary hematopoiesis were also present at the skull base and within the vertebral...
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Operation, Histopathological Findings, and Postoperative Course. The patient underwent an uncomplicated laminectomy from T-1 through T-3. Intraoperatively the lesion was noted to be thrombosed with a dark bluish appearance and was easily elevated off the dura mater. When the lesion was entered, thrombosed blood emanated under low pressure, facilitating removal and visualization of the underlying dura, which was not infiltrated. Histopathological examination showed fibroadipose tissue with a proliferation of venous channels, many with myxoid degeneration, admixed with thick-walled arteries. A larger vein contained blood clot with early organization. Focal hemorrhage into the surrounding tissue and fragments of viable bone and cartilage were also present (Fig. 3). After surgery, the patient was neurologically intact, with resolution of her pain. She was discharged home on postoperative Day 2.

Discussion

Blue rubber bleb nevus syndrome (BRBNS) is a rare condition with fewer than 200 documented reports. First described in 1860 by Gascoyen and later named by William Bean in 1958, BRBNS is also known as Bean syndrome. The condition is characterized by blue rubbery-appearing nevi throughout the skin. These skin lesions are vascular malformations that will blanch with pressure. Thrombosis of the vascular malformations can occur due to slow blood flow, and flow voids may not be present on imaging. Gastrointestinal vascular malformations are a hallmark and typically present in adolescence, and they frequently bleed. The skin vascular malformations are typically present at birth and multiple other organ systems involvement has been described, including muscle, bone, central nervous system, visceral pericardium, thyroid, heart, and lung. Paravertebral muscle involvement can extend to the spinal canal. Vascular lesions of the central nervous system have included cavernoma, dural arteriovenous fistula, arteriovenous malformations, and vein of Galen thrombosis and malformation. While gastrointestinal hemorrhage and consumptive coagulopathy can be life threatening, surveillance for malignancy is important in patients with BRBNS, as medulloblastoma, chronic lymphocytic leukemia, renal cell carcinoma (hypernephroma), and squamous cell carcinoma have been reported. Although most cases of BRBNS are sporadic, autosomal dominant inheritance with linkage to chromosome 9 has been described. MRI is the imaging modality of choice for identification of the lesions of BRBNS. Histological examination of vascular lesions will show thin vascular channels lined by a single layer of flattened endothelial cells. Treatment of BRBNS is aimed at controlling gastrointestinal hemorrhage with surgery, endoscopy, and medications, including interferon, steroids, and octreotide. With timely treatment of symptoms, children with BRBNS can have normal life spans and intelligence quotients.

We have been able to identify 3 other reports of spinal cord compression in the setting of BRBNS, and only 2 of these reports have been due to vascular malformations. The first report describes a 70-year-old man with thoracic epidural compression and leg pain and weak-
ness, the second report describes a 50-year-old man with pain and weakness following a minor back injury due to collapse of a vertebral hemangioma of T-4 with extension into the epidural space, and the third report describes an asymptomatic 36-year-old pregnant woman with cervical compression. The symptomatic lesions were treated surgically.

**Conclusions**

The lesions of BRBNS can have a multifocal presentation based on the literature. Spinal canal involvement of BRBNS is uncommon; however, it can represent a true neurological emergency if not recognized and managed appropriately.

**Disclosure**

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**References**

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