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Peutz-Jeghers Syndrome- A Rare Cause of Duodenal Obstruction: A Case Study in a Tertiary Care Hospital in Bangladesh

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Abstract **Case Report**

Peutz-Jeghers syndrome (PJS) is a rare hamartomatous polyposis of the gastrointestinal GI) tract, sometimes associated with pigmentation around lips and oral mucosa. The case of a 4-year-old girl who presented with duodenal obstruction is reported. Along with multiple small polyps in the duodenum, a large polyp was found to be the cause of the duodenal obstruction. Histologically most of the polyps were hamartoma. PJS is a rare autosomal dominant disease. Most patients have recurrent episodes of polyposis in the gastrointestinal tract causing gastrointestinal obstruction as well as these patients have an increased risk of both gastrointestinal and non-gastrointestinal malignancy and need regular screening.

Keywords: Peutz-Jeghers syndrome (PJS), Hamartomatous polyposis, Gastrointestinal (GI) tract, Polyps, Duodenum, Hematoma.

INTRODUCTION

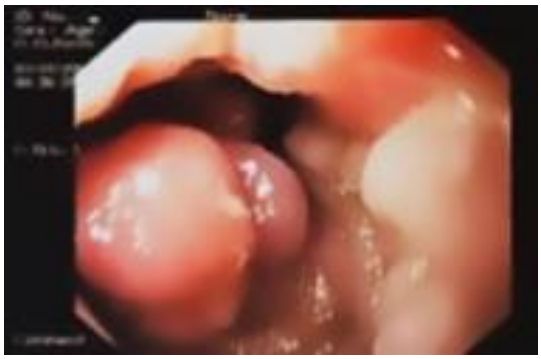
Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder resulting presented of GI tract polyposis causing obstruction and mucocutaneous melanin pigmentation [1]. Commonly resulting from mutation of serine-threonine kinase located on chromosome 19p34-p[36] known as STK11. Pigmentations are located mainly in the perioral region and appear in the infancy of life with increased intensity up to puberty then gradually fade in adult life. Sometimes pigmentation may be absent [2, 3]. Intestinal polyps are hamartomatous and the risk of developing cancer is variable [4]. In PJS GI tract polyps are hamartomatous with characteristics of histopathological appearance [5].

CASE REPORT

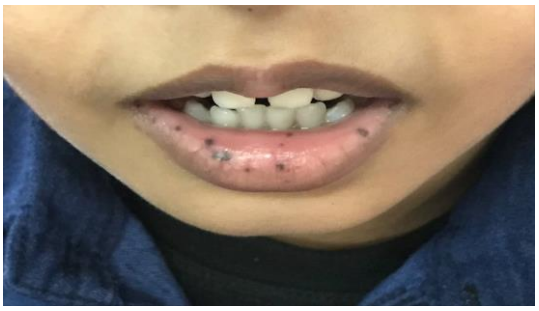
A 4-year-old girl presented to us with a history of recurrent episodes of vomiting for the last 6 months, the last episode lasting for 15 days. Vomiting occurred after each feed containing undigested food particles. She had no history of hematemesis, or malaena. Examination showed suspected upper gastrointestinal tract obstruction. As delayed gastric emptying on Ba meal follow-through x-ray, we have done upper gastrointestinal tract endoscopy showing one large pedunculated polyp in the 2nd part of the duodenum along with multiple small polyps in the duodenum (Picture-1). When we examined her, we didn't find any pigmented spots on her lip, buccal mucosa, or anywhere mucocutaneous junctions, but we have found pigmented spots on the lip of her one sibling (Picture-2). Intraoperatively we have found multiple polyps extending from the 1st part to the 3rd part of the duodenum. Duodenal mucosectomy along with the removal of all polyps was done (Picture 3).

Histologically, most of the polyps were hamartoma supported by broad bands of muscular mucosa with an arborizing pattern or Christmas tree appearance, thus confirming the clinical diagnosis of PJS (Picture 4).

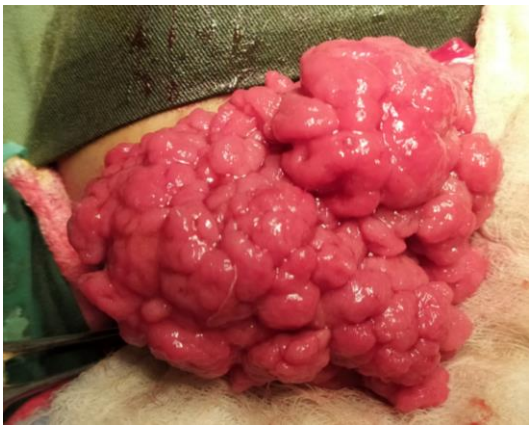
After the operation, her condition improved and the girl was discharged from the hospital in good condition she will be regularly monitored.



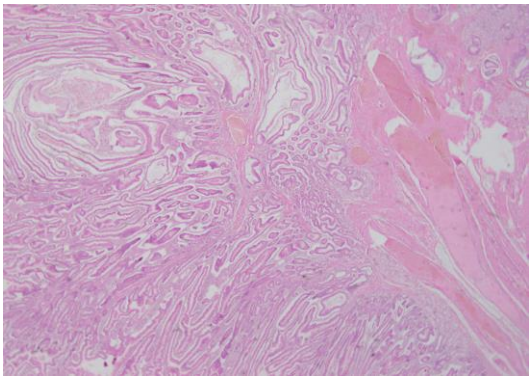
Picture 1: Polyp in the duodenum



Picture 2: Mucocutaneous spot on her sibling



Picture 3: Multiple polyps in the duodenum



Picture 4: Histopathology of Polyp

DISCUSSION

PJS is a rare autosomal dominant syndrome overall incidence is 1 in 120000 live births [6]. The most common clinical presentations are melanin pigmentations, commonly around the lip and buccal mucosa, also be present in other parts of the body like fingers, toes, hands, feet, and mucosa of the nose, conjunctiva, and rectum along with gastrointestinal polyps and positive family history. In some patients, pigmentation may be absent. Multiple hamartomatous polyps in the GI tract are the hallmarks of PJS [7]. Presentation of polyps includes colicky abdominal pain, bleeding, and bowel obstruction [3, 8]. Commencement of abdominal symptoms may vary and may present as early as the first year of life or at the age fourth decade [9]. 30% of patients with PJS may require laparotomy by the ten years of life. Sometimes patients with PJS may need multiple laparotomies in life. In our case polyps in the duodenum were found at operation, some of them were histologically confirmed as hamartomas, but she has no typical mucocutaneous pigmentation. In 20% of cases, typical mucocutaneous hyperpigmentation may be absent [10]. On searching, we have found hyperpigmentation in the lip of her one sibling who is under evaluation for PJS. Germline mutation in the tumor suppressor gene STK11 causes a very high risk of developing cancer. 66% increased risk of developing GI cancer by the time of 70 years of age. Female patients have a 32% increased risk of developing breast cancer at 60 years of age [6, 8, 9]. Treatment depends on the clinical presentation with severity. Surgical intervention is most often required in case of bowel obstruction. Radiographic imaging, endoscopy, virtual colonography, etc may be able to discover polyps in a patient with abdominal pain or GI bleeding that can be resected either endoscopically or by enterotomy, depending on the size of polyps.

CONCLUSIONS

The presence of perioral hyperpigmentation in early infancy should alert the clinicians to PJS and patients with PJS should be regularly monitored for the early detection of cancer.

REFERENCES

1. Giardiello, F. M., & Trimbath, J. D. (2006). Peutz-Jeghers syndrome and management recommendations. *Clinical Gastroenterology and Hepatology*, *4*(4), 408-415.
2. Spigelman, A. D., Arese, P., & Phillips, R. K. S. (1995). Polyposis: the Peutz-Jeghers syndrome. *Journal of British Surgery*, *82*(10), 1311-1314.
3. Homan, M., Dolenc Strazar, Z., & Orel, R. (2005). Peutz-Jeghers syndrome. A case report. *Acta Dermatovenerol Alp Panonica Adriat*, *14*(1), 26-29.

4. Konishi, F., Wyse, N. E., Muto, T., Sawada, T., Morioka, Y., Sugimura, H., & Yamaguchi, K. (1987). Peutz-Jeghers polyposis associated with carcinoma of the digestive organs. *Diseases of the colon & rectum*, *30*(10), 790-799.
5. Perzin, K. H., & Bridge, M. F. (1982). Adenomatous and carcinomatous changes in hamartomatous polyps of the small intestine (Peutz-Jeghers syndrome): Report of a case and review of the literature. *Cancer*, *49*(5), 971-983.
6. Georgescu, E. F., Stanescu, L., Simionescu, C., Georgescu, I., Ionescu, R., & Florescu, G. (2008). Peutz-Jeghers syndrome: case report and literature review. *Rom J Morphol Embryol*, *49*(2), 241-245.
7. Mathus-Vliegen, E. M. H., & Tytgat, G. N. J. (1985). Peutz-Jeghers syndrome: clinical presentation and new therapeutic strategy. *Endoscopy*, *17*(03), 102-104.
8. Sheperd, N. A., Bussey, H. J. R., & Jass, J. R. (1987). Epithelial Misplacement in Peutz-Heghers Polyps. *The American Journal of Surgical Pathology*, 11(10), 743-749.
9. Beggs, A. D., Latchford, A. R., Vasen, H. F., Moslein, G., Alonso, A., Aretz, S., ... & Hodgson, S. V. (2010). Peutz–Jeghers syndrome: a systematic review and recommendations for management. *Gut*, *59*(7), 975-986.
10. Spigelman, A. D., & Phillips, R. K. S. (1989). Management of the Peutz-Jeghers patient. *Journal of the Royal Society of Medicine*, *82*(11), 681-681.