Blue rubber bleb nevus syndrome complicated by a ventricular septal defect: a case report (Post-print)

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Abstract

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Full Text

Preamble

Blue Rubber Bleb Nevus Syndrome Complicated by a Ventricular Septal Defect: A Case Report

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Abstract: The co-occurrence of blue rubber bleb nevus syndrome (BRBNS) and ventricular septal defects is rare. Here we present a case of BRBNS in a 15-year-old boy who was born with multiple cavernous hemangiomas and a ventricular septal defect. Examinations revealed the presence of hemangioma lesions in the subcutaneous and mucosal tissues as well as in the cerebrum, nasopharynx, tongue, esophagus, gastric body, sigmoid colon and adrenal gland.

Combined imaging modalities played an important role in the diagnosis of hemangioma lesions.

Key words: blue rubber bleb nevus syndrome; cavernous hemangioma; ventricular septal defect

Introduction

Cavernous hemangiomas result from localized defects in vascular morphogenesis occurring during the embryonic period. They represent one type of multiple venous malformations characterized by benign lesions and are common during infancy and childhood. Cavernous hemangiomas commonly involve the subcutaneous tissues and occur occasionally in the brain or visceral organs such as the liver and adrenal glands. The occurrence of congenital cutaneous and gastrointestinal hemangiomatosis was named blue rubber bleb nevus syndrome (BRBNS) by Gascoyen. Approximately 200 cases of BRBNS have been reported worldwide.

The lesions can appear throughout the whole body, and in some cases, BRBNS may co-occur with malignant or benign tumors. The co-occurrence of cavernous hemangiomas and congenital heart disease is particularly rare. So far, only three BRBNS patients with congenital heart disease have been reported, including the case described by Schneeweiss in 1982. In this report, we present a case of BRBNS complicated by ventricular septal defect in a 15-year-old boy.

Case Report

A 15-year-old boy presented with numerous masses on the body trunk and extremities at birth and was diagnosed with BRBNS. Surgical repair of his ventricular septal defect (VSD) was performed when he was one year old. At six months of age, he underwent surgery for lesions on his back and right wrist. He received subsequent Chinese medicine and injection therapies at four years of age, and was treated intermittently in recent years with propranolol. Some of his lesions disappeared while new ones appeared during his childhood.

At 14 years of age, the boy was 160 cm tall, weighed 45 kg and appeared inactive. He had pale face and lips, indicating severe anemia. Laboratory analyses revealed iron deficiency anemia, which was confirmed by bone marrow biopsy. The patient had normal blood platelets with leukocytes of 5.14×10 /L, hemoglobin of 41 g/L, mean corpuscular volume of 62.8 fL, mean corpuscular hemoglobin of 17.7 pg, Fe of 2.65 mol/L, transferrin saturation of 15.7%, and ferritin of 7.4 ng/L. Stool occult blood tests yielded positive results on sporadic occasions, while urinalysis was normal.

Computed tomography (CT) of the chest showed inflammation in both lungs. Abdominal ultrasound revealed a hemangioma lesion in the right adrenal gland,

while the liver, gallbladder, pancreas and kidneys were normal. Capsule endoscopy showed the presence of multiple hemangiomas in the small bowel. Gastroscopic and colonoscopic examinations revealed multiple vascular blebs in the gastric body and sigmoid colon [Figure 1: see original paper], and the esophagus also contained hemangioma lesions.

Magnetic resonance imaging (MRI) of the brain showed hemangioma lesions on both sides of the occipital lobe [Figure 1: see original paper] and in the parotid glands, facial subcutaneous tissue, nose and pharynx. Whole-body blood pool scintigraphy (WBBPS) using single-photon emission computed tomography (SPECT) with labeled red blood cells (RBCs) displayed multiple radioactive foci in the subcutaneous tissues of the trunk, extremities, and other organs including the bilateral parotids and small intestine [Figure 2: see original paper].

Discussion

Hereditary cutaneomucosal venous malformations have been shown to follow an autosomal dominant inheritance pattern. Patients with this condition may also have cardiac malformations. In our case, however, family members do not present with similar conditions, so whether his condition was hereditary in nature remains to be determined.

The patient was found to have multiple cavernous hemangiomas in the subcutaneous and mucosal tissues and in the cerebrum, nasopharynx, tongue, esophagus, gastric body, sigmoid colon and adrenal gland. Notably, the gastrointestinal lesions appeared to lead to relapse of intestinal bleeding. This boy weighed less than normal for his age, with pale face and lips, indicating severe anemia; laboratory analyses revealed that he had a low hemoglobin level. His anemia improved after he was provided with iron supplementation; however, the pain caused by the hemangioma lesions was aggravated.

Imaging studies play an important role in the diagnosis of BRBNS, which is a rare condition and is difficult to diagnose. Color Doppler is widely used to diagnose hemangioma and has good sensitivity and specificity in detecting superficial hemangiomas. MRI and CT are useful for detecting hemangiomas located in deep organs. The WBBPS technique has been applied to diagnose congenital vascular malformations and has been shown to have better sensitivity in detecting lesions compared with MRI, angiography and Doppler sonography. We therefore recommend the use of combined imaging modalities in the examination of hemangioma lesions.

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Note: Figure translations are in progress. See original paper for figures.