



REVIEW OF LITERATURE AND CASE REPORT OF A PATIENT WITH RECURRENT MULTIPLE INTUSSUSCEPTION OF SMALL INTESTINE DUE TO PEUTZ JEGHER'S POLYPS

General Surgery

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ABSTRACT

Intussusception occurs when one portion of gut enters into immediately adjacent segment, almost invariably it is proximal into distal. It is associated with an associated lead point such as polyps, meckel's diverticulum, duplication or Hirschsprung's disease. Ileocolic is most common variety. It may lead to ischemic bowel segment. Multiple polyps in intestine may cause multiple intussusceptions. In children reduction of intussusception can be performed initially. In adults exploration is main choice of treatment

KEYWORDS

INTRODUCTION

Peutz-Jegher syndrome is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps, mucocutaneous pigmentation and an increased risk of malignancy with an incidence of one in 12 to 30,000 live birth. The main associated risks are acute small bowel obstruction and associated GI malignancy in adult life.

WHAT IS PEUTZ-JEGHER'S SYNDROME?

Peutz-Jegher's syndrome is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps in association with a distinct pattern of skin and mucosal macular melanin deposition and wide range of cancers

Epidemiology

- Prevalence 1 case per 60,000 people to 1 case per 300,000 people
- Males and females equally affected
- Mostly hereditary

AGE RELATED DEMOGRAPHICS

- Polyps commonly presents in adolescence and early adulthood
- One third individuals experience symptoms during the first 10 years of life
- Approximately 50% of individuals have experienced some symptoms by age 20 years

GENETICS

- The cause of (PJS) in most cases (> 90%) appears to be a germline mutation of the STK11/LKB1 tumor suppressor gene, located on chromosome 19p13
- somatic inactivation of the unaffected allele of STK11
- This mutation may occur sporadically and gets transmitted in autosomal dominant pattern

CLINICAL FEATURES

- Tan, dark brown, bluish black flat patches in the form of 1-5mm size freckles are seen around mouth, lips, gums, inner lining of mouth, eyes, hands, feet, anus and genitals
- GIT polyps occur late and causes bleeding and abdominal pain.
- In small intestine polyps cause INTUSSUSCEPTION, OBSTRUCTION
- Colicky abdominal pain
- Gastrointestinal and non-gastrointestinal cancers.

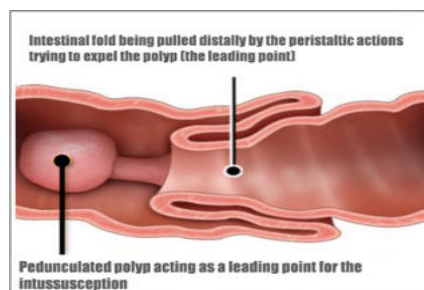
DIAGNOSIS-based on

Clinical examination, GI symptoms, family history, past history of surgery for intussusception, UGIscopy and COLONOSCOPY for polyps, Histopathology and Radiological Findings(USG & CECT (A+P)

INTUSSUSCEPTION

- **DEFINITION:-** INTUSSUSCEPTION occurs when one portion of the gut invaginates into an immediately adjacent segment; almost invariably, it is the proximal to the distal
- **Causes-** GI malignancies (colorectal carcinoma, metastasis, Benign neoplasm (intestinal polyps, lipoma, GIST), Congenital (Meckel's diverticulum, duplication cyst)

Multiple polyps in Peutz-Jegher syndrome may act as lead point for intussusception which may lead to bowel obstruction and ischemia and even perforation



POLYPS IN PEUTZ-JEGHER'S SYNDROME According to UTSUNOMIYA ET AL STUDY

- Small intestine - 64% of patients
- Colon - 63.2% of patients
- Stomach - 48.6% of patients
- Rectum - 32% of patients

Extra-intestinal polyps

- Nasal polyps
- Gall bladder polyps
- Ureteric polyps
- Respiratory tract polyps

GASTROINTESTINAL COMPLICATIONS According to UTSUNOMIYA ET AL STUDY

- Obstruction (INTUSSUSCEPTION more commonly localises small bowel) - 42.8%
- Abdominal pain - 23%
- Bleeding - 13.5%
- Gastric outlet obstruction

Risk of malignancy

- 15-fold increased risk of developing intestinal cancer
- 48% develop and die from cancer by age of 57 years

- 100 times increased risk of pancreatic carcinoma compared with normal population
- GB/ Bile Duct, Thyroid, Lung carcinomas
- Breast, Lung, Ovarian, Fallopian tube carcinomas in females
- Testicular carcinomas in males

CLINICAL PRESENTATION AND DIAGNOSTIC CRITERIA

- During the first 3 decades of life, anemia, rectal bleeding, abdominal pain, obstruction, and/or intussusception are common complications in patients with Peutz-Jeghers syndrome..
- As proposed by the Johns Hopkins Registry Diagnostic criteria include:

Histopathologically verified hamartomatous polyps with at least 2 of the following:

- Small-bowel location of polyposis
- Mucocutaneous melanotic pigmentation
- Family history of Peutz-Jeghers syndrome.

A CASE REPORT OF RECURRENT MULTIPLE INTUSSUSCEPTION IN PEUTZ-JEGHERS SYNDROME

CASE REPORT

PATIENT DETAILS

- A 19 year old male unmarried student belonging to Bhavnagar, Gujarat
- Muslim by religion
- And student by occupation
- Belonging to lower middle socioeconomic class
- Presented In the emergency on 26/12/24 with chief complains of colicky abdominal pain with vomiting for last 2-3 days

HISTORY

- **PRESENT ILLNESS** - Colicky Abdominal pain started 5 days ago and increased over past 5 days vomiting started 2-3 days back, greenish in colour, 2-3 episodes /day patient was taken to local hospital in Bhavnagar and was referred to CIVIL HOSPITAL AHMEDABAD.
- **past history** - Patient presented with similar complaints in the past. H/O emergency exploratory laparotomy with resection of INTUSSUSCEPTION (with polyps acting as lead point), containing gangrenous segment of small bowel with jejunio-ileal end to end anastomosis under GA on oct 2023

INVESTIGATIONS OF PREVIOUS SURGERY

USG

- Approx 34*48 mm sized target like lesion (bowel within bowel) noted from left lumbar region to right iliac fossa, length of involved segment is approx >8 cm, ileum as both intususcepiens and intususceptum s/o ileoileal INTUSSUSCEPTION
- Another 29*39 mm sized target like lesion noted in right lumbar fossa, involved segment approx 6cm s/o jejunojejunal INTUSSUSCEPTION

BIOPSY

- Specimen shows gross INTUSSUSCEPTION with gangrenous loop of intestine
- History iron deficiency anemia in may 2024
- personal history - non vegetarian diet
- no h/o addiction
- bladder bowel habit normal
- Family history - Patients father and grandfather died at around 40-50 years of age allegedly due to cardiac causes.
- (Note: Patient's mother has no signs or symptoms suggestive of PJS)

GENERAL EXAMINATION

- On general examination
- Pulse - 84/min Blood pressure - 110/74
- RR - 16/min SPO2 - 98% on room air
- RS- bilateral air entry was positive and equal
- CVS - S1 S2 heard and no abnormal sounds were heard on auscultation
- CNS - conscious and oriented
- P/A - distended diffuse tenderness on deep palpation
- P/R - ON DRE - no ballooning, normal anal tone, no fissure/fistula/mass
- ON PROCTOSCOPY- no visible polyps

- Pigmentation - Bluish black mucosal of about 2-3mm present over inner surface of both cheeks, Tan brown freckles present on palmer aspect of distal Phalynx of ring and middle fingers of right hand and over bilateral gluteal region. Brown freckles present scattered over sole of B/L foot.
- No clubbing lymphadenopathy jaundice or pallor present

CLINICAL IMAGES SHOWING PIGMENTATION



Left inner cheek



Tan brown freckles present over distal phalanx of right hand



Brownish black freckles present over gluteal region

INVESTIGATIONS

- Hemogram, LFT, RFT within normal range
- hiv hbsag non reactive
- Chest and abdominal x ray - N A D usg abdomen pelvis - multiple small bowel intussusception

ULTRASOUND ABDOMEN-PELVIS (26/12/24)

1. Approximately 28*37 mm sized target like lesion (bowel within bowel appearance) is noted in infra umbilical region. Length - approx 45mm. Part of small bowel loops (ileum) acts as intususcepiens. Part of small bowel loops (ileum) act as intususceptum. Bowel loops shows preserved vascularity & normal peristalsis.
2. Another approx 33x42 mm sized target like lesion is noted in right paraumbilical region. Length- 45mm. Part of small bowel loops (ileum) - intususcepiens. (ileum) - intususceptum. Bowel loops shows preserved vascularity & normal peristalsis.
3. Another approx 19x22 mm sized target like lesion noted in right illiac fossa. Length - approx 18mm. (Ileum) - intususcepiens. Ileum - intususceptum. Bowel loops shows preserved vascularity & normal peristalsis.
4. Another approx 32x46 mm sized target like lesion is noted in left lumbar region. Length - approx 45mm.

Part of small bowel loops (ileum) acts as intussuscepiens. Part of small bowel loops (ileum) act as intussusceptum..

Findings are in favour of multiple small bowel intussusception

Liver appears enlarge in size (17.6 cm) & normal in echotexture.

GB appears mild distended with few tiny calculi within average size measuring 3-4mm

Spleen appears enlarge (13.5cm) in size

CECT (A+P)

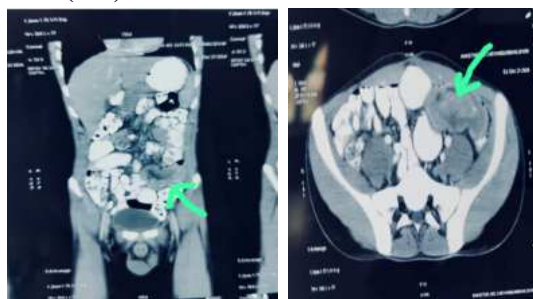
21/12/2024 (Sahyog-CHA)

- Multiple polyps seen involving jejunal and ileal lumen largest measuring 20*13 mm
- Multiple (at least 3 jejunal ileal and jejuno jejunal INTUSSUSCEPTION seen in left lumbar, centre left abdomen and right peri umbilical parasagittal abdomen noted measuring approx 70*80*40mm in length
- Resultant mild dilated (30 mm) proximal jejunal loop
- S/o small bowel obstruction due to multiple INTUSSUSCEPTIONS

25/12/2024 (outside) (note-Patient had taken Lama and got CECT A+P done outside)

- Enhancing polypoid lesion in proximal jejunal loop and ileal loop the large polyp measures 10*11 mm.
- Enhancing polypoid lesion of size 9*12mm near terminal ileum
- Bowel into bowel segment giving target appearance in umbilical region and right para umbilical region the involved segment contains polypoid enhancing focus intussusception Presence of multiple polyps may represent associated syndrome peutz jeghers syndrome and is to be ruled out

CT SCAN (A+P) IMAGES



MANAGEMENT

- On day of emergency based on clinical exam and investigations decision was taken to operate the patient on elective basis
- Patient was kept NBM
- IV fluids with antibiotics started and was closely monitored
- Patient was posted for diagnostic laparoscopy sos exploratory laparotomy on 27/12/2024 electively
- Diagnostic laparoscopy done which was converted to exploratory laparotomy as multiple polyps with multiple Intussusceptions were seen

INTRAOP FINDINGS

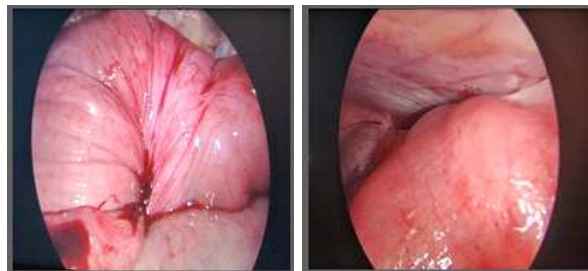
- Multiple Intussusceptions (4 in number) present without any adverse changes
- reduction of intussusceptions done and polyps as lead points were noted
- No bowel gangrenous or ischemic change
- 10 polyps were palpable 4 of which were lead points for intussusception
- approx 60 cm proximal jejunal segment containing 4 polyps was the shortest segment containing most number of polyps, 2 of which acted as lead points.
- Intra-op COLONOSCOPY findings
- Multiple colonic polyps were noted in sigmoid colon and ascending colon

Multiple Intussusceptions (4 in number) present with out any adverse changes

- reduction of intussusceptions done and polyps as lead points were noted
- Bowel was healthy without gangrenous or ischemic changes
- 10 polyps palpable 4 of which were lead points for intussusception
- Polyps were scattered inside all over the small intestine

- approx 60 cm proximal jejunal segment containing most number of polyps (4 polyps) of sizes varying from 4-6cm

DIAGNOSTIC LAPAROSCOPY IMAGES



INTUSSUSCEPTION

POLYP EXTERNAL VIEW

Multiple Intussusceptions (4 in number) present with out any adverse changes

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COLONOSCOPY findings

- Multiple polyps were noted in rectum and proximal sigmoid colon

CONSIDERATION FOR SURGICAL INTERVENTION

- Intestinal polyps <2cm can cause GI bleed and act as lead point for intussusception. These can be managed by conservatively under .
- Intestinal polyps between 2-5cm cause GI bleed, act as lead point for intussusception and may lead to Intestinal obstruction. These can be managed by endoscopic snare polypectomy
- Polyps >5cm can not only cause above mentioned complications but may also lead to intraluminal obstruction which is why they need to be excised surgically.
- Intraoperatively we found multiple polyps which were measured with a sterilised scale. The smallest measuring 1cm and largest polyp measuring about 5-6cm.

INTRA-OP VISUALS OF REDUCTION



Intra-op visuals

External view of the polyp



Internal view single polyp

PROCEDURES DONE

- Careful reduction of INTUSSUSCEPTIONS done
- Approx 60cm segment of small bowel (jejunum) containing major number of polyps (4 in number) resected two of which were lead points for INTUSSUSCEPTIONS
- INTUSSUSCEPTION created for delivering a polyp in proximal

- small bowel and the polyp ligated and resected
- INTUSSUSCEPTION created was reduced and primary anastomosis done leaving the remaining inaccessible polyps for endoscopic resection using snare
- Intra-op COLONOSCOPY done and colonoscopic biopsy taken from the colonic polyps

INTRA-OP COLONOSCOPY VISUALS



Rectal polyp in colonoscopy

HISTOPATHOLOGY

SPECIMEN 1) JEJUNUM

show histology of Peutz Jegher's polyp with focal changes of enteritis cystica profunda

SPECIMEN 2) JEJUNUM SECTION (POLYP)

show histology of Peutz Jegher's polyp with focal changes of enteritis cystica profunda

SPECIMEN 3) JEJUNUM BAND TISSUE

- Sections shows hyalinized fibrocollagenous tissue with vascular granulation tissue and scattered non specific inflammation

SPECIMEN 4) RECTAL POLYP (BIOPSY)

suggestive of hamartomatous polyp

- CONCLUSION- Peutz jegher's polyps with focal changes of enteritis cystica profunda

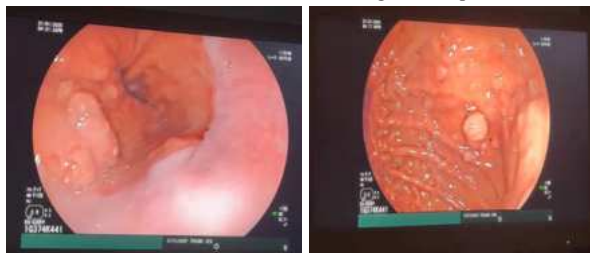
POST-OP SEQUELAE

- Was uneventful without any major complications
- Patient was kept NBM for 7 days
- RT removed and oral fluids started on post-op day 7 and soft diet on POD-8.
- Abdominal drains removed on post-op day 11
- Patient discharged on post-op day 13

FOLLOW UP

- Patient has been instructed for lifetime follow up
- Planned for UGIs copy
- Planned endoscopic snare polypectomy
- Planned for regular screening by colonoscopy ugi copy, CECT (A+P) and serial serological tests to detect malignancies as early as possible for management

SCREENING UGISCOPY VISUALS during follow up



MONITORING OF A PEUTZ JEGHER'S CASE

- Life time monitoring is required in a case of peutz jegher's syndrome for early detection of carcinomas
- A complete small bowel evaluation with EGD and capsule endoscopy should be done biannually, starting around age 8 years or younger if symptoms are present
- Colonoscopy should be done every 2 to 3 years, starting in the late teenage years.
- Breast surveillance (females):
- Gynaecological surveillance with pelvic exam and Pap smear

should be done every 3 years beginning at 25 years of age.

PATIENT INSTRUCTIONS AND COUNSELLING

- Patient should be counselled adequately about the intestinal complications and future risks of carcinomas and inheritance to future generations
- Patients should be told that they will require routine periodic (semiannual, annual, and biannual) examinations for gastrointestinal and extraintestinal cancer surveillance
- Women with PJS should begin self-breast exams at 18 years of age. They may benefit from involvement in disease-specific patient advocacy groups
- An online support group has also been established for PJS
- <https://www.chop.edu/health-resources/peutz-jeghers-syndrome-and-juvenile-polyposis-syndrome-online-support-group>.