

A Child with Peutz-Jeghers Syndrome Presenting in Emergency as Acute Intestinal - Obstruction and Malena

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ABSTRACT

Introduction: Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant hereditary disorder affecting males and females equally, presenting as mucocutaneous hyperpigmentation and intestinal polyposis and sometimes with complications, as acute intestinal obstruction.

Case report: We are presenting here a rare case of Peutz-Jeghers Syndrome in a eight year old female, who presented in emergency with features of acute intestinal obstruction. On examination, mucocutaneous pigmentations were present on and around lips, buccal mucosa and on planter aspect of feet. She was also anaemic. Bowel sounds were exaggerated. Black tarry stool on digital rectal examination. Intra-op finding on laparotomy was jejunio-ileal intussusception, for which resection and anastomosis of the involved loop was done.

Conclusion: Hyperpigmented lesions especially in and around buccal cavity should be investigated for PJS in early age group to treat polyposis effectively and to prevent its complications and further morbidity and mortality.

Keywords: Intussusception, Malena, MDCT, Peutz-Jeghers Syndrome (PJS).

INTRODUCTION

Peutz-Jeghers syndrome (PJS) was first described by Johannes Peutz, a Dutch Paediatrician, in 1921 and later by Harold Jeghers in 1949.¹ PJS is an autosomal dominant hereditary disorder characterized by mucocutaneous hyperpigmentations and gastrointestinal polyps. Jejunum is the most common site of polyps followed by ileum, stomach, duodenum and colon. These polyps grow during the first decade of life and become symptomatic. The most frequent complications of PJS is intussusception occurring in 47% of patients and mainly in small intestine.² These polyps may lead to gastrointestinal bleeding and intestinal obstruction by their own or intussusception occasionally, as they are present since childhood. There is a lifetime higher risk of gastrointestinal malignancy, exceeding 50% and also risk of breast, ovarian, testicular and pancreatic cancers.³ Here we present a child of Peutz-Jeghers Syndrome, with small bowel obstruction and malena.

CASE REPORT

An eight year old female child presented in emergency with complaints of recurrent colicky abdominal pain with passage of dark red blood in stool since last seven days on and off, with two to three episodes of vomiting containing greenish fluid. On general examination, she was pale and dehydrated. Around 20-30 black-bluish hyperpigmented spots were present on her lips near the vermilion border, perioral skin, nostrils, columella, nasal bridge, mucosa of buccal cavity and planter aspect of feet, especially on toes and digits and their size ranging from 1mm to 5mm transversely (Figures-1,2).

Tachycardia and tachypnoea was present. There was a palpable, rigid, vague, tender lump of size approx. 8 x 6 x 3cm in the umbilical region, encroaching in left hypochondrium, firm in consistency. Bowel sounds were exaggerated. Digital rectal examination showed dark black color stools with mucous. Soft tissue density displacing adjacent bowel loop gases were present on plain x-ray of the abdomen.

Ultrasonography of the whole abdomen suggested a 15 x 6.5 x 5.8cm size small-bowel loop intussusception (probably, jejunio-ileal intussusception, as a single mass). She was moderately anaemic (Hb% = 7.2gm%) with total leukocyte counts = 13500 cells/mm³ and polymorphs- 88% on blood reports. She received two units of blood transfusion each, before and during exploratory laparotomy.

Resection of the affected bowel loop and jejunioileal anastomosis was performed. Multiple pedunculated and sessile polyps of size (3mm x 5mm) to (10mm x 25mm) were seen predominantly in jejunum with pregangrenous/gangrenous changes within the intussusceptum and leading polyp was seen after laying open the specimen (Figures-3,4).

Histopathological examination (HPE) showed proliferating mucinous glands with smooth muscle fibres arranged in definite pattern. No nuclear atypia was seen. Few glands were dilated. Ultimately hamartomatous polyps were diagnosed with the above findings. A diagnosis of Peutz-Jegher syndrome was made based upon the above clinical and histopathological findings.

DISCUSSION

PJS has a reported incidence of 1:8300 to 1:128000 in general population, with an almost equal incidence in both sexes. The estimated approximate prevalence of PJS has been reported as 1:100000 in literature.¹

PJS manifests as mucocutaneous hyperpigmentations and hamartomatous polyps in the gastrointestinal tract. The predicted sites for hyperpigmentations are peri and intraoral regions; predominantly on lips, with variation in size, numbers and color.¹ The least common sites of melanin pigmentations are rectum, feet, vulva and conjunctiva. The melanin pigment distribution in perioral area may be present at birth or appear in childhood. Laugier-Hunziker syndrome, which is characterized by melanotic pigmentation of lips and buccal cavity but without

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gastrointestinal polyposis should be differentiated from PJS.⁴ They appear like spots with change in color.³ PJS is caused by mutation of the STK11/LKB1 (serine / threonine kinase11) tumor suppressor gene mapped on 19p13.3.⁵

The gastrointestinal hamartomatous polyps are the next most important features of PJS. Jejunum is the most common site followed by ileum and duodenum in decreasing order. Utsunomiya et al has noted a distribution of polyps in the gastrointestinal track as stomach (48.60%), small intestine (64%), colon (63.20%) and rectum (32%), in a study of PJS patients.² These polyps may also be present at extra-intestinal sites, as in renal system, nose, lungs and gall bladder.⁶

Approximately 35% patients of PJS are diagnosed in the first decade of life and around 30% patients have no previous positive family history, as seen in our case report. It seems to be due to spontaneous genetic mutations.⁴ In our case report, symptomatic presentation was at 8 years of age. The patients of PJS often present with on and off pain abdomen (23%) and features of intestinal obstruction (43%) and malena, due to ulceration / persistent intermittent bleeding of large polyps, resulting in anaemia.

The criteria of diagnosing PJS includes⁷

- i) Presence of at least two polyps,
- ii) one polyp and pigmented mucocutaneous lesions,
- iii) one polyp and positive family history

The patients of PJS have tendency for development of both gastrointestinal and non-gastrointestinal malignancies. The

non-gastrointestinal sites are thyroid, lungs, breast, uterus, cervix, ovary and testis.⁸ In a large study on 419 individuals, ninety-six cancers individuals were found among PJS and 297 patients documented as STK11/LKB1 mutations.

An exploratory laparotomy is required in cases of intestinal obstruction and persistent / intermittent bleeding, if not controlled non-operatively, since decades for PJS. It may be followed by resection and anastomosis of the involved section of bowel. The extensive resection of bowel may result in short bowel syndrome and malnutrition. Furthermore intra-operative enteroscopy was added to evaluate and treat small intestine pathologies with each laparotomy in PJS patients. In our case report, an emergency exploration of abdomen was done for acute presentation because of persistent intussusception and malena followed by resection and anastomosis.

MDCT (Multi Detector Computerised Tomography) enterography is a recent noninvasive adjunct to capsule endoscopy in excluding strictures, and clarifying doubtful findings. It could provide information of polyps and complicated intussusception in Peutz-Jegher syndrome patients.⁹ Videoendoscopy may be aimed for excision of polyps in PJS, so that complications like intussusception and resection of bowel is prevented.

The antiproliferative agents like sirolimus and a non-steroidal anti-inflammatory drug (strong activity of COX-2 in polyps) is under trial for prevention of PJS. The family surveillance and genetic counseling plays a major role in management of suspected cases of Peutz-Jegher Syndrome and therefore



Figure-1: Mucocutaneous hyperpigmentations on and around lips and buccal mucosa.



Figure-3: Pregangrenous leading polyp causing intussusception.



Figure-2: Hyperpigmentations on planter aspect of toes and digits.

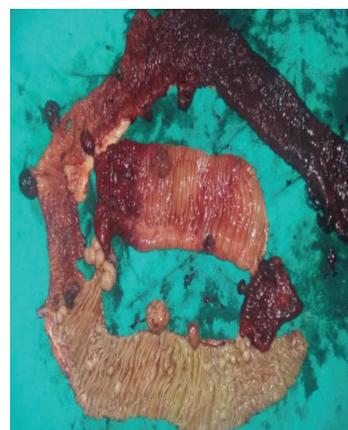


Figure-4: Small bowel specimen showing pregangrenous /gangrenous changes with multiple polyps.

molecular analysis of the polyps is recommended.¹⁰

CONCLUSION

The clinical course of the PJS is unpredictable; so patients presenting with incomprehensible pigmented lesions, especially in and around buccal cavity should be investigated for PJS because of its low incidence and rarity. There is no significant optimal screening programme recommended for Peutz-Jeghers Syndrome, hence these patients should undergo for lifetime follow up with regular clinical examinations, endoscopy, radiological investigations and laboratory tests, along with genetic counseling; as they are at high risk for development of cancer.

Routine screening with endoscopy in early age group is recommended to treat the polyposis effectively and to prevent complications like obstruction due to intussusception; otherwise it may require bowel resection unnecessarily, which can lead to further morbidity and mortality.

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