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Case Report

Blue rubber bleb nevus syndrome complicated by a ventricular septal defect: a case report

CHI Xiaohua[1], WANG Quanshi[1], XUE Yaoming[2], LIU Feng[1], QI Yongshuai[1], LI Guiping[1]*

[1]Department of Nuclear Medicine, [2]Department of Endocrinology and Metabolism, Nanfang Hospital, Southern Medical University, Guangzhou 510515, China

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 are localized defects of vascular morphogenesis caused by dysfunction 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 such visceral organs as the liver and adrenal glands. In 1860, the co-occurrence of congenital cutaneous and gastrointestinal hemangiomatosis was named Blue rubber bleb nevus syndrome (BRBNS) by Gascoyen. Over the last two centuries, only approximately 200 cases of BRBNS have been reported. The lesions can appear throughout the whole body, and in some cases, BRBNS may co-occur with malignant or benign tumors[1-4].

The co-occurrence of cavernous hemangiomas and congenital heart disease was first reported by Schneeweiss in 1982[5]. So far only 3 BRBNS patients with congenital heart disease have been reported[6-8]. In this report, we present a case of BRBNS complicated by ventricular septal defect in a 15-year-old boy.

CASE REPORT

The 15-year-old boy presented with numerous masses on the body trunk and extremities at birth and was diagnosed to have BRBNS. Surgery was performed on his back and right wrist at 6 months of age. He received subsequent Chinese medicine and injection therapies at 4 years of age, and was treated intermittently in recent years with propranolol. Some of his lesions disappeared while new ones appeared during his childhood. Surgical repair of his ventricular septal defect (VSD) was performed when he was one year old. At 14 years of age, the boy was 160 cm tall, weighed 45 kg and was not active in physical activities. His stool appeared chocolate-colored on sporadic occasions, fecal occult blood tests yielded positive results and urinalysis was normal. Laboratory analyses revealed severe iron-deficiency anemia, which was confirmed by bone marrow biopsy. Laboratory analyses showed that the patient had normal blood platelets with leukocytes of $(2.65\text{-}5.14) \times 10^{-9}$/L, hemoglobin of 41-91 g/L, mean corpuscular volume of 62.8-81.2 fL, mean corpuscular hemoglobin of 15.7-17.7 pg, Fe of 1.7-20 μmol/L, transferrin saturation of 2.58-3.21 g/L and ferritin of 3.5-7.4 ng/mL.

Computed tomography (CT) of the chest showed inflammation on both sides of the lower lungs. Abdominal ultrasound revealed a hemangioma lesion in the right adrenal gland, and the liver, gallbladder, pancreas and kidneys were normal. Capsule endoscopy showed the presence of multiple hemangiomas in the upper segment of the small bowel. Gastroscopic and colonoscopic examinations revealed multiple vascular blebs in the gastric body and sigmoid colon (Fig.1), and the esophagus also contained hemangioma lesions. Magnetic resonance imaging (MRI) of the brain showed the presence of hemangioma lesions on both sides of the occipital lobe (Fig.1) and in the parotid glands, facial subcutaneous tissue, nose and pharynx. Whole-body blood pool scintigraphy (WBBPS) using $^{99}Tc^{m}$-labeled red blood cells ($^{99}Tc^{m}$-RBCs) by single-photon emission

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***Corresponding author:** LI Guiping, MD, professor, E-mail: hyxknfyy@163.com.

computed tomography (SPECT) displayed multiple radioactive foci in the subcutaneous tissues of the trunk, extremities, and other organs including the bilateral parotids and small intestine (Fig.2).

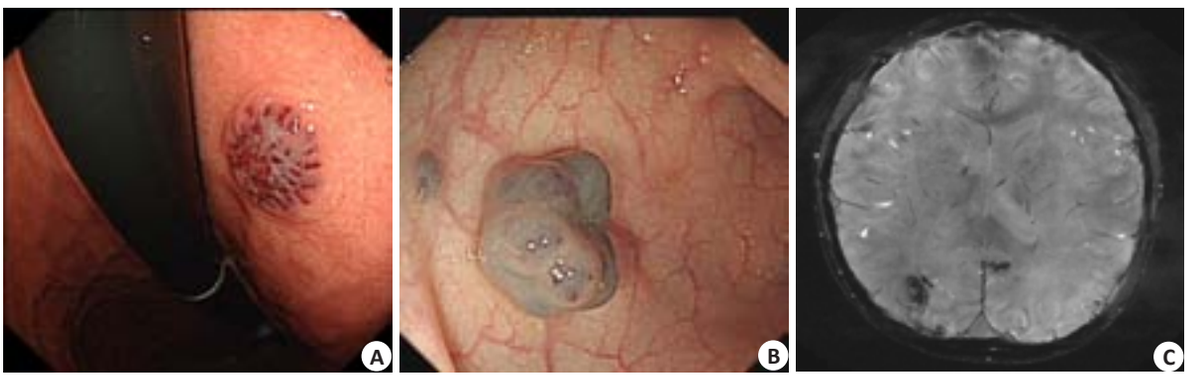

Fig.1 Gastroscopy revealed the presence of a single hemangioma lesion in the gastric body (***A***). Colonoscopy revealed multiple varicosities in the sigmoid colon (***B***). Susceptibility-weighted imaging (SWI) clearly displayed multiple draining veins on both sides of the occipital lobe detected by their abnormally low signals (***C***).

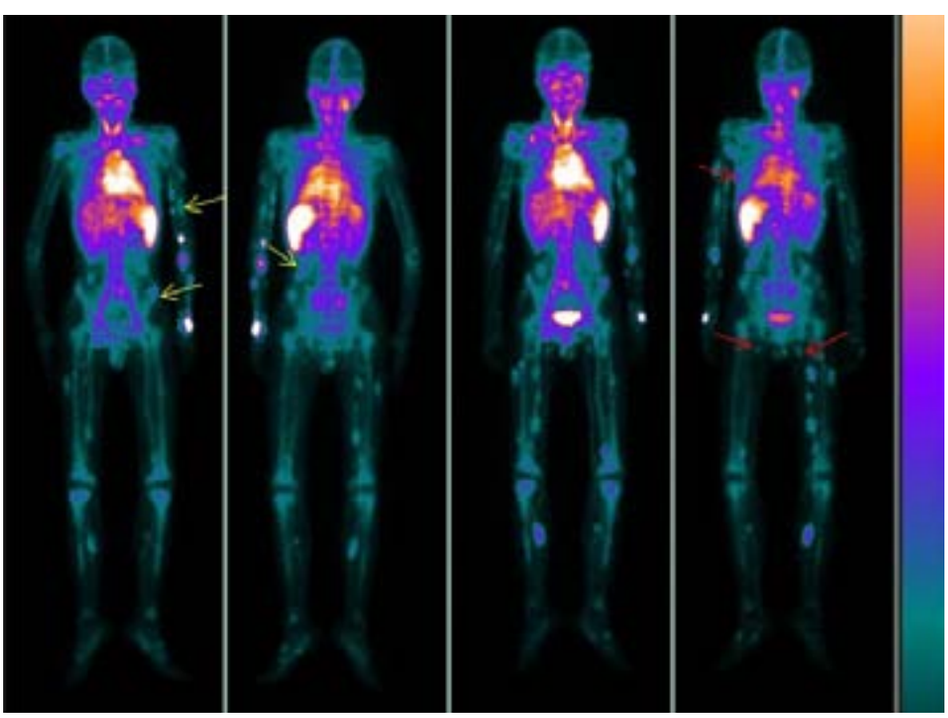

Fig.2 Whole-body blood pool scintigraphy of the patient. At 10 min, the images show extensive distribution of the radioactive tracer throughout the subcutaneous tissues of the extremities, trunk and small intestine (yellow arrows). At 3 h, radionuclide pooling abnormally increased as compared with that at 10 min (red arrows).

DISCUSSION

Wouters et al demonstrated that hereditary cutaneomucosal venous malformation had an autosomal dominant inheritance pattern [9]. Patients with this disease may also have cardiac defects (cardiac malformations). In our case, however, the patient's family members do not present with similar conditions, so that 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 Fe 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 a good sensitivity and specificity in detecting superficial hemangioma. MRI and CT are useful for detecting hemangiomas located in organs. $^{99}Tc^{m}$-RBCs technique has been applied with WBBPS to diagnose congenital vascular malformations, and has been shown to have a better sensitivity in detecting the lesions compared with MRI, angiography, and Doppler sonography [10-11]. We therefore recommend the use of combined imaging modalities in the examination of hemangioma lesions.

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蓝色橡皮疱痣综合征合并室间隔缺损 :1 例报告

池晓华[1],王全师[1],薛耀明[2],刘 峰[1],齐永帅[1],李贵平[1]
南方医科大学南方医院[1]核医学科,[2]内分泌与代谢科,广州 广东 510515

摘要:蓝色橡皮疱痣综合征合并室间隔缺损在临床上非常罕见。我们报道1例15岁患者,该患者以多发海绵状血管瘤为主要表现,同时合并室间隔缺损。多种影像学检查表明,大量的海绵状血管瘤病灶位于皮下组织和口腔粘膜组织,同时在患者大脑、鼻咽部、舌头、食道、胃、乙状结肠和肾上腺均发现血管瘤病灶。多种影像诊断技术的联合应用对病灶的发现具有重要意义。

关键词:蓝色橡皮疱痣综合征;海绵状血管瘤;室间隔缺损

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作者简介:池晓华,硕士,主治医师,E-mail: 546863745@qq.com

通信作者:李贵平,博士,主任医师,教授,硕士研究生导师,E-mail: hyxknfyy@163.com