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Case Study

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RARE PEUTZ JEGHERS SYNDROME IN PEDIATRIC AGE: MERELY ON UPPER GI WITH ADENOMATOUS CHANGE

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare, hereditary, and autosomal dominant disorder usually presented with mucocutaneous pigmentation and gastrointestinal (GI) polyps. GI polyps usually hamartomatous polyps located throughout the gastrointestinal tract and may be presented with bleeding and small bowel intussusception, potentially leading to the need for emergency surgery. We represent a case of Peutz-Jeghers syndrome in a 05-year-old female child who presented with abdominal pain, vomiting, and upper GI bleeding. This syndrome is a rare entity that required regular follow-up with screening for other malignancies due to increased chances of intestinal and extra-intestinal malignancy.

KEYWORDS: Hamartomatous polyp, Melanoma cutaneous pigmentation, Peutz-Jeghers syndrome, Intestinal polyps, Child polyposis.

INTRODUCTION

Peutz-Jeghers syndrome is a rare, autosomal dominant, hereditary polyposis syndrome characterized by gastrointestinal hamartomas and mucocutaneous pigmentations.[1] PJS was first described by Dr. Connor in 1895 in the scientific society of London and further by Johannes Peutz in 1921 and by Harol Joseph Jeghers in 1949.[2, 3] The Incidence has been estimated between 1 in 8,300 -200,000 live births.[1, 4] PJS presents with multiple hamartomatous GI polyps less than 100 in number with characteristic periorificial, palms and soles of the feet melanin pigmentation.[5] These polyps are usually 1 mm to 4 cm in diameter, mostly seen in the jejunum and small bowel than the colon. They can also occur at the nose, bronchi, renal pelvis, and biliary tree.[1, 5, 6] It is caused by a germline mutation in the serine/ threonine kinase 11 or liver kinase B1 (STK11/LKB1) genes and the mutation is localized to chromosome 19p13.3.[1, 7] Patients with Peutz-Jeghers syndrome have a There is a chance of 15-fold increased risk of developing intestinal cancer compared with the general population in patients with PJS but in children, cancer is extremely rare.[7, 8] Here we report a case of a 5-year-old girl who was presenting with GI polyp merely in upper GI.

CASE REPORT

Our case was a 5-year-old girl presented in our gastroenterology department with the complaint of abdominal pain and vomiting for 18 days and a single episode of hematemesis. Abdominal pain was intermittent, diffuse, dull aching in nature, not associated with a meal and there was no aggravating or relieving factor. Vomiting was non-projectile, not bilious. She had a history of occasional passes of melena during her illness. There was no history of altered bowel habits, fever, anorexia, weight loss, or any other significant systemic complaints. Family history was not contributory. Physical examination showed moderate pallor, malnourished, and few blackish pigmentations on lips [Figure 1] but other areas like the trunk, extremities, back, external genitalia, gums, and palate were spared.

Routine laboratory investigations [Table 1] showed Hb 5.8 mg/dl, stool OBT was positive. LFT, RFT, urine examination was within normal limits. Endoscopy of upper GI revealed, multiple polyps of variable size and shape are seen in the fundus and body of the stomach. One of the polyps was large and cauliflower shape, some are pedunculated, some were sessile and 3 polyps are seen in the 1st and 2nd part of the duodenum [Figure 2]. Polypectomy was done. But colonoscopy revealed normal findings. On histopathology, Microscopic examination revealed polyp with arborization of smooth muscle within the lamina propria resembling Christmas tree at low power [Figure 3A & 3B] and some dysplastic foci but not features of malignancy. We diagnosed her with Peutz-Jeghers syndrome.

Table 1: Laboratory and radiological investigations		
Investigation	**Results**	**Normal range**
Complete Blood Count		
Haemoglobin (g/dL)	5.8	13-17
Erythrocyte sedimentation rate (mm in 1st hr)	5	0-10
White blood cell count (/cu mm)	10,000	4,500-11,000
Neutrophil leucocytes (%)	70	40-80
Lymphocytes (%)	26	20-40
Platelet count (/cu mm)	4,50,000	150,000-400,000
Liver function tests		
Prothrombin Time (sec)	12.5	12-16
International normalized ratio	1.00	<1.4
Alanine aminotransferase (U/L)	29	35-50
Serum creatinine (mg/dl)	0.5	0.9-1.3
Urine R/M/E	Normal	
Endoscopy of the upper GI tract	Multiple polyps of variable size and shape are seen in the fundus and body of the stomach. One of the polyps was large and cauliflower shape, some are pedunculated, some are sessile and 3 polyps are seen in 1st and 2nd parts of the duodenum [Figure 2A & 2B].	

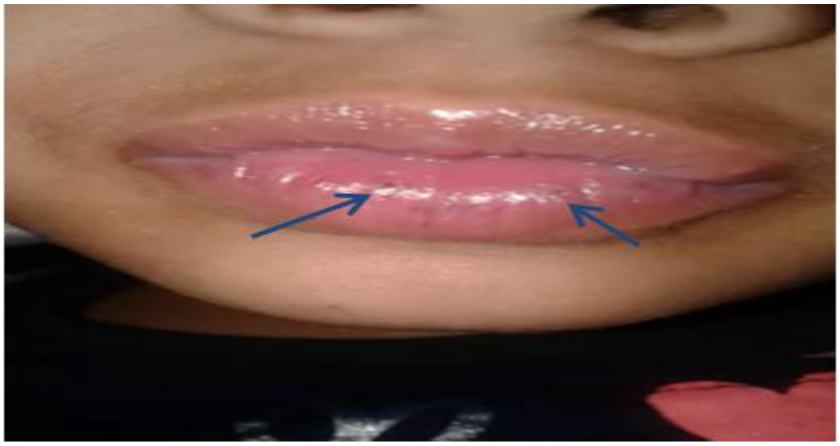

Figure 1: Multiple pin-headed pigmentations on the lower lip.

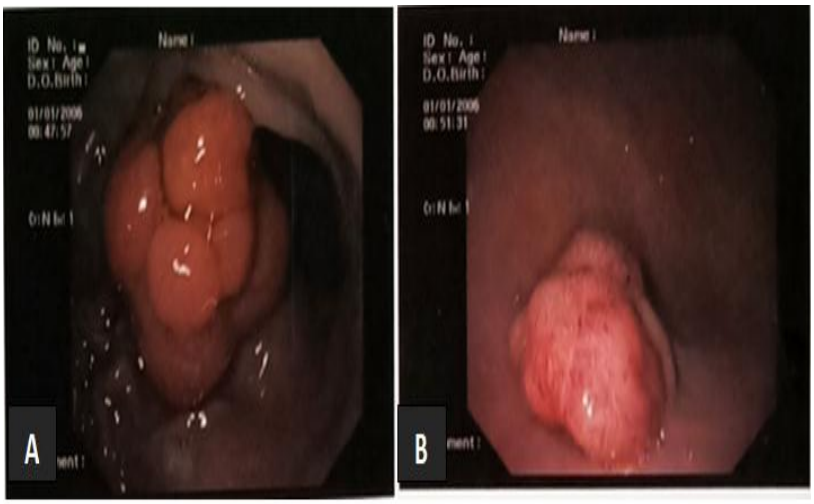

Figure 2: Cauliflower-like polyp in the fundus (A) and pedunculated polyp in the body of the stomach (B).

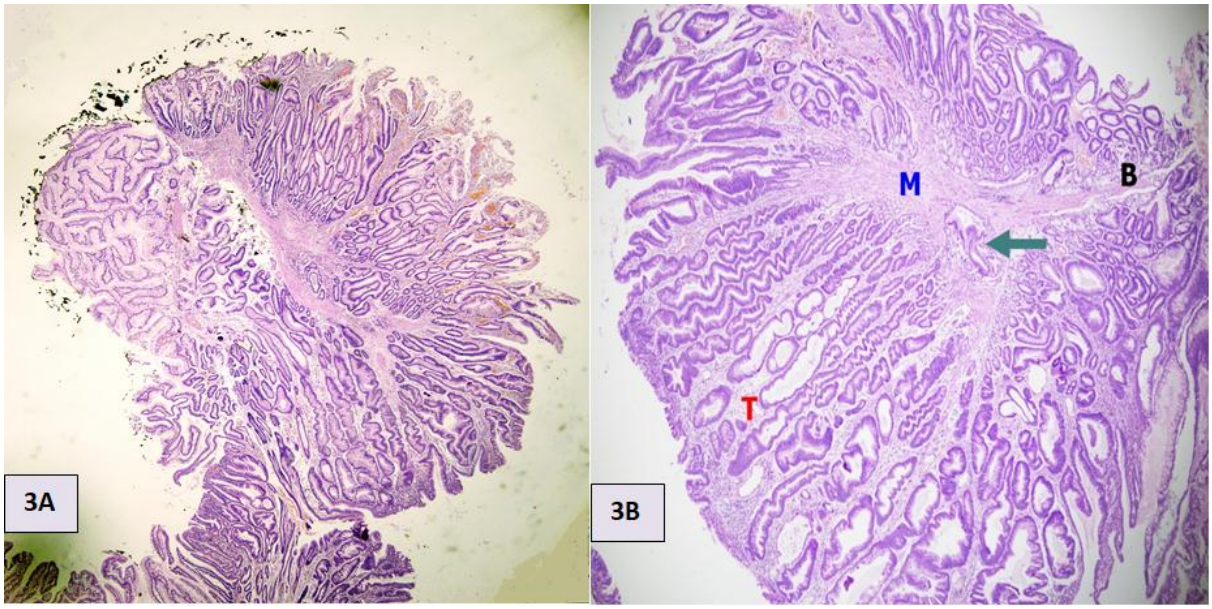

Figure 3A: Low magnification view of the polyp showing a pedicle and lobulated Christmas tree appearance (H&E x40) & Figure 3B: Peutz-Jegher's polyp showing arborization of smooth muscle (M) within the lamina propria, tip (T) and base (B) of the polyp. Arrow shows epithelial misplacement (pseudoinvasion) (H&E x120).

DISCUSSION

PJS is an autosomal dominant with variable inheritance disorder, manifested by usually hamartomatous polyps in the GI tract commonly in the small intestine, and pigmented mucocutaneous lesions.[9] It is associated with a significant risk of serious complications and an increased risk of malignancy.[1]

PJS having phenotypic variability that ranges from an asymptomatic patient with mucocutaneous melanic pigmentation, to emergencies due to GI invagination of polyps. Patients with PJS may develop abdominal pain, vomiting, and GI bleeding due to the intestinal polyp and symptoms from obstruction of large polyps, anemia, hematochezia, hematemesis, biliary obstruction, and gastric outlet obstruction. Acute blood loss or chronic anemia due to ulceration of Peutz-Jeghers polyps may also occur. Symptoms are related to the size, location, and character of the polyp.[1, 3] In this case, the child presented with abdominal pain, vomiting, hematemesis, and occasional melena.

PJS has two major clinical features: hamartomatous polyposis of the gastrointestinal tract and mucocutaneous pigmentation.[7,9] Hyperpigmentations are a common manifestation of PJS (in 95%), due to melanin-laden macrophages within the dermis. It usually manifests in infancy & early childhood as flat, greyish, or brown spots, up to 4mm in size commonly on the lips and buccal mucosa but may present on nostrils, fingers, palms, soles, perianal area, genitals, and intestinal mucosa. Absence alone cannot exclude the diagnosis of PJS because it may not be present in the first years of life or maybe subtle.[1,7,10] In our case pigmentation was found on the lower lip only.

Polyps in PJS vary in size from 1 cm to 3.5 cm in diameter and maybe pedunculated or sessile occur in the small intestine most frequently in the jejunum followed by the ileum, duodenum, the colon & the stomach.[1, 7, 11] Although Peutz-Jeghers polyps are most commonly found in the gastrointestinal system, they can also occur in extra-intestinal sites such as the kidney, ureter, gall-bladder, bronchial tree, and nasal passages.[7] Histopathological type of polyps is hamartomas in most of the cases but adenomatous, hyperplastic, inflammatory, and mixed polyps less frequently may found in some cases.[1,12] Histologically, hamartoma is characterized by an overgrowth of cells composed of non-neoplastic tissue native to the area in which they normally occur, however, the original architecture is markedly distorted.[1] PJS-associated polyps can be differentiated from sporadic hamartomatous polyps and hamartomatous polyps associated with other syndromes by a unique central core of smooth muscle that extends into the polyp in an arborizing fashion (Christmas tree-like appearance) and that is covered by either normal or hyperplastic mucosa native to the involved site.[1,7,9,14] In our case, we found multiple pedunculated and sessile polyps in the stomach and duodenum but no extra-intestinal polyp was seen.

The clinical diagnosis of this rare disease is made with one of the following criteria: (1) Two or more histologically confirmed PJ polyps (2) Any number of PJ polyps detected in an individual with a close relative with PJS (3) Characteristic mucocutaneous pigmentation in an individual with a family history of PJS or (4) Any number of PJ polyps in a patient characteristic mucocutaneous pigmentation.[3,13] In our case, we diagnosed PJS as the child having mucocutaneous pigmentation and histologically confirmed PJ polyp.

The standard therapy is polypectomy in symptomatic patients either endoscopically or laparotomy and long-time surveillance for symptoms and signs derived from polyps, such as anemia, GI bleeding, abdominal pain, and hormonal production, such as precocious puberty, gynecomastia, and intestinal or extra-intestinal malignancy. Many patients may require multiple surgical resections and bowel resection to remove symptomatic gastrointestinal polyps that cause persistent or recurrent intussusceptions which may develop short gut syndrome.[3,7,13]

CONCLUSION

Because of the risk of polyps-related complications, and its association with cancers although it is less frequent, PJS needs frequent monitoring of the symptoms and performing endoscopic procedures for those in whom the syndrome is suspected and patients already diagnosed, to avid-losing important bowel segments and death.

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