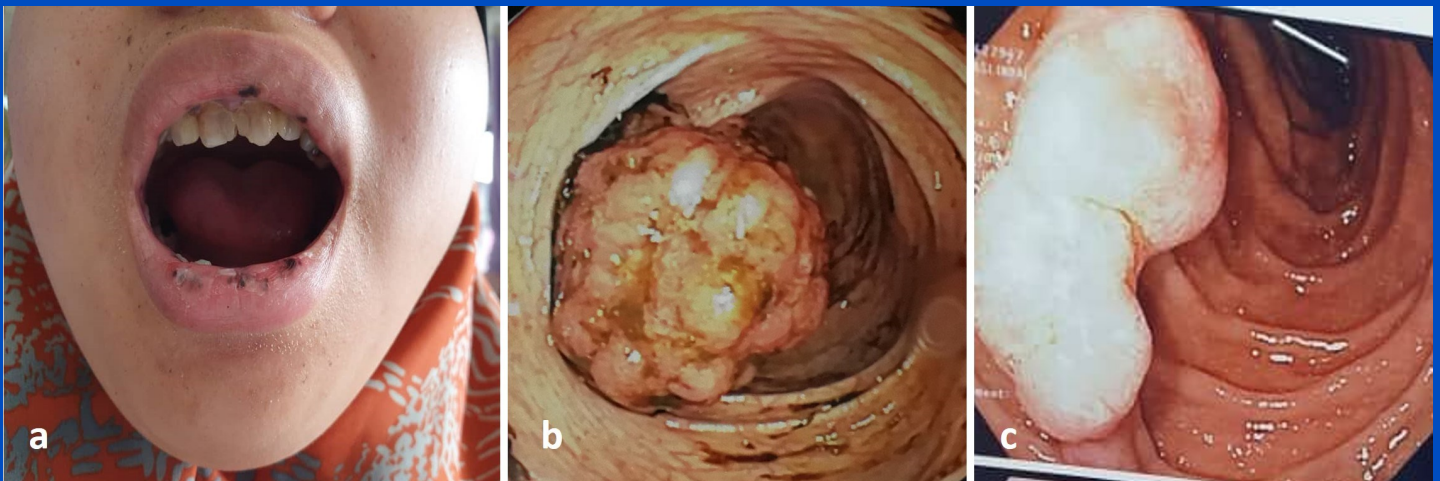


Brunei International Medical Journal

OFFICIAL PUBLICATION OF
THE MINISTRY OF HEALTH
AND
UNIVERSITI BRUNEI DARUSSALAM

Volume 16

7 December 2020 (21 Rabiulakhir 1442H)



Brunei Int Med J. 2020;16:137-138

Brunei International Medical Journal (BIMJ)

Official Publication of The Ministry of Health and Universiti Brunei Darussalam

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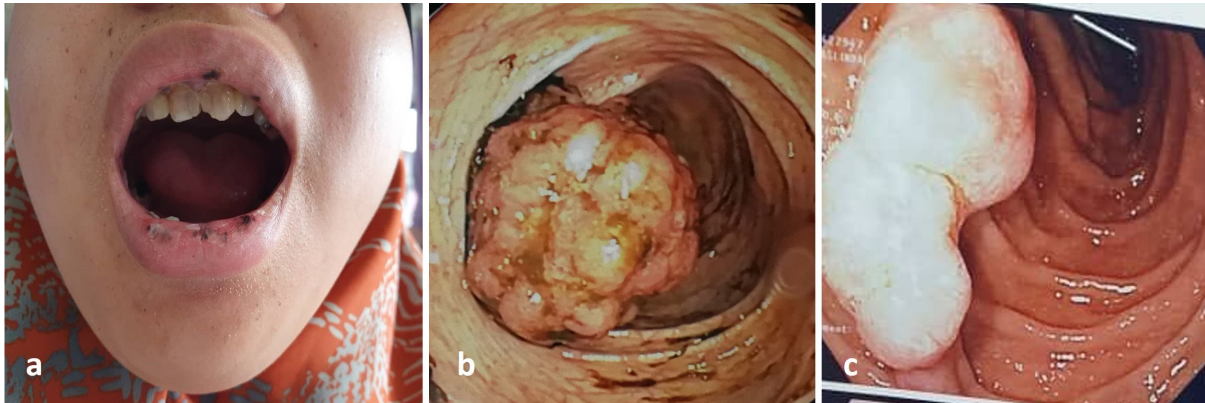


Figure 1

25 years old lady presented with anaemic symptoms of lethargy and dizziness. She had intermittent history of blood mixed with feces after defecation for the past one year. On examination, she had hyperpigmented lesion over lips as shown in figure 1a. Colonoscopy showed pedunculated polyps over multiple part of her large colon as shown in figure 1b. Esophagogastroduodenoscopy showed pedunculated polyps over duodenum as shown in figure 1c.

What is the diagnosis?

Answer: refer to page [138](#)

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(Refer to page [137](#))

ANSWER: PEUTZ-JEGHERS SYNDROME

Colonic polyp is one of the common cause of lower gastrointestinal bleed. However, Peutz-Jeghers syndrome (PJS), which is one of the spectrum of hamartomatous polyposis syndromes, is a rare autosomal dominant inherited disorder. PJS occur at approximately one tenth of the frequency of adenomatous polyposis syndrome and the incidence of Peutz-Jeghers syndrome is about 1 in 230,000 live births. The eponym of "Peutz-Jeghers Syndrome" originated from a Dutch physician named Jan Peutz, whom was the first to recognize the combination of intestinal polyposis, heredity and mucocutaneous pigmentation, and Joseph Jeghers, American physician whom written a published article regarding the syndrome in 1949.

The polyps of PJS are categorized as hamartomatous because they are comprised of epithelium consisting of the cellular elements usually present at that specific site of the gut and an extreme proliferation of smooth muscle encompassing into the lamina propria in an arborisation-like manner. Number of polyps can vary from one to hundreds and can occur in the stomach, small bowel and large bowel, which is similar to the case that we presented here. More than 50% of the individuals with PJS will experience polyp-related symptoms before the age of 20, and the median age of onset is 13 years old. They can come with small bowel intussusception and obstruction, abdominal pain, rectal bleeding and anaemia.

The estimated cumulative lifetime risk for any cancer in PJS individuals is in the range 85-93%. The commonest cancers identified were those located in the GI tract, with colorectal cancer as the highest cancer reported at 39%. Other PJS-associated cancers are breast cancer and cancers of the reproductive organs (fallopian tube, ovarian and uterine cancers). The management of individuals with PJS include surgical treatment of

curative intent and cancer risk management via routine surveillance.

Gastric and colonic polyps of individuals with PJS can be removed by endoscopic polypectomy. The management of small bowel polyp however is challenging as the patients usually come with intestinal obstruction secondary to small bowel intussusception. Complications of PJS such as bleeding and intussusception are managed by exploratory laparotomy with manual reduction of the intussusception and surgical polypectomy or segmental resection of the small bowel. In non-complicated PJS polyps, the option is polypectomy via push enteroscopy, but it can reach only beyond the ligament of Treitz up to 150cm.

The cause for PJS is mutation in tumor suppressor gene STK11 (alternatively known as LKB1) on chromosome 19p13.3, as there is correlation between the type of STK11 mutation and time to onset of PJS related symptoms. Genetic testing for STK11 mutation is recommended for PJS individuals. Once an STK11 mutation has been identified in the proband of genetic testing, at-risk blood relatives are recommended to undergo predictive testing.

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