中文題目：患有貧血的年輕男性：藍色橡皮泡痣症候群
英文題目：A young male with anemia: blue rubber bleb nevus syndrome
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Background：Blue rubber bleb nevus syndrome (BRBNS), a rare disease is characterized by multiple venous malformation in the skin, gastrointestinal tract and other visceral organs. Patients with BRBNS mainly presents with iron deficiency anemia from chronic GI bleeding or extensive hemorrhage, and require lifelong treatment with iron and blood transfusions. BRBNS is an important condition due to the potential for significant bleeding which can be fatal. We report here of a 19-year-old patient with multiple hemangiomas over his duodenum, jejunum and colon.

Case Presentation：The 19 year-old male was a student and had past medical history of right axillary hemagioma status post excision surgery on 2001/11. School health examination revealed Hgb: 9.3 g/dl in 2013, he was then taken to our pediatric clinic and blood examination revealed Hgb: 8.6 g/dl, ferritin: 2.4 ng/mL, Fe:19 ug/dL and iron deficiency anemia was suspected. Stool occult blood was also done showed negative finding. Iron suppelement was prescribed and nutritional education was performed. He had improved hemogobin from 8.6 to 13.1(g/dl) while OPD follow up. However, he presented occasionally shortness of breath and getting tired easily while playing basketball three years later. School healthy examination revealed Hgb: 5.4 g/dl. He denied bloody stool nor tarry stool. Due to decreased Hb level, he was then taken to our pediatric clinic for help again in 2016/05. Thus, upper and lower GI scopy examination were arranged.
He had esophagogastroduodenoscopy(EGD) showed one 0.3 cm purple submucosal lesion and colonscopy showed seven sessile purple polyps and one polypoid lesion, about 3 cm in size on 2016/06, colon hemangioma were suspected. Lower gastrointestinal endoscopic ultrasound and hemangioma ligation was done on 2016/07. He still had progressive decreased hemoglobin after hemangioma ligation. He had repeated EGD and colonoscopy showed one 4mm hemangioma over 2nd portion of duodenum and three hemangiomas up to 5mm over colon s/p ligation on 2017/02. He also received angiography after ligation and there was no active contrast extravasation or hypervascular lesion noted. Then, he received small bowel capsule scopy, and 8 variable size bluish polypoid lesions from duodenal bulb to ileum, compatible with hemangioma. He had the third time EGD and colonoscopy due to decreased hemoglobin still noted on 2017/07. There was no bleeder noted via EGD but one 8mm hemangioma noted over transverse colon s/p band ligation. He also had double balloon enteroscopy showed jejunal subpeithelial lesions with erythematous patches,

r/o blue rubber bleb nevus syndrome s/p endoloop ligation, EMR and hemoclip closure. After treatment with double balloon enteroscopy, he had stable hemoglobin without bleeding even if without iron supplement.

Discussion：Blue rubber nevus syndrome is a rare disorder which consists mainly of abnormal venous malformation over the skin, gastrointestinal tract and other visceral organs. Dr. Gascoyen recognized this condition initially in 1860. It was described by William Bennett Bean in 1958 and the disease has been termed "bean syndrome," and it was called blue rubber bleb nevus syndrome later. BRBNS was often noted from birth or early childhood (>75%). Cutaneous lesions were often asymptomatic, and mainly in trunk and upper extremities. GI lesions could be found from mouth to anus. Bleeding and secondary iron deficiency anemia were often noted due to GI lesions, including rupture, intestinal torsions, intussuception, and volvulus. The differential diagnosis of recurrent vascular malformation of gastrointestinal tract including Maffucci syndrome, Osler-Weber-Rendu syndrome, and Klipple trenaunay syndrome.
In this case, recurrent dropped hemoglobin was noted due to GI bleeding even if after band ligation of colon hemangioma. We need survey whole GI tract for possible bleeding due to GI lesions could be found from mouth to anus. Thus, capsule scopy was arranged for survey. Double balloon enteroscopy was arranged for further treatment, including EMR with hemoclip closure and endoloop ligation. For GI tract lesion, endoscopic interventions were including Argon plasma coagulation, band ligation, electrocauterization, and Histoacryl injection. Surgery including wedge resection and segmental resection could be considered.

Conclusion：
BRBNS should be a differential diagnosis in patients with unexplained anemia or gastroenterology bleeding. Further endoscopy including EGD, colonscopy and double balloon enteroscopy was helpful for further diagnosis and treatment.