CASE REPORT

Peutz- Jeghers’ syndrome, a rare genetic disorder: A case report

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Abstract
Hamartomatous polyposis syndromes or Peutz-Jeghers syndrome (PJS) is a hereditary autosomal dominant disease characterized by benign hamartomatous polyps and mucocutaneous pigmentation in the digestive tract. It occurs mostly in the small intestine during first decade of life but frequently in the colon and stomach. Only a few cases have been reported in the duodenum [1]. Polyposis syndromes are common cause of adult intussusceptions, with polyps acting as lead points. Adult intussusceptions are rare and is almost always associated with that lead point [2]. Although hamartomatous polyps are not pre-malignant, there is an increased risk of gastrointestinal and non-gastrointestinal malignancy, commonly involving the small bowel. Most patients of PJS presents with acute abdomen and diagnosed as intussusceptions, commonly entero-enteric type but colo-colic intussusceptions are rare in Peutz-Jeghers syndrome [3]. To the best of our knowledge, synchronous colo-colic intussusception association in Peutz-Jeghers syndrome has not been previously reported.

1 | INTRODUCTION

Peutz-Jeghers syndrome (PJS) is related to STK11 gene, with an incidence of 1 in 50,000-200,000 live births. It is an autosomal dominant condition with incomplete penetrance. Non-sense, frameshift and missense mutations inactivating the LKB1 gene on chromosome 19p13.3 have been implicated as the underlying abnormality [4], [5]. PJS presents with characteristic flat, pigmented, freckle-like cutaneous lesions mainly on the lower lip, perioral area, buccal mucosa, periorbital area and eyelids. The syndrome is also associated with gastrointestinal and extragastrointestinal Supplementary information

The online version of this article (https://doi.org/10.15520/jmrhs.v3i7.221) contains supplementary material, which is available to authorized users.

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hamartomatous polyps. The typical pathological feature of Peutz-Jegher polyp (PJP) is a smooth muscle core arising from the muscularis mucosae and ramifying into the substance of the polyp like the branches of a tree [6].

The World Health Organisation (WHO) clinico-pathological criteria for diagnosing this rare disorder are:

1. Three or more polyps, which show histological features consistent with PJS.
2. A family history of PJS with any number of PJPs.
3. A family history of PJS with characteristic mucocutaneous pigmentation.
4. Characteristic mucocutaneous pigmentation with any number of PJPs [7].

Individuals with this condition carry a very high risk of developing not only gastrointestinal adenocarcinoma but also extra-gastrointestinal malignancies in the breast, pancreas, testes and ovary. Compared with the normal population, PJS subjects have a relative risk of 15% for developing any such type of malignancy. Pseudo-invasion, mimicking adenocarcinoma, is described in nearly 10% cases of PJS. It is thought that the mechanical pressure resulting from intussusception of small bowel polyps in PJS may be responsible for misplacing luminal epithelial cells through normal anatomic defects in the intestinal wall, particularly the ones caused by traversing neurovascular bundles. Published review articles can be referenced for further information about this disease, of the many published case reports with solitary or multiple PJPs, most patients presented with bleeding and intestinal intussusception. To the best of our knowledge, in all those reports of patients with single or multiple enteric intussusception, abdominal signs and symptoms of some kind were present. This led the clinician to suspect the condition is a surgical emergency. Here we report a case of multiple small intestinal intussusception in a young adult man presented with melena, but with a completely unremarkable abdominal examination [8], [9].

2 | CASE PRESENTATION:

Master Fardin Mia 14 years old, 1st issue of non-consanguineous parents, hailing from Narayangong Dhaka, admitted in this hospital with the complaints of pain in the epigastric region for 14 days, which was severe in intensity, cramping in nature, radiate to whole abdomen, persistent for few minutes to hours, increased by taking food and sometimes relieved by defecation. He also complained of vomiting which was painless, projectile, large in volume, contains undigested food, not blood or bile stained. He also complains of substantial loss of weight and weakness. He also complains of constipation occasionally. His bladder habit is normal. He has history of volvulus which was operated and his mother has colon cancer for which she is taking chemotherapy. On examination he was ill looking, moderately anemic, vitals were normal. He had scattered hyperpigmented macules around lower lip which was irregular in shape with no definite boundaries, did not protrude skin or fade on pressure (Figure-1).

FIGURE 1: Hyperpigmented macules on the Patient's lips

On Gastrointestinal examination, hyperpigmented spots were present in the lower lip, vertical scar mark present, non-tender, no organomegaly, percussion note was tympanic, bowel sound present, on DRE no blood or mucus was present. Other systemic examinations were normal. On investigations, Hb was 6.0 gm/dl, MCV 14.1 pg, urine R/E was normal, S.Amylase was 19 U/L,
SGPT, S.Creatinine was also normal, USG shows multiple intussusception of bowel loops in left hypochondriac region and pelvic cavity, mild splenomegaly and cystitis. OBT was positive, Upper GI endoscopy shows gastro duodenal polyposis and non-erosive gastritis, colonoscopy shows recto colic polyps (Figure-2).

FIGURE 2: Endoscopic view of multiple scattered polyps in Rectum, Sigmoid & descending Colons

Conservative treatment was given and patient conditions improved. So, discharge was given with advice of periodic follow up. Frequent medical examination and testing is essential to allow early detection of polyps and cancer.

3 | DISCUSSION

Peutz-Jeghers syndrome is a more than rare inherited disorder with particular features of mucocutaneous pigmentation macules around mouth and cheek, multiple polyps in the gastrointestinal tract, and recognizable family history. Conditions with either mucocutaneous pigmentation or gastrointestinal polyps are defined as incomplete PJS, which usually presents only typical pigmentation or enterorrhagia or symptoms of intussusception. In 1998, Jenne and Hemminki colonized PJS-related gene at the proximity of D19S886190kb, and named it STK11. Papp J found, in 21 cases from 13 families in Hungary, that all patients had STK11 mutation [10].

In treating PJS, the treatment for mucocutaneous pigmentation were usually considered as unnecessary; however, if it is necessary, laser therapy can be a choice. For polyps’ treatment, opinions diverge. Some consider these polyps as precursors of cancer, which should be paid considerable attention or removed by surgery [11]. However, others found out that canceration of these polyps was rare, and periodic follow-up is enough. Enteric intussusception in adults is very rare and more than 90% of cases are associated with a pathological leading point. In the study conducted by Chiang and Lin, nearly 18% of incidences of hamartomas caused enteric intussusception. Ileo-ileal intussusception was the most common type. About 90% of cases presented with abdominal pain, while 40% exhibited signs of proximal small bowel obstruction. Rare presentations were diarrhoea, bleeding and anaemia. Abdominal CT scan was shown to be the most effective diagnostic instrument [12]. These polyps are 1 mm to 4 cm in diameter, mostly seen in jejunum and small bowel > colon. They can also occur at the nose, bronchi, renal pelvis, and biliary tree. Pigmentation is seen in perioral, buccal mucosa, digits of hands and feet and perianal and genital region. These patients mostly present in 3rd decade with abdominal pain due to intussusception. Other less common presentations: Symptoms from obstruction of large polyps, anemia, hematochezia, hematemesis, biliary obstruction, and gastric outlet obstruction. They are at an increased risk of both GI and extra GI malignancy such as that of pancreas, breast, thyroid, lungs, gallbladder, ovary, and testes [13].

4 | CONCLUSION

Peutz-Jeghers syndrome patients should examined periodically to detect any form of cancer. Risk and complications related with polyps should also considered in PJS.

Conflict of Interests:

The authors declare no conflict of interests. No financial involvement in the preparation of this manuscript.

REFERENCES


