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Case Report

Peutz-Jeghers syndrome associated with eventration of left hemidiaphragm in a child: A case report

Jiledar Rawat, Sudhir Singh,*

Department of Pediatric Surgery, King George's Medical University, Lucknow, UP, India

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ABSTRACT

Background: Peutz–Jeghers syndrome (PJS) is a rare genetic disorder. PJS with ileo-ileal intussusception and left-sided eventration of the diaphragm is an extremely rare presentation and not been reported so far.

Case Presentation: An 8-year-old child of PJS presented with acute intestinal obstruction. Examination and investigations of the child revealed intussusception with left side elevated hemidiaphragm. Surgical correction of both conditions was done.

Conclusion: PJS with intestinal obstruction due to intussusception needs surgical correction. Sometimes these cases may present with other associated surgical conditions that too need simultaneous surgical correction.

Keywords: Intussusception, Eventration of the diaphragm, Intestinal polyps, Peutz-Jeghers syndrome (PJS)

INTRODUCTION

Peutz-Jeghers syndrome (PJS) an autosomal dominant hereditary genetic disorder characterized by the development of benign hamartomatous polyps in the gastrointestinal tract and mucocutaneous pigmentation.[1] It has an incidence of approximately 1 in 25,000 to 300,000 births.[1] It may develop various complications such as gastrointestinal bleeding, intussusception, and malignant transformation.[2] Occasionally other surgical maladies can coexist. We are presenting a case of PJP with intestinal obstruction due to intussusception and left side eventration of the diaphragm.

CASE REPORT

An 8-year-old boy referred to us with abdominal pain, distention, and bilious vomiting for the last 3 days. He also had intermittent colicky pain in the epigastrium and per-rectal bleeding for the last one year. The bleeding was occasional with fresh blood; there was no history of melena. The child had poor nutritional status and was not thriving well in addition to the history of recurrent respiratory tract infections since birth. The child had been treated by local practitioners, but no diagnosis was labeled. On examination, pulse was 130/min with moderate dehydration and pallor. A palpable abdominal mass was found in the peri-umbilical area. There were hyperpigmentation spots on the mucocutaneous junction of the oral cavity. Digital rectal examination revealed an empty rectum with blood mixed mucus; multiple polyps were present approximately 5 cm from the anal verge.

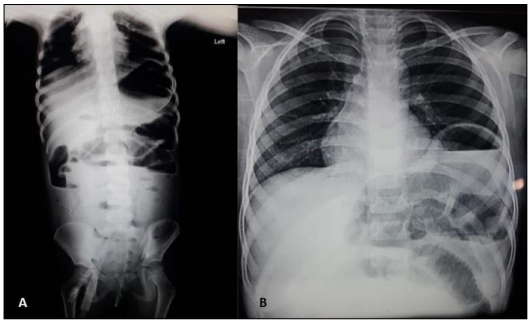

Figure 1A, 1B: Radiograph of abdomen and chest, air-fluid levels in the abdomen and elevated left hemidiaphragm.

Resuscitation of the child was performed with intravenous fluid, antibiotics, and a nasogastric tube

Correspondence*: Sudhir Singh, Associate Professor, Department of Pediatric Surgery, King George's Medical University, Lucknow, UP, India
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inserted. Hematological and biochemical investigations were unremarkable except for hemoglobin which was 7.5 gram/deciliter. Plain radiographs of the abdomen and chest suggest multiple air-fluid levels in the upper abdomen and elevated left hemidiaphragm (Fig. 1A. 1B). Ultrasonography (USG) of the abdomen showed distended loops of bowel in the upper abdomen with an ileo-ileal intussusception. On Contrast-enhanced computer tomography, besides the findings of USG, multiple intestinal polyps and eventration of left diaphragms found. (Fig. 2A, 2B)

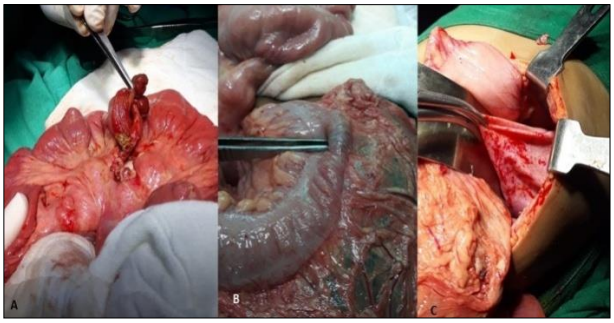

Figure 2: Intussusception (A) with colonic polyps (B) with left side eventration of the diaphragm (C).

On exploratory laparotomy, an ileo-ileal intussusception with multiple ileal and colonic polyps were found with left-sided eventration of diaphragm (Fig. 2A, 2B and 2C). On reduction of intussusception, the gangrenous ileal segment found, so resection of gangrenous segments and primary anastomosis were done, and resection of three big polyps was done by performing an enterotomy in the small bowel. Repair of eventration of the left diaphragm was also performed by excising the lax hemidiaphragm giving the doubt of diaphragmatic hernia with sac (Fig. 3A, 3B). Postoperatively, the patient improved uneventfully.

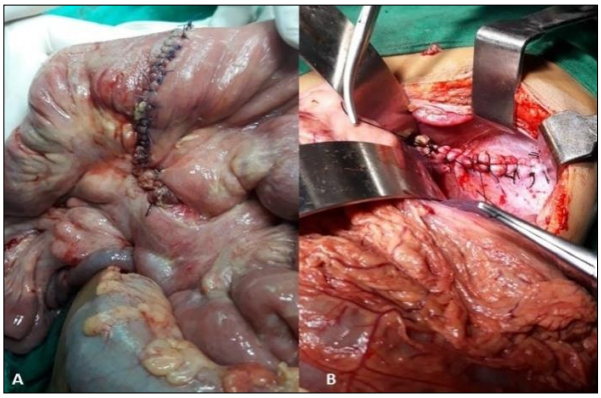

Figure 3: Primary anastomosis of the ileum (A), and repair of eventration of the left hemidiaphragm (B).

Histology was consistent with hamartomatous polyps and excised diaphragmatic sac confirmed the diagnosis of eventration. The diagnosis of PJS was confirmed by the identification of a heterozygous pathogenic variant in STK11 by molecular genetic testing. The child is on regular follow up and is doing fine for the last 2 years.

DISCUSSION

PJS is an autosomal dominant disease characterized by hamartomatous polyposis throughout the gastrointestinal tract, as well as mucocutaneous pigmentations, mostly on the lips, oral, and gingival mucosae.[1] This is due to a germline mutation in the STK11 (LKB1) gene.[3] The index case was also confirmed as PJS on genetic testing. Patients with PJS often present with a history of intermittent abdominal pain and per-rectal bleeding due to intraluminal polyps. There is a high frequency of hypo-proteinemia and anemia, in our case, there was a history of intermittent abdominal pain and bleeding per rectum for the last one year. Few patients develop intestinal obstruction due to intussusceptions, as also seen in our case.[4, 5] The mechanism of intussusception is not fully understood, but polyps are believed to act as pathological lead points.

Sometimes the intussusceptions are spontaneously reduced, however surgical intervention is required if the intestinal obstruction is developed. Though our patient had recurrent respiratory infections, eventration of the diaphragm was not detected earlier and found during the radiological evaluation of the child. PJS and eventration of the diaphragm in a child are extremely rare findings and not been previously reported to the best of authors' knowledge. The eventration of hemidiaphragm is occasionally very thin which may become indistinguishable from a sac of congenital diaphragmatic hernia. Plication of the thin diaphragm is a preferred method with a series of non-absorbable sutures without injury to the phrenic nerve.[6] In our case, repair of the eventration of left hemidiaphragm was done by excising the sac, as the sac was quite lax and very thin, giving us the doubt of a diaphragmatic hernia with sac.

To conclude, PJS associated with the eventration of hemidiaphragm is an extremely rare co-occurrence. Any history of repeated chest infections should be investigated to rule out correctable causes of recurrent respiratory tract infections.

Consent to Publication: Author(s) declared taking informed written consent for the publication of clinical photographs/material (if any used), from the legal guardian of the patient with an understanding that every effort will be made to conceal the identity of the patient, however it cannot be guaranteed.

Authors Contribution: Author(s) declared to fulfill authorship criteria as devised by ICMJE and approved the final version. Authorship declaration form, submitted by the author(s), is available with the editorial office.

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