ABSTRACT Blue rubber bleb nevus syndrome (BRBNS) is a rare condition characterised by venous malformations in the skin, gastrointestinal tract and other parts of the body. Its presentation is usually sporadic, although cases of autosomal dominant inheritance have also been reported. Usually seen in children, BRBNS presentation in adults is rare. Symptoms at presentation depend on the organs involved; patients with BRBNS may present with acute or chronic gastrointestinal bleed. We herein report a rare presentation of BRBNS in an adult who suffered from intermittent abdominal pain and melaena for three years. Contrast-enhanced computed tomography revealed a jejunojejunal intussusception with a vascular malformation as the lead point. The patient underwent laparotomy with resection of the intussuscepted bowel segment. Recovery was uneventful. In spite of a wide range of therapeutic options for the management of BRBNS described in the literature, the efficacy of those available therapies, including surgical excision, is not well established.

Keywords: blue rubber bleb nevus syndrome, chronic gastrointestinal bleed, intussusception

INTRODUCTION
First described by William Bean in 1958, blue rubber bleb nevus syndrome (BRBNS) is a rare condition characterised by venous malformations in the skin, gastrointestinal tract and other parts of the body. Its presentation is usually sporadic, although cases of autosomal dominant inheritance have also been reported. BRBNS is usually seen in young children, and presentation in adults is rare. The skin and gastrointestinal tract are the most commonly involved, and symptoms at presentation vary according to the organ affected. Patients may present with acute gastrointestinal haemorrhage (in the form of haematemesis, melaena, or haematochezia) or chronic gastrointestinal bleed (in the form of iron deficiency anaemia). A wide range of surgical and nonsurgical modalities for the management of BRBNS have been described in the literature. However, the efficacy of the available therapies, including surgical excision, is not well established. We herein report a very rare case of BRBNS in an adult, who was successfully treated via surgical resection.

CASE REPORT
A 40-year-old man presented to our outpatient surgical department with symptoms of tiredness and easy fatiguability. The patient had no significant medical history apart from intermittent abdominal pain and melaena for three years. He appeared pale at presentation, but was haemodynamically stable, with a haemoglobin level of 6.7 g/dL. The rest of the physical examination was unremarkable. Oesophagogastroduodenoscopy and colonoscopy did not reveal the source of bleed. Contrast-enhanced computed tomography (CT) of the abdomen was performed to rule out other possible causes of bleed, showing a spectrum of findings, including a jejunojejunal intussusception with a probable vascular malformation as a lead point (Fig. 1), and the presence of multiple enhancing soft tissue density lesions in the subcutaneous plane of the anterior abdominal wall, which is suggestive of subcutaneous haemangioma and multiple hepatic haemangiomas (Fig. 2).

The patient underwent laparotomy with resection of the intussuscepted small bowel segment. The resected bowel revealed a polyp with superficial ulceration (Fig. 3) as a possible source of the bleed, and the lead point of the intussusception. Microscopic examination of the resected polyp showed dilated venous channels in the mucosa and submucosa. This spectrum of clinical, radiological and pathological findings is consistent with BRBNS.

DISCUSSION
Gastrointestinal lesions in BRBNS typically present as chronic continual bleed, requiring lifelong iron supplements or repeated blood transfusions. Very rarely do they present as overt bleeding with intussusception, as was seen in our patient. Upper and lower gastrointestinal endoscopy, contrast-enhanced CT of the abdomen,
and mesenteric angiography are the recommended diagnostic modalities for BRBNS. As part of the evaluation, gastrointestinal endoscopy (oesophagogastroduodenoscopy and colonoscopy) is the first investigation of choice, and may help in localising and detecting the extent of the disease. However, in cases with a small bowel lesion, as in our patient, gastrointestinal endoscopy may fail to locate the site of bleed. In such cases, contrast-enhanced CT of the abdomen may be helpful in the diagnosis. In conclusion, BRBNS is a rare surgical entity, and a wide range of therapeutic options including medical management, endoscopic therapies and surgical excision has been described. Since this syndrome is commonly associated with either occult or overt gastrointestinal bleed, it should be considered in any patient investigated for gastrointestinal bleed, as it will help in appropriate diagnosis and adequate management.

REFERENCES