Blue Rubber Bleb Nevus Syndrome

BLUE RUBBER BLEB NEVUS SENDROMU

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Summary

Aim: Blue rubber bleb nevus syndrome (BRBNS) is a rare condition characterized by gastrointestinal and skin hemangiomas. These hemangiomas may cause gastrointestinal bleeding and anemia.

Case: A 19-year-old male patient was admitted with anemia to our clinic. Physical examination revealed pale conjunctiva, surgical incision scar on right auricula, and hemangiomas over dorsum of right hand, right big toe and gluteal region. In laboratory findings, there was iron deficiency anemia. Colonoscopic examination revealed multiple hemangiomas approved by histologically in all sites of colon and rectum. BRBNS must be considered as a rare cause of iron deficiency anemia.

Key Words: Blue rubber bleb nevus syndrome, Hemangioma, Anemia

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Özet

Amaç: Blue rubber bleb nevus sendromu (BRBNS) gastrointestinal sistem ve deride hemanjiomlarla karakterize nadir bir durumdur. Bu hemanjiomlar gastrointestinal kanama ve anemiye neden olabilirler.

Olgu: On dokuz yaşında bir erkek hasta kliniğimize anemiyle başvurdu. Fizik muayenede konjonktival solukluk, aurikulada insizyon skarı ve sağ el sırtı, sağ ayak baş parmağı ve gluteal bölgede hemanjiomlar mevcuttu. Laboratuvarda demir eksikliği anemisi saptandı. Kolonoskopik bakıda kolon ve rektumun tüm alanlarında histolojik olarak ıspatlanan hemanjiomlar mevcuttu. BRBNS'u demir eksikliği anemisinde nadir sebep olarak göz önünde bulundurulmalıdır.

Anahtar Kelimeler: Blue rubber bleb nevus sendrom, Hemanjiom, Anemi

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Blue rubber bleb nevus syndrome is an uncommon condition manifested by gastrointestinal and skin hemangiomas that lead to gastrointestinal bleeding and anemia. This association was first described in the nineteenth century, but named in 1958 (1.2). More than 200 cases have been reported until today (3). The cutaneous lesions usually appear at birth or in early childhood and typically increase in size with age. The fragility of the gastrointestinal lesions leads to blood loss and anemia. If possible, therapy for this syndrome should be conservative. This report demonstrates a case of Blue Rubber Bleb Nevus Syndrome (BRBNS) with occult gastrointestinal hemorrhage and iron deficiency anemia. Clinic and pathology of this rare condition are discussed.

Case

In this case, a Blue Rubber Bleb Nevus Syndrome (BRBNS) in a 19 years old male patient with multiple cutaneous and gastrointestinal hemangiomas causing anemia is described. He was born with hemangioma located on his right auricula (Figure 1). In the first three years' period, hemangioma enlarged, and he had an operation on his ear. Apart from pallor on his face, he did not have any significant complaints for 13 years. In the last 3 years, he has suffered from constipation. In the last 1 year, fatigue, tiredness have developed in the patient. The patient has not had a history of gastrointestinal hemorrhage. On admission, physical examination revealed pale conjunctiva, surgical incision scar on auricula, and hemangiomas over

Figure 1. Hemangioma located on his right auricula

dorsal part of right hand, right big toe and gluteal region. The rest of the systemic examination was normal. Results of laboratory tests were as follows: The stools were guaiac-positive, hemoglobin: 8.7 (N:13-18) g/dL, hematocrit: 27.2% (N:42-52%), red blood cell count: 4.8 (N:4.15-4.9)x10^6/mm^3, mean corpuscular volume (MCV):56 (N:86-98) fl, mean corpuscular hemoglobin (MCH):30 (N:28-33) pg/cell, mean corpuscular hemoglobin concentration (MCHC):33 (N:32-36) g/dL, white blood cell count: 8.7 (N:4.3-10.8)x10^3/mm^3, platelet count: 301 (N:130-140)x10^3/mm^3, peripheral blood smear: hypochromia, microcytosis, aniso-poikilocytosis, serum iron: 10 (N:50-150) μg/dL, total iron-binding capacity (TIBC):280 (N:250-370) μg/dL, serum ferritin: 2.4 (N:150-400) ng/mL, transferring saturation: 2.8 (N:20-45)%, reticulocyte count: <1% (N:<2%), Urea nitrogen:13 (N:10-20) mg/dL, Creatinin: 0.8 (N:<1.5mg/dL), glucose: 86 (N:75-115) mg/dL. Bone marrow examination: Non-diagnostic; bone morrow iron staining: Negative; abdominal ultrasonography: Normal; thoracic, abdominal and cranial computerized tomography: Normal; cranial magnetic resonance imaging: Normal; upper GI tract endoscopy: Normal; colonoscopy, revealing hemangiomas in all part of colon and rectum (Figure 2). The lesions varied in size from 3 to 25 mm. Hemangioma in gluteal region was resected, and histological examination revealed cavernous hemangioma.

Discussion

Blue rubber bleb nevus syndrome, an uncommon condition, is manifested by gastrointestinal and skin hemangiomas and gastrointestinal hemorrhage.

This association was first described by Gascoyen in 1860, and later named as blue rubber bleb nevus syndrome (1,2). A family history is infrequent, although a few cases of autosomal-dominant transmission have been reported (4). A gene for venous malformations was mapped to chromosome 9p from an extended family, in which several members had BRBNS-like cutaneous, mucocutaneous, and visceral lesions (5). Histologically, these lesions are composed of large, thin-walled vascular spaces with vessels lined by a hyperplastic endothelium and separated by a scant network of elastin-deficient connective tissue similar to cavernous hemangiomas seen in other clinical situations (6). Lesions may be single or multiple; they are blue and raised, vary from 0.1-10 cm in diameter, and have a wrinkled surface. Cutaneous lesions may be present throughout the body, but

Figure 2. Hemangiomas in the colon

they are usually found on the trunk extremities and face. In addition to cutaneous lesions, hemangiomas can be present in any portion of gastrointestinal tract, but the small bowel is the most frequently involved site. In the colon, they are more commonly distally. Furthermore, the lesions may be observed in the nasopharynx, oropharynx, esophagus, stomach, peritoneal cavity, mesentery, liver, lung, glans penis, eye, and central nervous system (7). In contrast to cutanous lesions, gastrointestinal hemangiomas are fragile and tend to bleed easily. Although the bleeding may be acute in the form of hematemesis, melena or hematochezia, it is usullay occult and chronic, and leads to iron deficiency anemia. Thrombocytopenia, chronic consumptive coagulopathy, intussusception, bowel infarction, rectal prolapsus, epileptic disorder, ocular lesions, and orthopedic abnormalities have been reported in association with this syndrome (8-13). The diagnosis is usually made on findings of clinical, histological, and imaging studies including upper endoscopy, colonoscopy, barium studies, computed tomography and magnetic resonance imaging. Magnetic resonance imaging (MRI) has recently been shown to be an excellent method for identifying the presence and extent of lesions in BRBNS. Hemangiomas are revaled as a bright signal on T2-weighted MR images. This is probably the result of slow flow or thrombosis, typically present in these lesions, and allows for easy recognition (14). However, we could not reveal hemangiomas in MRI. Enteroclysis, a special barium study of small bowel, is another significant diagnostic tool. This reveals the hemangioma as an oval fillind defect in the lumen of bowel (15). Differential diagnosis includes glomangiomatosis, Maffucci's syndrome, Osler-Weber-Rendu syndrome, Kaposi's sarcoma. The patients should be treated with only conservative treatment including oral iron supplement or blood transfusions whenever bleeding episodes are mild. Resection of the involved segment of bowel is recommended for recurrent hemorrhage. Endoscopic laser coagulation may be dangerous because these lesions may involve the full thickness of the bowel wall (4). However, some have suggested that endoscopic control of the nevi using sclerosants is a reasonable first-line therapy, and if operations are performed, assisted enteroscopic sclerotherapy or hemangioectomy and repair rather than resection should be favored in most instances to reduce the risk of malnutrition (16). Although oral prednison, aspirin, interferon-alpha, vincristine, octreotid have been used for acute hemorrhage, the effectiveness of these drugs have not been proven.

This patient admitted with anemia did not have any history of hemorrhage. Our patient was treated with per-oral iron supplementation, and no blood transfusion was required.

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