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Case report

A RARE CASE OF PEUTZ-JEGHERS SYNDROME PRESENTING WITH MELENA

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ABSTRACT

Peutz-Jeghers Syndrome (PJS) is a rare autosomal dominant disorder characterized by hamartomatous polyps throughout the gastrointestinal tract and distinctive mucocutaneous pigmentation. We present a case of PJS in a 17-year-old male who presented with melena, which ledto the discovery of multiple hamartomatous polyps in the small intestine, colon, and stomach. Despite the rarityof this syndrome, prompt recognition of its clinical manifestations, including gastrointestinal bleeding, is crucial for early diagnosis and management. This case underscores the importance of consideringPJS in the differential diagnosis of patients presenting with melena, as timely intervention can prevent serious complications such as bowel ischemia and malignant transformation. Additionally,it highlightsthe significance of comprehensive surveillance and screening protocols for individuals with PJS to detect and manage associated malignancies and gastrointestinal complications effectively.

KeyWords: Peutz-JeghersSyndrome,melena,Hamartomatouspolyps,Mucocutaneouspigmentations

INTRODUCTION

Peutz-Jeghers syndrome is a rare autosomal dominant disorder characterized by hamartomatous polyposis of the gastrointestinal tract, melanin hyperpigmentation of the skin and mucous membranes, and an increased risk for intestinal and extraintestinal malignancies. The estimated incidence of PJS ranges from 1:50,000 to1:200,000 births with no gender or racial predilection. The most common and worrisome manifestationin children andadolescentsistheoccurrenceof polyp induced small bowel intussusception, which presentsas melena. It could represent a serious surgical emergency and be life-threatening which requiresrepeated laparotomies. Other important presentations include bowel obstruction and anemia due to the polyps.

CASEREPORT

A17-year-oldmalepatientcamewithchiefcomplaintsof melaenaforthelast2daysandpainabdomenwithvomiting for thelast3days.Onexamination,multiplehyperpigmented macules were noted on the lips and inner aspect of cheeks. Per abdominal and per rectal examination was under normal limits. The hemogram revealed mild anemia (Hb-12gm%). Other biochemical lab investigations were under normal limits. B-mode and Doppler scan was done using GE-M6 Versana BalanceUSGmachine.Contrastenhanced CTofthe abdomen was done using 128-slice SIEMENS machine inour hospital. On Ultrasound, telescoping of jejunal bowel loop into jejunal bowel loop was noted in the left hypochondrium and telescoping of ileal loop into caecum was noted in the right ileac fossa suggesting multiple intussusceptions. Multiple well-defined hypo to isoechoic lesions were noted arising from the wall of the bowel loops acting as lead points for the intussusceptions. On contrast enhanced Computed Tomography, multiple well defined enhancing lesions arising from the bowel wall were noted in stomach, duodenum, jejunum, ileal loops and the ascending colon. Other findings include bowel within bowel appearance at the ileocecal junction in the right iliac fossa and jejunal bowel loops within jejunal bowel loops in left hypochondrium signifying the intussusceptions. Laparotomy was done for correction of intussusception followed by polypectomy and it was sent for biopsy. On biopsy, hamartomatousnatureofthepolyp was confirmed. The imaging findings were correlatedwith colonoscopy and intraoperative findings.

DISCUSSION

The Peutz-Jeghers Syndrome (PJS) is an autosomal dominant neoplastic syndrome caused by a germline mutation in the STK11 (LKB1) gene with hamartomatous polyps throughout the GI tract, distinctive mucocutaneous pigmentations,andan increasedcancerriskover aperson's lifetime. Multiple hamartomatous polyps are seen, most commonlyinvolvingthe smallintestine(predominantlythe jejunum), but also colon and stomach. Theycan also occur in extraintestinal sites such as the kidney, ureter, gallbladder and lungs.Thecommon presentation is usually relatedtogastrointestinalsystem,likeintussusception,

obstruction or melena. Most common and grave manifestation being recurrent intussusceptions which may cause multiple laparotomies and complications like short bowel syndrome. Rectal bleedingis seen asa complication of hamartomatous polyp. Both small and large bowel polyps tendtobe pedunculatedandstomach polyps tendto be sessile. The large polyp size and pedunculated morphology contributes to recurrent intussusceptions and obstructive symptoms, frequently requiring surgical intervention. Intussusception occurs in around 70% of PJS patients with the intestinal polyps acting as lead points. Unlike most pediatric intussusceptions which occur frequently in the ileocecal area, the PJS-related intussusceptions are usually ileo-ileal or jejuno-jejunal. Thirty percent of all the PJS mortalities are related to intussusceptions. Malignancies in the Peutz-Jegher syndrome are broadly of two types: gastrointestinal and non-gastrointestinal cancers, with regular screenings recommended to detect and prevent malignancies. Gastrointestinal cancers are the most commonmalignancies in these patients, accounting for up to two- thirdsofmalignanciesinthispopulation.These malignancies predominately include colorectal, small bowel, esophageal, and gastric cancers. Nongastrointestinal malignancies include breast, pancreatic, testis, cervix, uterus, ovary and lung. Upper and lower GI endoscopies every two years starting from age 10, along with other surveillance measures like abdominal ultrasounds and breast examinations, are advised tomanage cancer risk effectively.

Figure 1. Showinghyperpigmanted lips in a 17 year old boy.

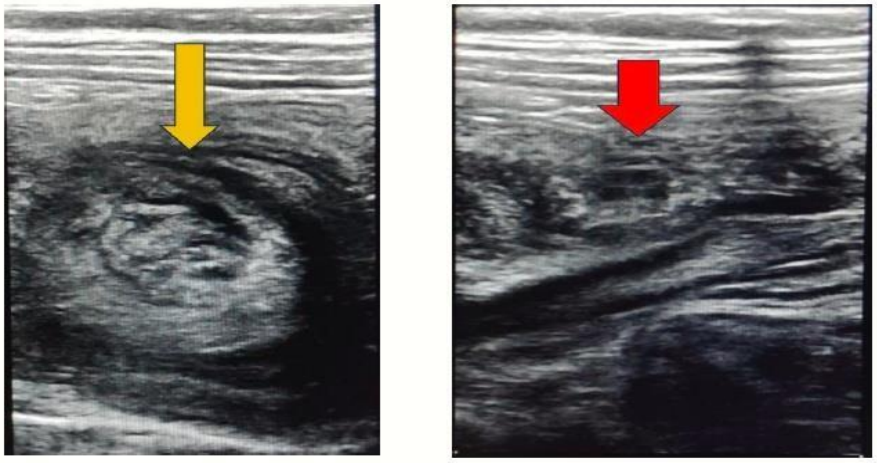

Figure 2. USG image showing a “doughnut” appearance (yellowarrow) on images obtainedtransverselytothe long axis. The hypoechoic outer area,represents the edematous bowel wall of the intussuscipiens, and,the hyperechoic centralarearepresentsmesentericfatthatisdragged into,theintussusception between theenteringandreturning limbs of,the intussusceptum. A well defined lobulated hypoechoic polyp (red arrow) ismnoted at the entry pointof the intussusceptum acting as a lead point

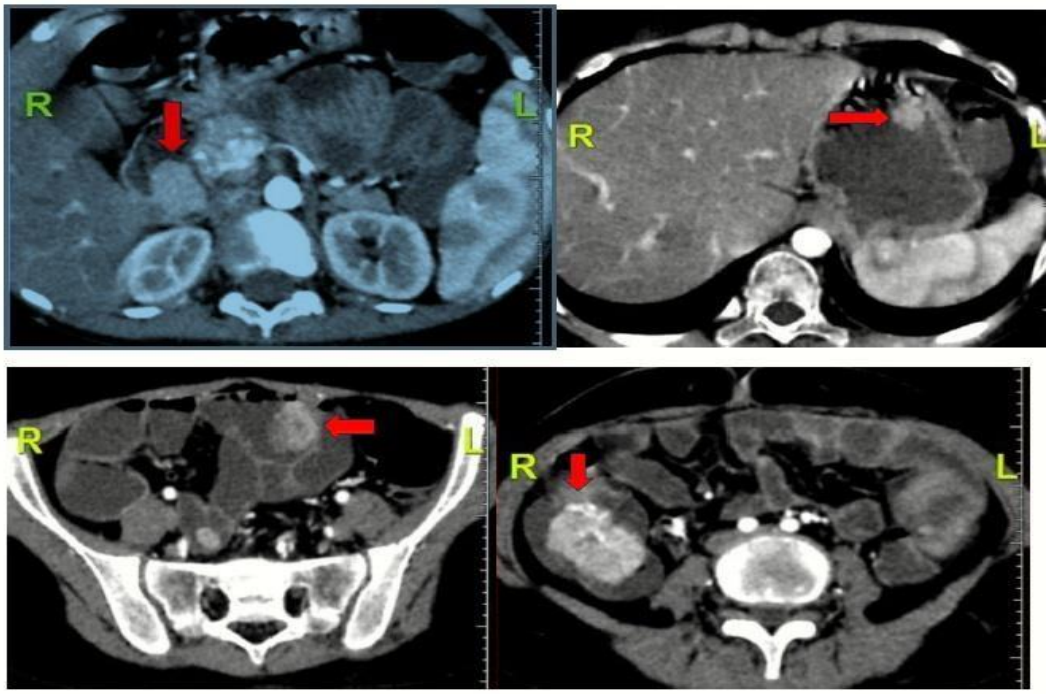

Figure3.AxialcontrastenhancedCTAbdomenimagesarrows) arising from the wall noted inn stomach, duodenum, showing-Multiplewelldefinedenhancinglesions(redjejunum, ileal loops and the ascending colon.

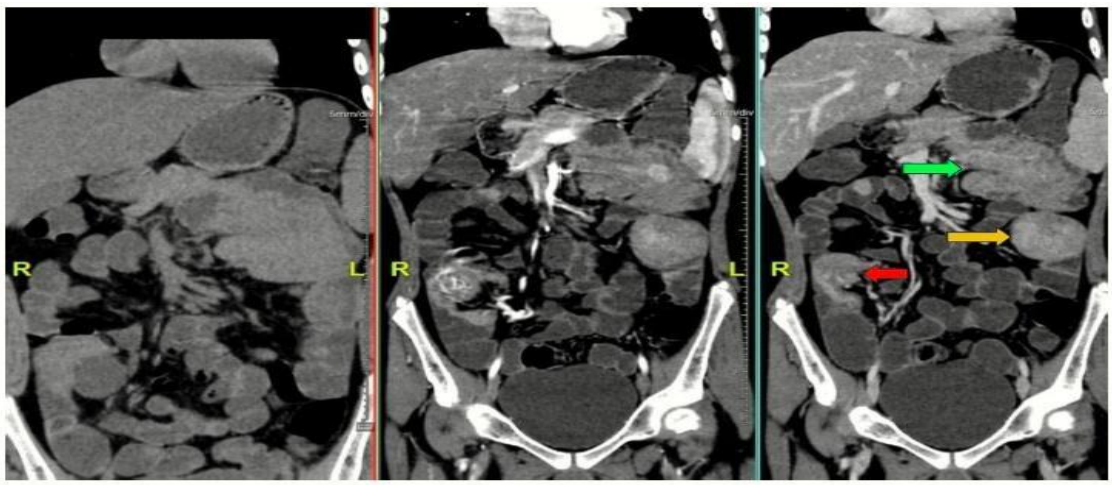

Figure 4. Coronal Plain and post contrast CT images of the abdomen showing bowel within bowel appearance at the ileoceal junction (red arrow) and at the jejuno-jejunal region in left hypochondrium (yellow arrow). Multiple enhancing polyps can be seen in the jejunal loops (green arrow).

CONCLUSION

Melaena in a case of PJS may indicate an underlying surgical emergency like intussusception, which if overlookedcanresultinbowelischaemiawithsevere consequences. Intussusception occurring at a young agecan be caused by the presence of a hamartoma polyp as a trigger. When multiple polyps are found in the gastrointestinal tract and other pathognomonic signs are found, such as hyperpigmented macular lesions on the lip and buccal mucosa, PeutzJeghers Syndrome should be suspected. Aggressive screening should be done for early detection of malignancies. Given its rarity, managingPeutz-Jeghers Syndrome necessitates a specialized approachtoaddresstheheightenedrisksofcancerand

complications associated with gastrointestinal polyps. Radiologists play a pivotal role in managing these patients by maintaining a high level of suspicion when reviewing surveillance studies of confirmed Peutz-Jeghers syndrome patients and their family members.

REFERENCES

1. Bhattacharya S, Mahapatra SR, NangaliaR, Palit A, Morrissey JR, RubanE, et al. Melaena with Peutz-Jeghers syndrome: a case report. J Med Case Rep. 2010 Feb;4:44.
2. Shetty KH, Shetty D, Pai MR. An unusual case of Peutz–Jeghers syndrome with polyposis-associated adenomatous change. Int J Case Rep Images 2020;11:101117Z01KS2020.
3. Kalliakmanis V, Perysinakis I, Koutsouvas K,Karras P, Margaris E, Angelakis C. Massive intussusception caused by a solitary Peutz– Jeghers type hamartomatous polyp. Ann R Coll Surg Engl. 2018; 100(4): e91-e93.
4. PitiakoudisM,MimidisK,TsarouchaA, KartalisG, Simopoulos K. Intussusception of the small bowel due to Peutz-Jeghers syndrome: a case report. Ann Ital Chir. 2004 Jan-Feb;75(1):75-7. PMID: 15283392.
5. Homan M, Dolenc Strazar Z, Orel R. Peutz-Jeghers syndrome. A case report. Acta Dermatovenerol Alp PannonicaAdriat.2005Mar;14(1):26-9.PMID: 15818443.
6. Fan RY, Sheng JQ. A case of Peutz-Jeghers syndrome associated with high-grade intramucosal neoplasia. Int J Clin Exp Pathol. 2015 Jun 1;8(6):7503-5. PMID: 26261661; PMCID: PMC4525995.
7. Klimkowski S,IbrahimM,IbarraRovira JJ,Elshikh M, Javadi S, Klekers AR, Abusaif AA, Moawad AW, Ali K, Elsayes KM. Peutz-Jeghers Syndrome and the Role of Imaging: Pathophysiology, Diagnosis, and Associated Cancers. Cancers(Basel).2021Oct13;13(20):5121.doi: 10.3390/cancers13205121. PMID: 34680270; PMCID: PMC8533703.
8. Mozaffar M, Sobhiyeh MR, Hasani M, Fallah M. Peutz-Jeghers syndrome without mucocutaneous pigmentation: a case report. Gastroenterol Hepatol Bed Bench. 2012 Summer;5(3):169-73. PMID: 24834220; PMCID: PMC4017480.
9. HearleN., Schumacher V., Menko F.H., Olschwang S., Boardman L.A., Gille J.J., Keller J.J.,Westerman A.M., Scott R.J., Lim W., et al. Frequency and spectrum of cancers in the Peutz-Jegherssyndrome.Clin.Cancer Res.2006;12:3209–3215. doi: 10.1158/1078-0432.CCR-06-0083
10. McGarrity T.J., Kulin H.E., Zaino R.J. Peutz-Jeghers syndrome. Am. J. Gastroenterol. 2000;95:596–604. doi: 10.1111/j.1572-0241.2000.01831.x.
11. GammonA.,JaspersonK.,KohlmannW.,Burt R.W. Hamartomatous polyposis syndromes. Best Pract. Res. Clin. Gastroenterol. 2009;23:219–231. doi: 10.1016/j.bpg.2009.02.007.
12. UtsunomiyaJ.,GochoH.,MiyanagaT.,Hamaguchi E., Kashimure A. Peutz-Jeghers syndrome: Its natural course and management. Johns Hopkins Med. J. 1975; 136:71–82.