

# World Journal of *Gastrointestinal Surgery*

*World J Gastrointest Surg* 2024 November 27; 16(11): 3381-3642



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The primary aim of *World Journal of Gastrointestinal Surgery* (WJGS, *World J Gastrointest Surg*) is to provide scholars and readers from various fields of gastrointestinal surgery with a platform to publish high-quality basic and clinical research articles and communicate their research findings online.

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**INDEXING/ABSTRACTING**

The WJGS is now abstracted and indexed in Science Citation Index Expanded (SCIE, also known as SciSearch®), Current Contents/Clinical Medicine, Journal Citation Reports/Science Edition, PubMed, PubMed Central, Reference Citation Analysis, China Science and Technology Journal Database, and Superstar Journals Database. The 2024 Edition of Journal Citation Reports® cites the 2023 journal impact factor (JIF) for WJGS as 1.8; JIF without journal self cites: 1.7; 5-year JIF: 1.9; JIF Rank: 126/292 in surgery; JIF Quartile: Q2; and 5-year JIF Quartile: Q3.

**RESPONSIBLE EDITORS FOR THIS ISSUE**

Production Editor: Zi-Hang Xu, Production Department Director: Xiang Li, Cover Editor: Jia-Ru Fan.

**NAME OF JOURNAL**

*World Journal of Gastrointestinal Surgery*

**ISSN**

ISSN 1948-9366 (online)

**LAUNCH DATE**

November 30, 2009

**FREQUENCY**

Monthly

**EDITORS-IN-CHIEF**

Peter Schemmer

**EDITORIAL BOARD MEMBERS**

<https://www.wjgnet.com/1948-9366/editorialboard.htm>

**PUBLICATION DATE**

November 27, 2024

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<https://www.wjgnet.com/bpg/gerinfo/204>

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**PUBLICATION ETHICS**

<https://www.wjgnet.com/bpg/gerinfo/288>

**PUBLICATION MISCONDUCT**

<https://www.wjgnet.com/bpg/gerinfo/208>

**ARTICLE PROCESSING CHARGE**

<https://www.wjgnet.com/bpg/gerinfo/242>

**STEPS FOR SUBMITTING MANUSCRIPTS**

<https://www.wjgnet.com/bpg/gerinfo/239>

**ONLINE SUBMISSION**

<https://www.f6publishing.com>



## Blue rubber blister nevus syndrome: A case report

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**Specialty type:** Gastroenterology and hepatology

**Provenance and peer review:** Unsolicited article; Externally peer reviewed.

**Peer-review model:** Single blind

**Peer-review report's classification**

**Scientific Quality:** Grade B

**Novelty:** Grade B

**Creativity or Innovation:** Grade B

**Scientific Significance:** Grade C

**P-Reviewer:** Fernandez-García R

**Received:** April 23, 2024

**Revised:** September 5, 2024

**Accepted:** September 25, 2024

**Published online:** November 27, 2024

**Processing time:** 189 Days and 23.6 Hours



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### Abstract

#### BACKGROUND

Blue rubber blister nevus syndrome (BRBNS) is a congenital, rare disease characterized by venous malformations of the skin and internal organs, affecting all systems throughout the body. The pathogenesis is unknown. There is no consensus on the treatment of BRBNS. Most of the previously reported cases were mild to moderate with a good prognosis, and this case was a critically ill patient with severe gastrointestinal hemorrhage, disseminated intravascular coagulation (DIC), and severe joint fusion that was different from previously reported cases.

#### CASE SUMMARY

An 18-year-old man with early onset of BRBNS in early childhood is reported. He presented with recurrent melena and underwent malformed phlebectomy and partial jejunectomy and ileal resection. The patient had melena before and after surgery. After active treatment, the patient's gastrointestinal bleeding improved. This was a case of atypical BRBNS with severe gastrointestinal bleeding and severe joint fusion, which should be differentiated from other serious joint lesions and provide clinicians with better understanding of this rare disease.

#### CONCLUSION

This case of critical BRBNS with gastrointestinal hemorrhage, DIC and severe joint fusion provides further understanding of this rare disease.

**Key Words:** Blue rubber blister nevus syndrome; Gastrointestinal hemorrhage; Joint fusion; Maffucci's syndrome; Case report

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**Core Tip:** Blue rubber blister nevus syndrome is a rare disease. The mechanism of the disease is not fully understood. Lack of awareness among clinicians leads to delays in diagnosis and treatment. This severe case had a long course of illness and difficult diagnosis and treatment. The patient was treated with surgery because the gastrointestinal bleeding did not resolve after medication, repeated intervention and other treatments. The patient had severe joint fusion and was unable to walk, which seriously affected his quality of life. Clinicians should pay attention to the differential diagnosis of rare diseases while learning about them, so as to avoid misdiagnosis and missed diagnosis.

**Citation:** Wang WJ, Chen PL, Shao HZ. Blue rubber blister nevus syndrome: A case report. *World J Gastrointest Surg* 2024; 16(11): 3584-3589

**URL:** <https://www.wjgnet.com/1948-9366/full/v16/i11/3584.htm>

**DOI:** <https://dx.doi.org/10.4240/wjgs.v16.i11.3584>

## INTRODUCTION

Blue rubber blister nevus syndrome (BRBNS) is a disease characterized by multiple widespread venous malformations. The most common symptom is venous malformations of the skin and internal organs. Cutaneous angiomas are bluish or purple, soft, rubbery, nipple-like nodules with wrinkled and hyperhidrotic surfaces[1]. Cutaneous angiomas are usually diagnosed in infancy or childhood, with only 4% of reported cases diagnosed in adulthood. After the skin, the gastrointestinal tract is the second most commonly affected organ, with most lesions occurring in the small intestine, followed by the colon. A review[2] of 101 cases reported that distribution of BRBNS-related lesions was variable, but predominantly oral (55.9%), followed by small bowel (49.5%), colorectal (35.6%) and stomach (26.7%). Another review[3] reported that the most commonly affected area of the gastrointestinal tract was the small intestine, with the highest prevalence of ileal involvement at 73.5%. Gastrointestinal nevi can cause overt or occult bleeding[4]. Skin moles vary in size and number and are usually asymptomatic but can cause pain or hyperhidrosis. Clinical diagnosis is mainly based on characteristic skin lesions. Endoscopy is an important diagnostic method for gastrointestinal hemangioma. In addition, ultrasound, angiography, computed tomography (CT), magnetic resonance imaging and other auxiliary examinations can find other affected organs. BRBNS is thought to be related to mutation of the short arm gene on chromosome 9[5], which is inherited chromosomal dominantly. However, the pathogenesis is not clear. There is no consensus on the treatment of BRBNS. Conservative treatment, endoscopic treatment and surgical resection are usually based on the degree of gastrointestinal involvement and other organ involvement. Most of the patients reported so far are mild to moderate patients with good prognosis. This case was a critically ill patient with life-threatening gastrointestinal bleeding, systemic vascular involvement and severe joint fusion.

## CASE PRESENTATION

### Chief complaints

An 18-year-old man was admitted to the hemangioma department with the main complaint of multiple blue-purple cutaneous nevus for 18 years, intermittent melena for 4 years and repeat black stools for 2 weeks.

### History of present illness

The patient was born with multiple blue angiomas on the buttocks and back, which gradually enlarged with age, covering the chest, back and limbs. These angiomas were bluish or purple, soft, rubbery, nipple-like nodules and were not treated. Eight years ago, the patient often felt pain when walking. However, no treatment was given because there were no positive signs such as fractures. Six years ago, he could not straighten his legs or even get out of bed and went to several hospitals without a clear diagnosis. Four years ago, the patient started developing blood in his stools, and underwent genetic testing in Shanghai East Hospital, which revealed the *TEK* gene mutation. BRBNS was confirmed. He was given hemostatic drugs, blood transfusion, oral sirolimus treatment, and injected with absolute ethanol > 10 times for cutaneous angiomas. Two weeks ago, he was admitted to the above hospital due to recurrent melena. Capsule endoscopy showed multiple jejunal venous malformations and active bleeding. The patient was given hemostatic drug application, blood transfusion and oral sirolimus again. However, the gastrointestinal bleeding did not stop and he was transferred to our hospital.

### History of past illness

The patient had impaired joint mobility for 6 years and took sirolimus irregularly for 4 years. He was injected with absolute ethanol > 10 times for cutaneous angiomas 4 years ago and underwent capsule endoscopy 2 week ago.

### Personal and family history

The patient denied any family history of cutaneous angiomas or gastrointestinal bleeding.

### Physical examination

On physical examination, the vital signs were as follows: (1) On admission: Body temperature, 36.3 °C; blood pressure, 110/70 mmHg; heart rate, 80 beats/minute; respiratory rate, 19 breaths/minute; and (2) On admission to the intensive care unit (ICU): Body temperature, 36.8 °C; blood pressure, 137/81 mmHg (plasma and cryoprecipitate were transfused daily before the ICU); heart rate, 75 beats/minute; respiratory rate, 15 breaths/minute. The patient had multiple blue-purple, cutaneous nevi all over the body (Figure 1). The left upper limb was hypertrophic, deformed with palpable phleboliths under the skin (Figure 1A). The left elbow and bilateral knee joints had limited extension (Figure 1A and B). The maximum extension of the left elbow and both knee joints was 60° and 160°, respectively. The patient was not able to walk.

### Laboratory examinations

Table 1 shows coagulation parameters at the time of admission, during hospitalization, and at discharge.

### Imaging examinations

Capsule endoscopy at the other hospital revealed multiple venous malformations and active small intestinal bleeding (Supplementary Figure 1).

### Genetic testing

The patient's previous peripheral blood gene test showed a missense mutation in the *TEK* gene on chromosome 9, with a mutation site NM\_000459: Exon17: C.C2740T: p.Leu914Phe, and a variation of 0.62%.

## FINAL DIAGNOSIS

(1) BRBNS; (2) Gastrointestinal bleeding; (3) Severe anemia; and (4) Disseminated intravascular coagulation (DIC).

## TREATMENT

Capsule endoscopy revealed multiple venous malformations and active bleeding in the small intestine (Supplementary Figure 1). The patient was recommended for gastrointestinal surgery due to active bleeding, low fibrinogen level, high risk of bleeding during bowel preparation, and potential gastrointestinal perforation due to endoscopic venous hemostasis. After this evaluation, phlebectomy of intestinal vein malformation, and partial intestine resection were performed. The lesion in the intestinal tract was dark red, and the different sized blue-purple nodules with a maximum diameter of 3.2 cm were scattered throughout the serous and mucosal layers of the jejunum and ileum. The length of the diseased intestine was about 2.1 m, and the malformed blood vessels involved the mesentery (Figure 2A and B). Pathological findings show multiple malformed veins under the mucosal layers of the jejunum and ileum (Figure 2C). On postoperative day 5, the patient was transferred to the ICU because there was > 1 L of dark blood in the stools, and 1 L of dark blood was released again that night.

From day 2 to 4 in the ICU, 2-5 L/day of bloody stools were released. In addition to component blood transfusion, the patient was given thrombin 2 U/day before surgery, which was boosted to 6 U/day after admission to the ICU, along with vitamin K 10 mg twice daily, tranexamic acid 0.5 g once daily, and fibrinogen concentrate 4-8 g daily. During the patient's 17-day hospital stay, the patient was given almost daily transfusions of red blood cells and cryoprecipitate. Hemoglobin, fibrinogen and fibrinogen degradation products fluctuated at 38-70 g/L, 0.30-1.08 g/L, and 129.77-506.16 mg/mL, respectively (Table 1).

## OUTCOME AND FOLLOW-UP

The drainage fluid of the abdominal drainage tube changed from bloody to pale yellow on day 3 in the ICU (postoperative day 8). Gastrointestinal bleeding was significantly reduced to 530 mL on day 5 (postoperative day 10). On day 6 in the ICU, the patient was transferred back to the local hospital due to personal circumstances. At follow-up, the patient was transfused with 1-2 U red blood cells and 2-4 U cryoprecipitate U per day. The melena was reduced to 100 mL/day on day 2 in the local hospital. The abdominal drainage tube was removed and the liquid diet was started on day 4.

## DISCUSSION

This patient developed in infancy systemic cutaneous nevi as the first manifestation, and then movement disorders, joint fusion and severe gastrointestinal bleeding. The patient's condition did not alleviate significantly after oral treatment with several drugs and anhydrous ethanol injections. This was a critical case of BRBNS with life-threatening gastrointestinal bleeding and joint fusion different from previously reported cases.



**Table 1 Coagulation function at admission and discharge**

	Hb (110-160 g/L)	Plt (100-300 10 <sup>9</sup> /L)	FBG (1.8-3.5 g/L)	FDP (0-5 g/mL)	DD (0-0.55 g/mL)	APTT (22-35 seconds)	PT (9-14 seconds)	TT (14-21 seconds)
At admission	58	114	0.52	222.03	69.12	30.6	16.2	26.4
H/L	38	48	1.08	506.16	144.50	23.3	13.0	18.0
At discharge	41	52	1.08	188.40	66.54	31.4	13.0	18.0

Hb: Hemoglobin; Plt: Platelets; FBG: Fibrinogen; FDP: Fibrinogen degradation product; DD: D dimer; APTT: Activated partial thromboplastin time; PT: Prothrombin time; TT: Thrombin time; H/L: The highest/lowest during hospitalization.

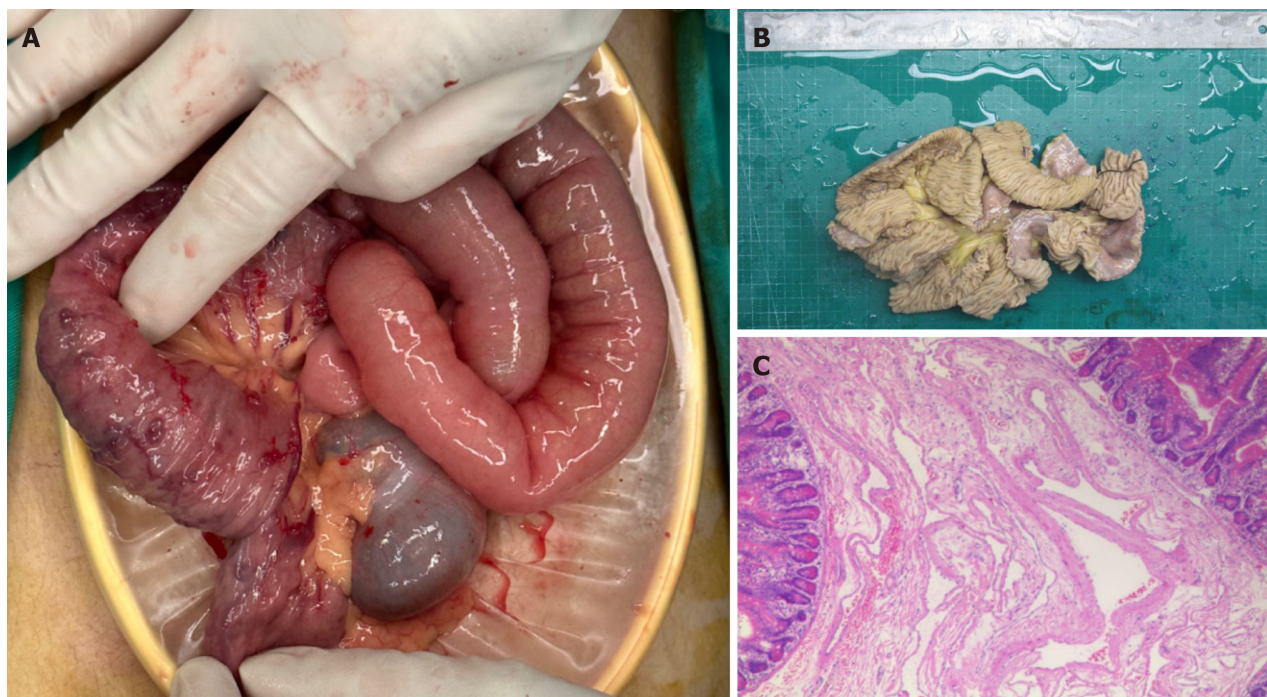


**Figure 1 Cutaneous angiomas and joint fusion.** A: Multiple cutaneous angiomas, thickened and hypertrophic deformity of the left upper extremity, and left elbow joint fusion; B: Multiple cutaneous angiomas of left lower extremity and bilateral knee joint fusion; C: Cutaneous angiomas of the feet; D: Enlarged view of cutaneous angiomas of the left lower extremity.

BRBNS is characterized by multiple blue-purple nodules or masses on the skin and gastrointestinal tract, and can affect different organs. The lungs, bronchi, urinary tract, brain, liver, spleen or heart can be involved. Gastrointestinal bleeding [6] leads to hematemesis, melena, iron deficiency anemia, and even life-threatening hemorrhagic shock. Small bowel lesions [7] can lead to intussusception and intestinal obstruction, brain lesions [8] can lead to cerebral infarction, vertebral lesions can lead to spinal cord compression, and bronchial lesions [9] can lead to chronic cough. This patient mainly presented with gastrointestinal symptoms such as bleeding and movement disorder due to joint fusion. Clinical differential diagnosis with familial hemorrhagic telangiectasia (Osler Weber-Rendu disease), angiokeratoma corporis diffusum (Fabry's disease), dyschondroplasia with hemangiomas (Maffucci's syndrome), multiple glomangiomas and Kaposi's sarcoma may be difficult [1]. In particular, this case should be distinguished from Maffucci's syndrome, which is a clinical syndrome with a tendency to malignant transformation due to somatic mutations leading to mesodermal dysplasia, mainly characterized by multiple chondroma and hemangioma [10]. Maffucci's syndrome has mostly bilateral involvement. Lesions often occur in short tubular bones, such as in the hands and feet, and can also occur in long bones such as the humerus, tibia, fibula, ulnar and radius, as well as scapula, pelvis and ribs [11,12]. Clinicians should pay attention to the differential diagnosis of rare diseases while learning about them, so as to avoid misdiagnosis and missed diagnosis. Unfortunately, the patient refused further imaging due to gastrointestinal hemorrhage.

The patient had the typical skin and gastrointestinal lesions of BRBNS, a positive gene report and no family history. During the long medical treatment, the patient was given sirolimus tablets orally, and the systemic nodular lesions and gastrointestinal bleeding were not in remission. Gastrointestinal bleeding and severe anemia persisted after oral hemostatic drugs, repeated transfusions of blood products and cutaneous vascular embolization. The patient had severe hypofibrinogenemia at admission. Despite the administration of a large number of blood components (red blood cells, plasma, and cryoprecipitate) before and during surgery, the patient still had severe gastrointestinal bleeding after





**Figure 2 Intestinal lesions.** A: Diseased small intestine (left) and normal small intestine (right); B: Diseased small intestine after resection; C: Multiple venous malformations under the small intestinal mucosa.

surgery, which may be related to secondary DIC induced by combination of primary disease and surgery. Anastomotic hemorrhage cannot be completely ruled out, because the patient refused CT scanning. However, combined with the complete resection of the vascular lesions, and clear abdominal drainage, we considered anastomotic bleeding to be unlikely. After positive treatment with hemostatic drugs, antifibrinolytic drugs, massive red blood cell transfusions to alleviate anemia, cryoprecipitate (24 U/day) and plasma to replenish coagulation factors, and target oriented transfusion of fibrinogen concentrate (4-8 g/day), the bleeding was significantly reduced. Thromboelastography showed that there was no abnormality in the activity of coagulation factors at admission and discharge. Coagulation dysfunction was mainly caused by severe fibrinogen deficiency. Careful perioperative management and close monitoring are necessary when the coagulopathy caused by the primary disease increases the risk of perioperative bleeding, but nonsurgical treatment cannot stop the bleeding.

It is important to note that this case differs from previous ones. Firstly, the patient had severe joint fusion of the left elbow and both knees, which was not seen previously. Secondly, extraintestinal vascular lesions differed from previously reported BRBNS with a single, clumpy dilated irregular vascular lumen. In this case, the lesion involved the whole thickness of the intestinal wall from the mucosal to the serosal layer, and the venous malformation was not a solitary, isolated hemangioma. Thirdly, the patient's previous oral administration of sirolimus and iron did not alleviate bleeding and anemia. All the above indicate that this may be an atypical case of BRBNS, or that there were comorbidities that we have not yet identified. Although the gastrointestinal bleeding of the patient was temporarily stopped, joint fusion persisted, and long-term prognosis and quality of life were poor.

## CONCLUSION

We report a case of critical BRBNS with gastrointestinal hemorrhage, DIC and severe joint fusion, which provides clinicians with further understanding of this rare disease.

## ACKNOWLEDGEMENTS

We would like to thank Shanghai Dongfang Hospital for providing the gastroscopy report of the patient.

## FOOTNOTES

**Author contributions:** Wang WJ collected, wrote and followed up the cases; Chen PL provided language corrections, and Shao HZ supervised the patient's treatment and was responsible for the final review of the manuscript.

**Informed consent statement:** Informed written consent was obtained from the patient for publication of this report and any accompanying images.

**Conflict-of-interest statement:** The authors declare that they have no conflict of interest to disclose.

**CARE Checklist (2016) statement:** The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

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**S-Editor:** Li L

**L-Editor:** A

**P-Editor:** Xu ZH

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