

## Peutz–Jegher’s syndrome: Three case reports

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Peutz-Jegher syndrome (PJS) is an autosomal dominant disease characterized by hamartomatous intestinal polyps associated with muco-cutaneous melanocytic macules. STK11/LBK1 is proved to be the responsible gene for inheritance<sup>1</sup>. Patients with PJS have 15 fold increase risk of developing malignancy<sup>2</sup>, which could be gastrointestinal as oesophageal, gastric, pancreatic and colonic or non intestinal cancer such as lung, breast, ovary and testicular cancer. Here we reported three patients presented to the Gastroenterology Department at Ibn Sina Hospital Sudan over the last ten years



### Case 1

A 30- year lady from western Sudan presented with recurrent episodes of colicky abdominal pain, distention, vomiting and absolute constipation relieved after conservative treatment. She underwent laparotomy and resection of bowel segment when she was ten years of age. Physical examination was unremarkable apart from an old midline scar. Her blood investigations were normal. Small bowel meal showed huge dilated small bowel loops with multiple filling defects (Fig 1).



Fig 1: Dilated jejunum with filling defects

Oesophago-gastro-duodenoscopy (OGD) showed no polyps. Surgical exploration revealed dilated jejunum with multiple jejunal polyps and jejuno jejunal intussusception (Fig 2) for which resection anastomosis was done. Polyps were resected through small entrotomy incisions. In retrospect physical examination showed oral melanosis (Fig 3). Histopathology confirmed PJS



Fig 2: Multiple Hamartomatous polyps

- 1-Surgeon
- 2- Surgical registrar
- 3- Gastroenterologist
- 4- Surgeon
- 5-Professor gastroenterologist
- 6- Professor of pathology



Fig 3: Melanotic spots in the mucosa of the lips

### Case 2

A 17- year old lady with presented with clinical features similar to those of the previous case (Case 1). In addition she has axillary freckle and underwent laparotomy twice for intestinal obstruction when she was four and eleven year of age. Endoscopy showed multiple polyps in stomach and colon, endoscopic polypectomy was performed. Small bowel meal was normal. Bowel obstruction was considered post-adhesive subacute intestinal obstruction and hence managed successfully conservatively. Histopathology proved hamartomatous polyps consistent with PJS.

### Case 3

A 9-year old boy presented with frequent vomiting for two month. Physical examination showed circumoral, palmar and planter dark depigmentations. OGD showed multiple gastric polyps and two large polyps in the second part of duodenum. Polypectomy was performed and PJS was diagnosed on the bases of histopathology.

The three cases are under regular follow up. Compliance for endoscopic screening of the family of the victims was rather poor except for the brother of the last case in whom no polyps were detected.

### Discussion

PJS was diagnosed for the first time in a Dutch family in 1921 by Peutz<sup>3</sup> and since that time many cases were reported, Duodenal polyps and intussusception seen in our cases have similar presentation to Wu Yk<sup>4</sup> reported case of an 18 years old man with repeated episode of vomiting due to gastroduodenal intussusceptions, but in

ours case obstruction was caused by the polyps themselves. Our diagnosis was rater late after multiple polyps were excised and revising the physical examination. laparotomies in childhood in the first and second cases did not give a clue because probably the specimens after the first surgery were not examined histologically or the patient was not well informed of the cause of his disease. Entrotomy for removal of polyps instead of repeated resection of bowel should be avoided to decrease the chance of short bowel syndrome. Intestinal and non intestinal malignant transformation is considerable. 50% of patients will die by the age of 57 due to malignancy<sup>5</sup>. For this reason patient education, family screening should continue life long. Fortunately none of our patients develop cancer probably because all are still younger than 40 years and the follow up period is not more than 10 years. Follow up need careful physical examination of breast, chest, abdomen and testes with request of relevant investigation as mammography, chest X-ray and ultrasound for abdomen, pelvis and testes. Family screening usually start in 2<sup>nd</sup> decade using stool for occult blood and upper and lower

gastrointestinal endoscopy every three to five years.

### Conclusion

PJS is rare in sudan with delay in detection of disease and lack of programme to follow patients and screen families.

### References

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