



## Report of Peutz-Jeghers syndrome in two sisters with intussusception at the age of 10 and 12 year old

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### General Note

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## ABSTRACT

Peutz-Jeghers syndrome (PJS) is an inherited autosomal dominant condition that is characterized by various hamartomatous polyps in the gastrointestinal tract (GIT) and hyperpigmentation of the skin and mucous membranes. PJS increased risk for developing intestinal malignancy. *Cases presentation:* we report two sisters with intussusception at the age of 10 and 12 year old presented to the emergency department (ED) with features of intestinal obstruction. After thoroughly investigating, they diagnosed intussusception. Intraoperative was found intestinal polyps and by history, intraoperative findings, and histopathology, they confirmed diagnosis of PJS. These cases are reported because of the rarity of PJS and rarity of intussusception occurrence at 10 and 12 years. *Conclusions:* PJS is a rare familial disease. Early detection, treatment, and closely monitored can improve the prognosis of PJS and prevent complications related to polyps. PJS should be included as a differential diagnosis if intussusception happens at an atypical age.

**Keywords:** Peutz-jeghers syndrome; Intussusception; Hyperpigmentation; hamartomatous polyps; Intestinal malignancy

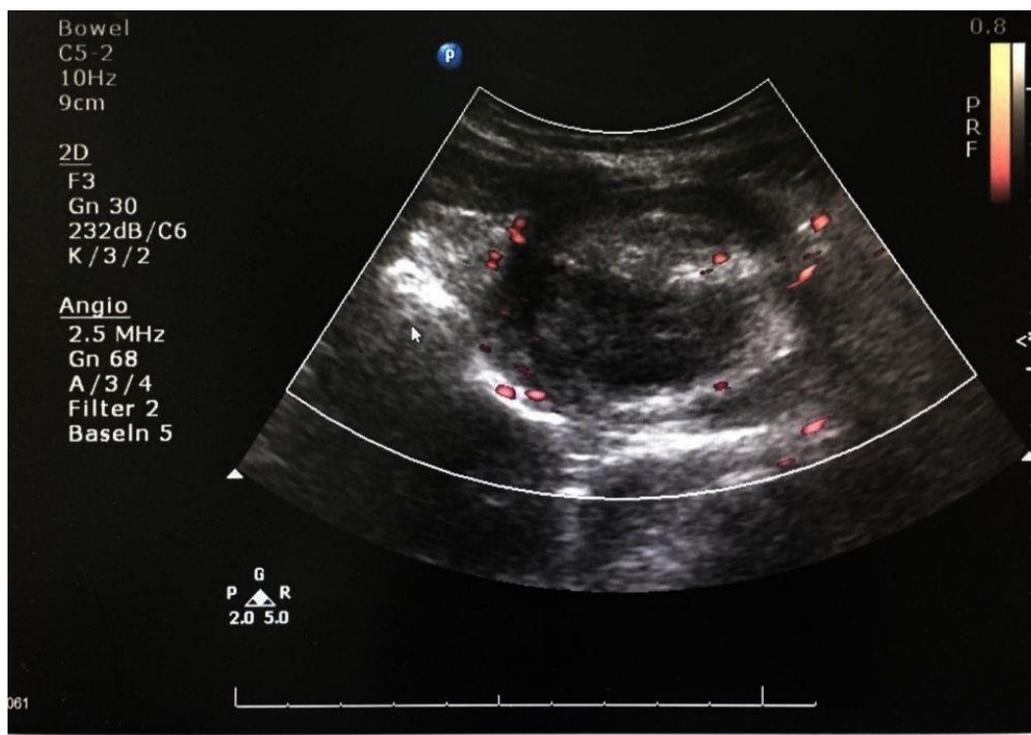
## 1. INTRODUCTION

PJS is a rare inherited disease which described by hamartomatous polyps, usually in GIT and hyperpigmentation of skin and mucous membranes (Shrivastava et al., 2013). The incidence of approximately one per 200000 births has PJS (Cai et al., 2020). The modes of inheritance in PJS are an autosomal dominant with incomplete penetrance (Bhattacharya et al., 2010). Both man and women's affected equally (Chung et al., 2019). PJS associated with a 15 fold increased risk for intestinal malignancy (Shrivastava et al., 2013; Choudhury et al., 2018). The majority of patients have a presentation of frequent abdominal pain and intermittent intussusception induced by polyps (Krishnan et al., 2015).

## 2. CASE PRESENTATIONS

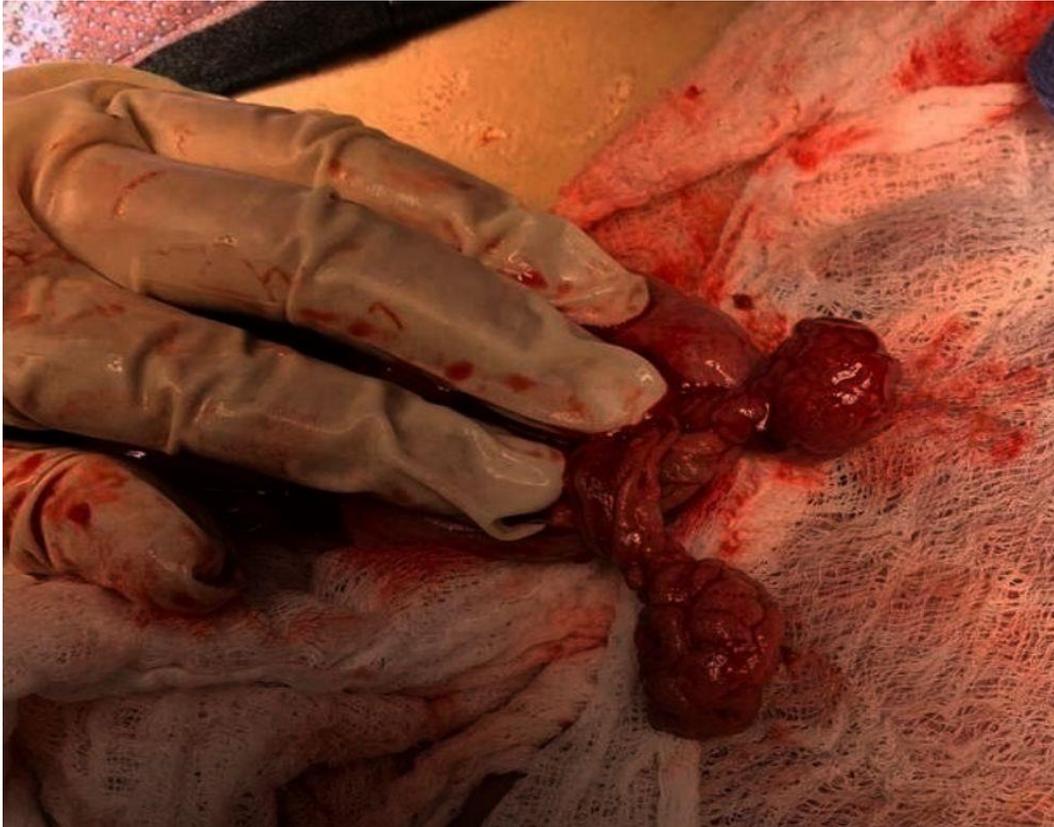
**Case A**

A 12-year-old Saudi girl, who appeared normal and without any history of previous diseases, was brought to ED with a history of sudden abdominal pain and vomiting for 24 hours. Abdomen examination revealed generalized tenderness with a huge tender mass in the mid-abdomen. She was vitally stable. Abdomen x-ray and US abdomen were done (Figure 1).



**Figure 1** Large comma shaped mass likely lesion seen in central of abdomen, seems lobulated, giving the appearance of bowel within bowel, as there is some fluid inside it with evidence bowel motion of its content intussusception for assessment

She was diagnosed to have intussusception and booked for surgery. Intraoperatively, a midline incision was done. Big intussusception mass was found in the abdomen. Intussusception mass was exteriorized outside the abdomen, and reduction trial was failed then to facilitate the reduction, the mesentery cutting at the proximal side of the intussusception. Small bowel polyps were found to be the cause of intussusception. The leading point was two polyps 140 cm from duodenojejunal flexure (DJ) (Figure 2), the small bowel was palpated, another three polyps were found two were found 30 cm from DJ and one 120 cm from DJ. Markedly distended bowel with questionable blood supply was resected. The leading point also resected. Then two enterotomies were done over polyps longitudinally. The polyps removed by transfixing the pedicel and closed transversally the colon contained a lot of small polyps. Two anastomoses were done, and the abdomen closed in ordinary fashion.



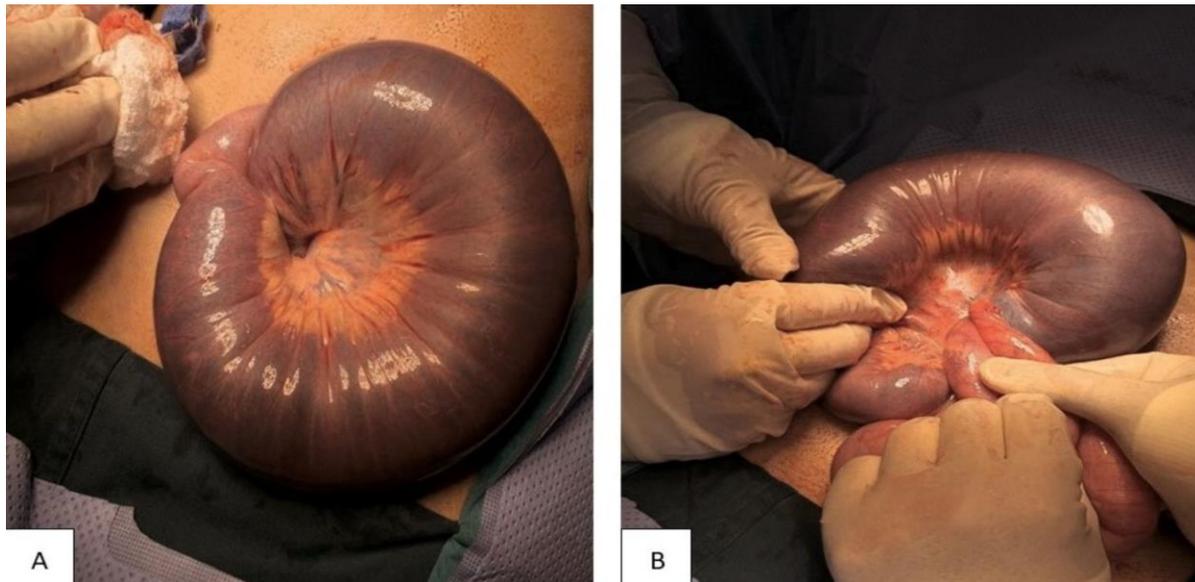
**Figure 2** The leading point was two polyps

Histopathology revealed villous adenomas. A further physical examination pigmented spots found in the inner surface of the lower lip. Her mother said she noticed the pigmented spots around her mouth when she was nine months old. A further family history, her brother, has pigmented spots in lips, and her mother underwent abdominal surgery at the age of 26 years due to intestinal obstruction due to polyps induce intussusception. While her sister, ten year old, came with the same complained before four days and was diagnosed as small bowel intussusception due to Meckel diverticulum.

Postoperative course: NGT removed on the third day, and liquid started on the fourth day, normal diet in five days, and she discharged one-week post-operative with regular and closely monitored. She diagnosed to have PJS. After one month, the patient was retained to the hospital with abdominal pain, and she managed conservatively and was discharged with good condition. 3 days after discharge patient came to the hospital with the same complained and diagnosed intestinal obstruction due to adhesion from previous surgery and treated surgically.

### Case B

A 10-year-old Saudi girl brought to the ED with a history of sudden abdominal pain for 5 days. The abdominal pain was associated with vomiting and constipation. She looks unwell and hemodynamically unstable. Abdomen examination revealed abdominal distention and generalized tenderness. X-ray and CT were done. She was diagnosed to have small bowel intussusception due to Meckel diverticulum. Intraoperatively, a midline incision was done. Big intussusception mass was found in the abdomen (Figure 3).



**Figure 3** A) Big intussusception mass B) Reduction trial

Intussusception mass exteriorized outside the abdomen, reduction tried was failed, then to facilitate the reduction, the mesentery cutting at the proximal side of the intussusception and anastomosis was done by using a surgical stapler, and the abdomen closed in ordinary fashion. Meckel diverticulum excision was done.

*Postoperative course:* she was on TPN for ten days. She was diagnosed to have mild pancreatitis and treated conservatively. She discharged with regular and closely monitored. She diagnosed to have PJS after the diagnosis of her sister. After two month, patient also retained to hospital with abdominal pain with partially intestinal obstruction due to adhesion. She vomited nine times per day the vomiting was associated with abdominal pain first periumbilical pain then shifted to RIF pain. Surgery was done, and lysis segment of the small bowel and prior to distal anastomosis was found fibrotic, adhesiolysis, resection, and anastomosis were done and discharged home after five days. Pigmented spots are seen in the inner surface of the lower lip and around the mouth. Further Family history is as mentioned above in case of A. Histopathology lab report revealed villous adenomas.

### 3. DISCUSSION

Peutz and Jegher is the first persons described Peutz-Jeghers syndrome in 1921 and 1944, respectively (Wang et al., 2011). PJS is an inherited autosomal dominant condition. PJS is a rare disease that is characterized by various hamartomatous polyps in the GIT and hyperpigmentation of the skin and mucous membranes. Approximately 90% of pigmentations around lips and buccal mucosa, but they can be seen in fingers, toes, nostrils, and perianal. The majority of pigmentations present during the first 12 months of life, and it may be subsided at adulthood or puberty. Hamartomatous polyps are predominant seen in GIT but although can be seen in other sites in urinary tracts, lung, bronchi, and gallbladder (Shrivastava et al., 2013; McGarrity et al., 2016). Approximately one per 200000 of births has a PJS (Cai et al., 2020). The most common cause of PJS is due to occurrence of Germline mutations of the serine/threonine kinase 11 (STK11) (LKB1) gene, which is the tumor suppressor gene located on chromosome 19 p 13 (Shrivastava et al., 2013; Derqaoui et al., 2020). It may occur with a family history or without family due to de novo mutations. Diagnostic criteria of PJS required the presence of a hamartoma accompanied by two of the following three signs: mucocutaneous pigmentation, polyps of the small intestine, or positive family history of PJS. Spots usually present during the first 12 months of life (Duan et al., 2017). As in these cases, case A presented at nine months, and case B presented at one year. Moreover, in this paper, patients have PJS manifestations involved a positive family history, polyps of the small bowel, and hamartoma are presenting.

PJS peak of occurrence of clinical presentations at the age of 30's years (Chung et al., 2019). In contrast, in the present case reports present at the age of 10 and 12 years. Sizes of polyps ranges from a few mm to 6 or 7 cm (Shrivastava et al., 2013). These polyps cause frequent abdominal pain due to intussusception, especially in the jejunum. Intussusception may resolve spontaneously, or it may cause an intestinal obstruction as in these cases. The occurrence of ulcers in these polyps may cause bleeding or chronic anemia (Shrivastava et al., 2013; Wang et al., 2011; McGarrity et al., 2016). These polyps can be detected through barium studies, Computer Tomography (CT scan) or ultrasonography (US) video capsule endoscopy, gastroscopy, colonoscopy, and magnetic resonance imaging. The US typically reveals 'target' or 'doughnut' signs. CT scan considered the most helpful modality for diagnosis intussusception (Wang et al., 2011). There is a strong Recommendation from the European Society for Paediatric Gastroenterology

Hepatology, and Nutrition (ESPGHAN) for PJS patients with symptomatic intussusception advisable to urgent underwent to surgical reduction without any role for radiological or endoscopic reduction of intussusception (Latchford et al., 2019).

Patients with PJS have an increased risk of intrainestinal and extraintestinal cancer. The polyps are commonly seen in the small intestine stomach, duodenum, and colon, while the extraintestinal can occur in pancreatic, breast, uterine, testicular, and ovary neoplasms (Wang et al., 2011; Chen et al., 2017). In this paper, the cases of PJS were surgically treated due to complications related to polyps. Regular screening is the primary way to prevent complications from PJS (Capasso et al., 2001).

#### 4. CONCLUSIONS

PJS is a precancerous syndrome. PJS should be included as a differential diagnosis if intussusception happens at an atypical age. The early detection and diagnosis of PJS in at-risk family members is very important in preventing the complications related to polyps, and closed lifelong follow up for these cases and screening their family required because they at risk of developing any gastrointestinal and extraintestinal cancers. It is necessary to investigate all first- degree relatives.

#### Authors' contributions

Mohammad Alnoaiji: Primary author, read and approved the final manuscript

Asmaa Salah Ghmaid, Ahmad Khaled Aleliwi, Tahani Nasser M Alrashidi: this work carried out in collaboration among all authors. All authors read and approved the final manuscript

#### Conflict of interests

The authors declare no conflict of interest.

#### Ethical approval and patient consent

Ethical approval was taken by ethics research committee at king Salman Armed forces hospital, Tabuk, Saudi Arabia (Ref. Number KSAFH-REC-2019-315). Written and Oral informed consent was obtained before writing these cases from all individual participants included in the study and their legal guardian.

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#### Abbreviations

Peutz-Jeghers syndrome (PJS), Gastrointestinal tract (GIT), Emergency department (ED), Computer Tomography (CT scan), Ultrasonography (US)

#### Data and materials availability

All data associated with this study are present in the paper.

#### Peer-review

External peer-review was done through double-blind method.

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