Case Report

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A 15-year-old male with Peutz-Jeghers syndrome: a rare case report from Syria

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Introduction and importance: In addition to extra gastrointestinal hamartomatous polyps, Peutz-Jeghers syndrome (PJS), a rare but well-known hereditary disorder, generates mucocutaneous lesions that resemble certain coloured freckles and gastrointestinal symptoms. Intussusception or polyps blocking the gastrointestinal lumen are examples of PJS consequences. Additionally, the polyps may cause ongoing bleeding that causes anaemia.

Case presentation: A 15-year-old male patient with generalized stomach discomfort, frequent vomiting, and decreased appetite reported to the hospital's ambulance department. A month and a half prior, the patient underwent a surgical laparotomy for intussusception. The clinical examination revealed many pigmentations near the mouth. The specialists decided to do an urgent laparotomy on the patient, during which a 60 mm necrotic intestinal intussusception was observed. The patient had an ileoileostomy and an amputation, and a pathology test discovered numerous benign hamartomatous polyps in the sample."Putz-Jeghers Syndrome" had been determined to be the ultimate diagnosis.

Clinical discussion: It is autosomal dominant and more prevalent in children and teenagers. According to some research, 30% of diseases are passed from parents to children while 70% may result from gene mutations.

Conclusion: There is no evidence that the transformation of hamartomatous polyps led to the neoplastic tumours in these patients. It is suggested to carry out a complete screening program and detect PJS early in order to prevent gastrointestinal problems and dangerous malignancies.

Keywords: laparotomy, peutz-jeghrers, pigmentations, polyps

Introduction

A rare but well-known genetic condition called Peutz-Jeghers syndrome (PJS) causes mucocutaneous lesion that look like particular pigmented freckles and gastrointestinal symptoms in addition to additional gastrointestinal hamartomatous polyps[1]. Peutz in 1921 and Jeghers in 1944 and 1949 were the first to describe it. This disorder is equally prevalent in people who are male and female and is typically diagnosed in childhood or early adulthood[2]. PJS has a prevalence of one per 100 000people. Serine/threonine kinase 11 (STK11/LKB1)tumour suppressor gene mutations are theprimary .cause of PJS[3]. PJS complications can include intussusception or polyps obstructing the gastrointestinal lumen. The polyps may also result in continuous bleeding that results in anaemia[4]. Despite the limited malignant potential of hamartomatous polyps, those who have this condition have a higher chance of developing malignancies[2]. Neurofibromatosis, the Cowden and Bannayan-Riley-Ruvalcaba syndromes, multiple endocrine neoplasia, and inherited mixed disorders are among the differential diagnoses[3]. The diagnosis is mainly based on medical history, family history, physical examination (characteristics of melanin spots), combined with gastrointestinal endoscopy, gastrointestinal imaging, and histopathology . The main therapeutic choices at this time are polypectomy using endoscopic procedures, reexamination, and follow-up. Intussusception is the condition that most frequently necessitates surgery in this patient population, and it may not

HIGHLIGHTS

- There is no proof that these patients' neoplastic lesions are brought on by the transformation of hamartomatous polyps.
- To avoid gastrointestinal issues and potential cancers, it is advised to execute a thorough screening programme and diagnose Peutz-Jeghers syndrome early.
- Patients with Peutz-Jeghers syndrome are advised to undergo complete blood counts, liver function tests, specialised blood tests, and chest radiography in addition to annual general internal medicine, chest disorders, gynaecological, or urology clinical examinations.

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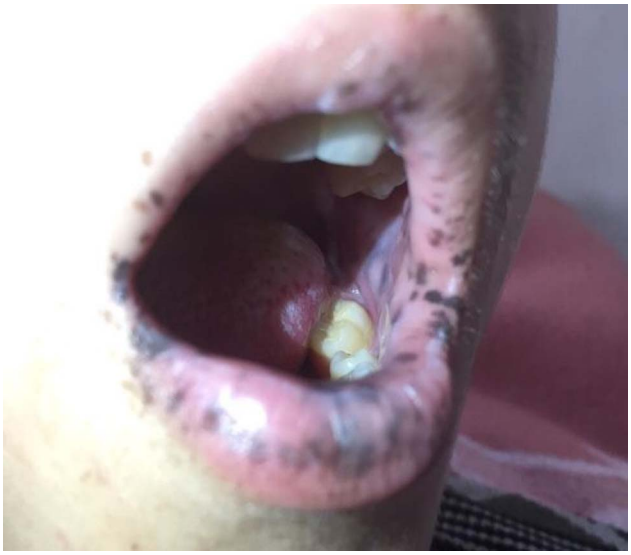

Figure 1. Multiple pigmentations were observed around the mouth.

always be possible to identify before surgery[5]. Herein, we report a case of a 15-year-old male who presented to the Ambulance Department of hospital for Generalized abdominal pain with not passing stool and frequent vomiting accompanied by decreased appetite to turn out later that he had peutz-jeghers Syndrome.

Case presentation

A 15-year-old male presented to the Ambulance Department of hospital for Generalized abdominal pain with not passing stool and frequent vomiting accompanied by decreased appetite. There was no family history of such symptoms in our patient. The patient had a history of surgical laparotomy for a month and a half ago due to intussusception. During the clinical examination, multiple pigmentations were observed around the mouth (Fig. 1). The necessary radiographic and laboratory investigations were performed. The abdominal radiography of the standing patient demostrated air-fluid levels (Fig. 2). An abdominal Ultrasound demonstrated Severe dilatation of the intestinal loops with intense peristaltic movements and An intestinal loop was observed behind and above the bladder measuring 80×46 mm. The wall of loop was thickened and oedematous with 20 mm. Peristaltic movements were very weak. The experts decided to perform an emergency laparotomy for the patient, where was watched a necrotic intestinal intussusception measuring 60 mm (Fig. 3).

Amputation and an ileoileostomy were performed, and the sample was sent to the laboratory of pathology to find out several polyps (Benign hamartomatous polyps). To exclude other differential diagnoses like familial adenomatous polyposis (FAP), multiple endocrine neoplasia type 2 (MEN2), Carney complex, etc. Genetic testing for the identification of disease-causing (pathogenic) mutations in the STK11 gene was performed, and it was positive. The final diagnosis had been made: Peutz-Jeghers syndrome.

The patient was followed up and discharged from the hospital when he improved. Recommendations were given to the family to perform upper and lower gastrointestinal endoscopy, with monitoring the patient.

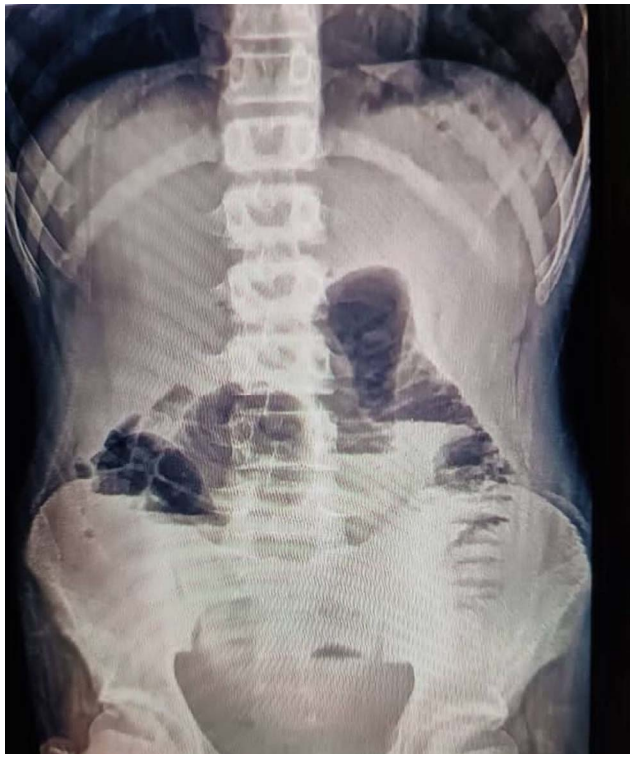

Figure 2. The abdominal radiography of the standing patient demostrated air-fluid levels.

Discussion

PJS was originally recognized in a case report written in 1921 by a Dutch physician called Jan Peutz, who discovered a connection between mucocutaneous macules and gastrointestinal polyps in a family. In his research in 1949, American physician Harold Jeghers verified that the illness constituted a syndrome[3]. It is

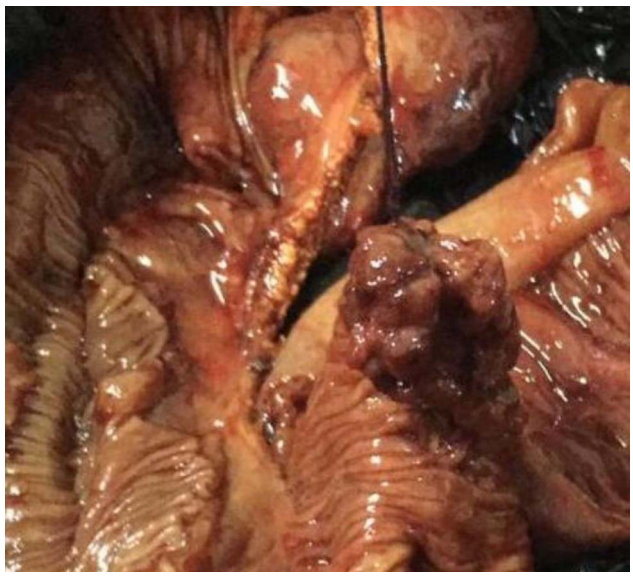

Figure 3. A necrotic intestinal intussusception measuring 60 mm.

autosomal dominant and more prevalent in children and teenagers. According to some research, 30% of diseases are passed from parents to children while 70% may result from gene mutations[5,6]. A germline mutation on the tumour suppressor gene serine/threonine kinase 11 (STK11/LKB1) on chromosome 19p13.3 has been linked to this extremely rare condition, which has an incidence of 1 in 12–30 000 live births[7]. Recent research has revealed that the PJS gene is a serine/threonine kinase, also referred to as LKB1 or STK11, and it corresponds to chromosomal subband 19p13.3[8]. This gene functions as a tumour suppressor in the hamartomatous polyps of PJS patients as well as in the other neoplasms that emerge in PJS patients. Its putative coding area is 1302 bp and has been divided into nine exons. Although the development of these neoplasms from hamartomas is probable, it is still possible that the LKB1 locus contributes to a separate genetic pathway of tumour growth in PJS patients' malignancies. In 50–75% of Peutz-Jeghers patients, previous research employing genomic DNA or cDNA sequencing as a primary screen discovered germline LKB1 mutations[9]. A shortened protein is the outcome of the majority of these mutations, which are either frameshifts or nonsense alterations. Missense mutations and in-frame deletions are less common and typically occur at conserved amino acids in the kinase core (codons 50–337)[10]. Despite being uncommon, PJS has grown in recognition and has distinct, clinical and histological-based diagnostic criteria. Beggs and colleagues recently recommended to evaluate the diagnosis in the presence of (1) two or more PJS polyps that have been histologically proven, (2) any number of PJS polyps in a person who has a close relative who has the condition, (3) distinctive mucocutaneous pigmentation in a person who has a close relative who has the condition, or (4) any number of PJS polyps in children[1]. PJS is typically discovered in the second decade of life as a result of polyposis-related problems such abdominal pain, bowel obstruction, intussusception, or occult gastrointestinal bleeding[4]. PJS patients may also experience acute blood loss and chronic anaemia as a result of the ulceration of polyps[2]. In our case, An unusual 15-year-old male was admitted to the emergency department with a generalized abdominal pain with not passing stool and frequent vomiting accompanied by decreased appetite . According to experts, the jejunum is the preferred place for PJ polyps, which may serve as a leading point for intussusceptions in more than 95% of instances. Furthermore, polyp size was presumably an important risk factor for small bowel intussusception, as they were frequently caused by hamartomas greater than or equivalent to 15 mm in diameter[1]. In our case, there was a jejunojejunal intussusception (JI) with complete intestinal obstruction. As in our case presentation, the literature search suggests that small intestinal polyps are frequently of this origin. There have also been reports of polyps outside the small intestine in the literature. The polyp in the Hammouda and colleagues case presentation originates from the ascending colon. When PJS is suspected during pre-diagnosis, the diagnosis can be made in childhood if there is a positive family history because the condition is inherited autosomally dominantly and there is mucocutaneous pigmentation, which is common from birth[3]. An important characteristic for an early diagnosis is lentiginosis of the lips and oral mucosa[2]. Only 5% of all occurrences of intussusception are found in adults, and most cases are documented in children. One to five percent of adult mechanical ileal obstructions are caused by intussusception. In 80% of cases, this condition, which is typically benign and rare in children, responds to medical intervention. On the other hand, a pathogenic condition is secondary in around 90% of documented cases of intussusception in adulthood. In cases of adult intussusception, the differential diagnosis should take into account the existence of the main pathology that causes intussusception, such as polyps, carcinomas, Meckel's diverticulum, colon diverticulum, strictures, and benign neoplasms[3]. To make a primary diagnosis for our patient, Abdominal ultrasound showed Severe dilatation of the intestinal loops with intense peristaltic movements and An intestinal loop was observed behind and above the bladder measuring 80×46 mm and the wall of loop was thickened and oedematous with 20 mm. Peristaltic movements were very weak. An emergency laparotomy for the patient was required, where was watched a necrotic intestinal intussusception measuring 60 mm (Fig. 3). Amputation and a ileoileostomy were performed, and the sample was sent to laboratory of pathology to find out several polyps(Benign hamartomatous polyps. The final diagnosis had been made"Peutz-Jeghers Syndrome. Hamartomatous polyps are the main disease that causes intussusception in patients with PJS. Intussusception is the condition that most frequently necessitates surgery in this patient group, and it may not always be able to identify before surgery[3]. In the Wang *et al.*[6] cohort research, only one case of intestinal blockage caused by intussusception was identified intraoperatively; all other cases were identified preoperatively. In our case presentation, intraoperative exams revealed the existence of intussusception caused by a jejunal polyp. In light of the available research, we believe that the size of the polyp, the length of the invaginated loop, and the efficacy of pre-operative imaging examinations all have a role in the pre-operative or intraoperative diagnosis of intussusception secondary to jejunal polyp[3]. According to a review of the medical literature on PJS treatment, the early management of gastrointestinal polyposis is the main focus of clinical PJS care. According to several research, a thorough system of endoscopic procedures, meticulous follow-up, combining surgery and preventive medications, and close monitoring are an effective diagnosis and treatment strategy for PJS gastrointestinal polyps. For people with scattered polyps, endoscopic resection can be done at various periods and is the primary procedure used among them. According to certain research, double-balloon enteroscopy is extremely valuable and significant for the diagnosis and care of PJS. It not only enables complete small intestinal inspection but also microscopic resection procedures, avoiding some surgical resection of the small bowel. Despite the challenging endoscopic removal of polyps, malignant transformation, and acute abdomen in PJS patients, surgical treatment is still required. However, from a genetics standpoint, both endoscopic and surgical procedures are still only partially effective in the treatment of PJS because it is a dominant hereditary condition[5].

Conclusion

In conclusion, PJS is an autosomal dominant illness characterized by mucocutaneous pigmentation and hamartomatous polyps in the gastrointestinal system, which are brought on by a germline mutation of the serine threonine kinase 11. Individuals diagnosed with PJS are more likely to acquire cancer or develop benign polyps in the stomach and other organs that turn into malignant ones. This patient's primary complaint is sporadic blockage of the

upper intestine brought on by large duodenal polyps, which hardly ever clog the duodenum. Since there are no clear guidelines for treating duodenal PJS hamartomatous polyps, each case needs to be managed specifically.

Methods

The work has been reported in line with the SCARE criteria[11].

Ethical approval

Not applicable because all date belong to the authors of this article.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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None.

Author contribution

M.S. wrote most of the manuscript and performed data analysis or interpretation and designed the study. M.A. wrote a part of the manuscript . I.A. wrote a part of the manuscript and designed the study . O.A. wrote a part of the manuscript. A.K. wrote a part of the manuscript. B.S. wrote a part of the manuscript . H.A. wrote a part of the manuscript. All authors reviewed the final manuscript.

Conflicts of interest disclosure

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References

[1] Rahma C, Najoua BK, Mohamed Z, *et al*. Peutz–Jeghers syndrome revealed by recurrent small bowel intussusceptions in children: a case report. Clin Case Rep 2022;10:1–5.
[2] Ben-Hammouda S, Njima M, Ben-Abdeljelil N, *et al*. An unusual presentation revealing Peutz-Jeghers syndrome in adult. Ann Med Surg 2020;58:87–90.
[3] Aytin YE, Türkyılmaz Z. A rare cause of mechanical intestinal obstruction due to small bowel intussusception: "A solitary Peutz-Jeghers type hamartomatous polyp." Turkish J Trauma Emerg Surg 2022;28:879–83.
[4] Shakil S, Aldaher Z, Divalentin L. Peutz-Jeghers syndrome presenting with anemia: a case report. J Cureus 2022;14:1–4.
[5] Jiang HH, Lu F, Tan SG, *et al*. Peutz-Jeghers syndrome manifested as multiple polyps in jejunum with intussusception. J Med Cases 2022;13:302–6.
[6] Wang H, Luo T, Liu WQ, *et al*. Clinical presentations and surgical approach of acute intussusception caused by Peutz-Jeghers syndrome in adults. J Gastrointest Surg 2011;15:22–18.
[7] Altamish M, Dahiya R, Singh AK, *et al*. Role of the serine/threonine kinase 11 (STK11) or liver kinase B1 (LKB1) gene in Peutz-Jeghers syndrome. Crit Rev Eukaryot Gene Express 2020;30:245–52.
[8] Su GH, Hruban RH, Bansal RK, *et al*. Germline and somatic mutations of the STK11/LKB1 Peutz-Jeghers gene in pancreatic and biliary cancers. Am J Pathol 1999;154:35–40.
[9] Wang Z, Wu B, Mosig RA, *et al*. STK11 domain XI mutations: candidate genetic drivers leading to the development of dysplastic polyps in Peutz-Jeghers syndrome. J Hum Mutat 2014;35:851–8.
[10] Jishage K, Nezu J, Kawase Y, *et al*. Role of Lkb1, the causative gene of Peutz-Jegher's syndrome, in embryogenesis and polyposis. Proc Natl Acad Sci USA 2002;99:89–3.
[11] Agha RA, Franchi T, Sohrab C, *et al*. The SCARE 2020 guideline: updating consensus Surgical Case Report (SCARE) guidelines. Int J Surg 2020;84:226–30.