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Acute abdomen in a 6 year old disclosed as an inherited autosomal disorder: A case report

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Abstract: A rare syndrome presenting with muco-cutaneous pigmentation with hamartomatous polyposis of intestine associated with increased risk of gastrointestinal and other systemic malignancies popularly termed as Peutz- Jeghers Syndrome present as varied course of symptoms, some report to dermatologist for pigmented oral macules, some report in emergency with acute abdomen while others are diagnosed incidentally. We report a 6 years old male child brought to emergency department with acute abdomen subsequently diagnosed with intussusception caused by a hamartomatous polyp which later on combined with pigmented macules on buccal mucosa ended up as an autosomal dominant syndrome.
Keywords: Hamartomatous polyp, intussception, target sign, bilious vomiting.

INTRODUCTION:

Peutz-Jeghers syndrome with equal male and female distribution characterized by typical pigmented perioral macules, pigmented spots in the buccal mucosa which are present in 90% of patients, and multiple, although rarely more than 20 hamartomatous polyps predominantly in the gastrointestinal (GI) tract [1]. Most patients have a characteristic clinical course of recurrent episodes of polyp induced bowel obstruction and bleeding. Diagnosis is based on at least 1 hamartomatous polyp associated with 2 of the 3 following criteria: family record of PJS, polyposis localised on small bowel, and muco-cutaneous pigmentation. The risk of gastrointestinal and extra-gastrointestinal malignancies is significantly increased in PJS patients [2]. The gene for this inherited autosomal disorder is localized to chromosome 19p34-p36 and is known as STK 11, a serine-threonine kinase involved in growth control regulation. The relative risk of dying from a gastrointestinal cancer is 13 times greater. The risk of any other malignancy including cancer of the reproductive organs and breast, and also of the pancreas and lung is 9 times greater than in the general population [3] .By early diagnosis, prompt treatment, continuous follow up, and appropriate lifetime screening for malignancies, mortality and morbidity of PJS can greatly be reduced [4-6].

CASE REPORT:

6 years old male brought to emergency department with complaint of sharp, intermittent abdominal pain, sudden in onset around umbilical area associated with bilious vomiting, there was no history of per rectal bleeding or melena. Family history was not contributory. Physical examination revealed pallor, black pigmented macules on buccal mucosa

Figure 1 which were present since childhood as per parents. On abdominal examination, periumbilical tenderness present with diminished bowel sounds.

USG Abdomen was performed, classic TARGET SIGN of intussuception was seen Figure 2. Hematological study revealed microcytic hypochromic anaemia. Patient was taken in for emergency exploratory laprotomy under general anaesthesia, intussception was reduced Figure 3, small intestine revealed multiple polypoidal masses Figure 4. Post-operative period was uneventful. Histopathology of the section confirmed hamartomatous poylp with sparse arborization of muscularis mucosae covered with normal villi. A close follow up was kept and patient is disease free.

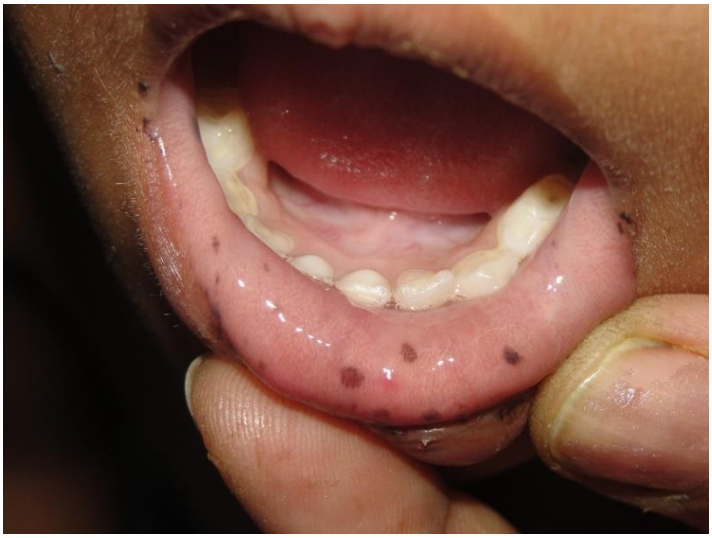

Fig 1: Pigmented macules on oral mucosa

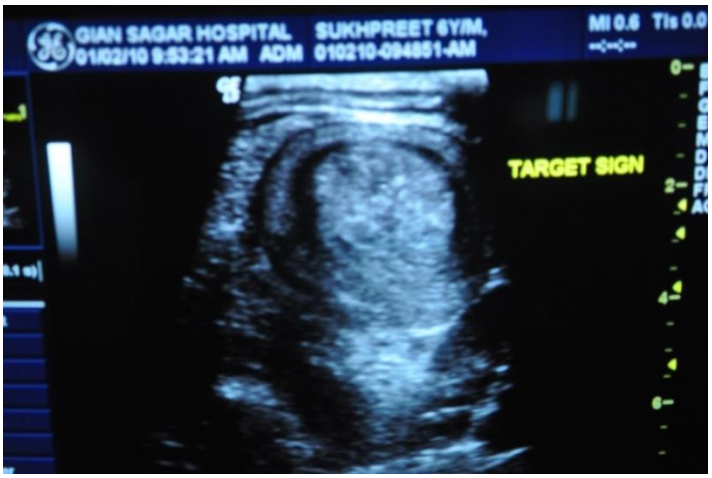

Fig 2: Classic TARGET SIGN of intussuception

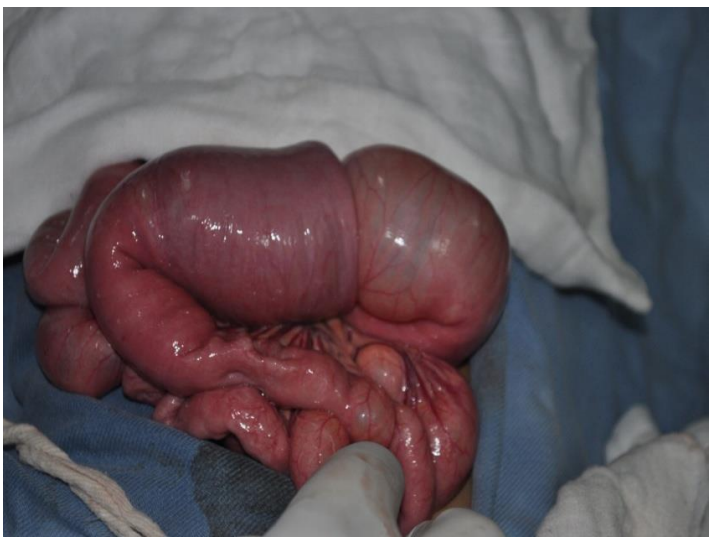

Fig 3: Intraoperative image of intussuception

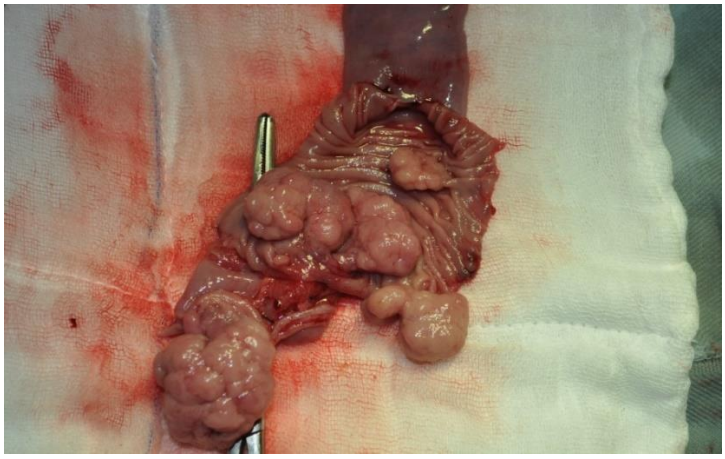

Fig 3: Small intestine revealed multiple polypoidal masses

DISCUSSION:

The primary description of PJS was published by Peutz in 1921 in one Dutch family (the Harrisburg family) as a gastrointestinal familial polyposis with pigmentations. Jeghers specified the description in 10 cases from different families in his work in 1949 and defined the relations between pigmented lesions, gastrointestinal polyposis and increased risk of carcinoma; approximately half of his patients suffered from gastrointestinal malignancy [7, 8]. The Peutz-Jeghers syndrome consists of two major components: hamartomatous polyposis of the gastrointestinal tract and muco-cutaneous pigmentation .The hyperpigmented lesions contain melanotic deposits and commonly manifest in infancy and childhood. Pigmented lesions could fade during puberty and adulthood. Peutz-Jeghers polyps can also ulcerate, leading to acute blood loss or chronic anemia. The Peutz-Jeghers polyp varies in size from<1cmto>3.5 cm in diameter, and may be pedunculated or sessile. Prophylactic colectomy is an approach if numerous polyps are encountered. In a known case of PJS, upper GI endoscopy, colonoscopy and pelvic examination every 2 years is recommended [9]. Recently, intraoperative endoscopy and endoscopic polypectomy, rather than segmental resection of the bowel, have been recommended. The new mouth to anus (M2A) capsule endoscopy will probably become the most useful screening tool in the near future.

CONCLUSION:

The present paper emphasizes the importance of collateral associations of symptoms and signs in the diagnosis of concealed systemic disease. Prompt diagnosis is crucial for early diagnosis and minimizes the significant morbidity. A lifetime screening for gastrointestinal and other systemic malignancies is an important aspect for management of this syndrome.

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