CASE REPORT

Chronic Rectal Bleeding in Proteus Syndrome

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SUMMARY

Proteus Syndrome is a rare congenital hamartomatous disorder that typically manifests itself in overgrowth, vascular malformation and disregulation of fatty tissue. The tissues affected are commonly the limbs but can be of any tissue. Vascular anomalies are common and appear at random sites on the body. Diagnosis is often difficult leading to wrong treatment. We describe a case of a 17- year-old girl with Proteus syndrome presented with symptomatic anaemia secondary to chronic rectal bleeding. Computed Tomography Angiogram of Abdomen and Pelvis confirmed the presence of rectal vascular malformations.

KEY WORDS:
Proteus Syndrome, Rectal vascular malformations, Rectal Bleeding, Anaemia

INTRODUCTION

Proteus syndrome (PS) was originally described by Cohen in 1979[1], and in 1983 named by Wiedemann and colleagues[2] after Proteus, the giant Greek god of the sea, who could change the shape of his body to avoid capture. The name Proteus therefore refers to polymorphism. Its aetiology to date is unknown[2,3], although somatic mosaicism offers the best explanation[3]. The manifestations of PS are highly variable[2,3]. While connective tissue and limbs are commonly affected, any tissue of the body can be affected[2,3]. The degree of severity is also variable[2,3]. Patients with Proteus Syndrome may appear healthy at birth[3]. Overgrowth is usually asymmetric[2,3]. Soft tissue abnormalities include vascular malformations are common in patients with PS[3,4], Diagnosis of PS is difficult as it overlaps with many other overgrowth syndromes[3,4].However, partial gigantism of hands or feet is diagnostic for the syndrome[2,3]. Management of patients with PS can be challenging especially since multiple procedures over a long period of time are required. Therefore, it is necessary for a multidisciplinary approach for the follow-up and management of these patients. We report a case of chronic rectal bleeding from vascular malformations in a young lady with a diagnosis of PS since childhood.

CASE REPORT

A 17-year-old girl presented with symptomatic anaemia and chronic haematochezia. She described as having intermittent minor episodes of painless fresh rectal bleed since childhood with no clots and with blood before bowel output. Stool was soft. On admission, she had iron deficiency anaemia with haemoglobin of 7.4g/dl and was transfused two units of blood. She was already diagnosed with PS since childhood, with bilateral partial gigantism of the feet (Figure I), which have been growing steadily in size. She is the third of four siblings, and all her other siblings are normal. No documented family history of similar disease. She was previously treated as rectal haemangiomas and started on steroid in which the lesion did not respond to. She was admitted for similar symptoms in 2006 and 2009 and given blood transfusion in 2009. But, she defaults subsequent follow up until the recent admission. Colonoscopy performed showed irregular, raised submucosal bluish lesions with intact mucosal involving the rectum, extending from the dentate line up to 10cm. Other areas of the large bowel were normal. Biopsies were not performed. Computerised tomography angiography scan of abdomen and pelvis showed a well defined, homogenous perirectal and anorectal soft tissue mass with multiple small calcifications, consistent with vascular malformations. The patient responded well to blood transfusion and was discharged to enable her to better prepare for her upcoming Sijil Pelajaran Malaysia examination.

DISCUSSION

Proteus syndrome is a rare hamartomatous condition with a wide spectrum of manifestations including an association with vascular anomalies. Hoeger et al reported that vascular anomalies were identified in all the 22 PS patients presented to his outpatient clinic and found that in 10 of them, more than one type of vascular anomalies were present[5]. He further reviewed 100 PS patients reported between 1983 and 2001 and found that 70% of the patients have vascular anomalies[5]. The distribution of vascular anomalies appears at random sites on the body and the varying types further support the concept of somatic mosaicism. While anal bleeding has been reported in association with PS, none of the literature we have come across describes bleeding caused solely by the presence of rectal vascular malformations. Vascular anomalies are commonly wrongly diagnosed and therefore wrongly treated. The accepted classification by International Society of Vascular Anomalies in Rome divides vascular anomalies into two categories: haemangiomas and vascular malformation. Haemangiomas are benign neoplasm of hyperplastic endothelium of capillaries, usually present a few weeks after birth but spontaneously involutes within 5-10 years. On the contrary, vascular malformations are single endothelial lined vascular spaces of different types: capillary, venous, lymphatic, arterial or combinations. They are present at birth, never regress and grow during lifetime. Non-pulsatile bluish submucosal swelling denotes the diagnosis of venous malformation. It was later confirmed on CT angiography by phlebolith which is the hall mark of venous malformation. Phleboliths are calcified

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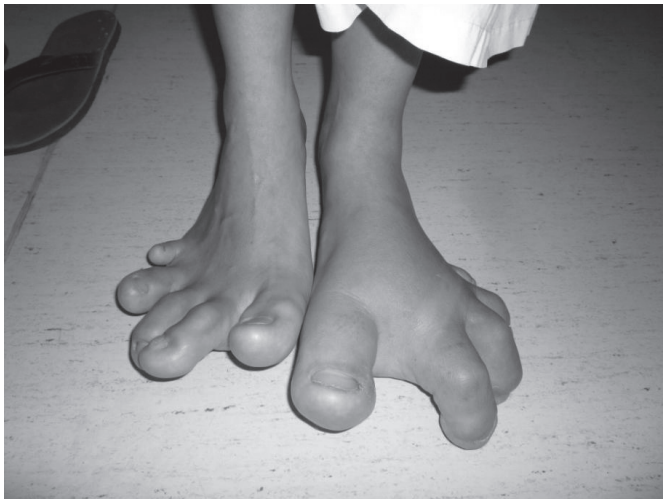

Figure 1: Partial Gigantism of the feet

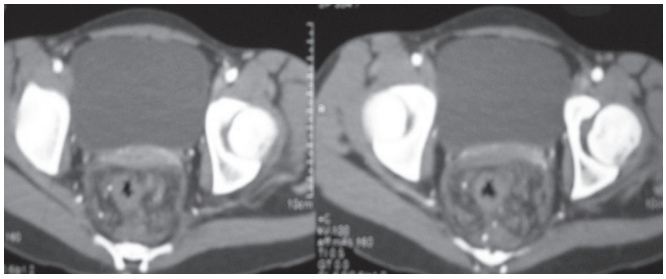

Figure 2: CT angiography scan of abdomen and pelvis showing a well defined, homogenous perirectal and anorectal soft tissue mass with multiple small calcifications

body formed in blood clots in these slow malformations. If the diagnosis of the type of vascular anomalies in doubt or not clear, the best investigation is to biopsy the lesion. However, expertise and facilities must be available to manage complications such as torrential bleed.

Given that the incidence of rectal vascular malformations is low, it is difficult to establish therapeutic guidelines. Most of the rectal vascular malformations can be managed conservatively. In centre where the interventional radiologists are available and the feeding vessels can be outlined, superselective microcoil embolization is a safe option. If not, surgical resection is recommended for patients with severe bleeding. A sphincter-saving procedure with anterior resection and coloanal anastomosis is most appropriate. The main technical difficulty of the surgery is to identify and divide the feeding vessels entering the vascular malformation which could pose the risk of severe intra-operative bleeding and such potential risk has to be discussed with the family members. The option of abdominoperineal resection is also an effective procedure with low mortality and morbidity but it is less desirable to give a young patient with benign disease a permanent colostomy.

In conclusion, rectal malformations are rare and often confused with rectal haemangiomas, which have a different management approaches.

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