Case Report

Peutz Jeghers Syndrome presented as intermittent gastric outlet obstruction and ileoileal intussusceptions

Abdul Hafeez A. Al-Selwi[1], Abdullah Dahan[2], Yasin MA Kadir[3]

Introduction:

Peutz Jeghers Syndrome (PJS), was first described in 1921 by Peutz[1], followed by Jeghers et-al in 1949[2]. It is characterized by muco-cutaneous melanin pigmentation, gastrointestinal hamartomatous polyps. It is not rare, several cases have been reported[3-7]after the first description by Peutz in 1921[1,8]. It is an autosomal dominant disease, one (PJS) locus has been mapped to chromosome 19p13.3, second locus is suspected on chromosome 19q13.4 in a minorities of families, the gene on 19p13.3 encodes the serine/thereonine kinase LKB1[9,10] . the patients of (PJS) is not only at higher risk of developing gastrointestinal cancers ,but also other cancers like breast, thyroid, ovaries, endometrial, pancreatic, and testicular tumours[3,5,11,12]. The Afro- Caribbean people are rarely affected[3,5]. The polyps are most commonly located in the small intestine (64-96%), but occur also in the stomach (24-49%), and in the colon (60%)[13,14]. These polyps vary in size from few millimetres to several centimetres, with lobulated surface, they could be pedunculated or sessile[15].

The clinical symptoms of the disease is recurrent abdominal pain, intestinal intussusception, , gastrointestinal bleeding with symptoms of iron deficiency anaemia, the symptoms usually takes place in the second and third decade of life[(6, 16)].

The polyps may develop in extra-intestinal sites like, bronchus, oesophagus, nose, bladder, renal pelvis, and ureters[17]. The pigmentation is usually muco-cutaneous, developing on the lips, buccal mucosa, fingers, toes, and periocular pigmentation[18].

The treatment is combined surgical endoscopic approach using intra-operative endoscopy or combined push enteroscopy and intra operative endoscopy have been recently advocated[21,22]. The malignant changes of hamartomatus polyps and other organs cancers have been reported in patients with (PJS)[23].

The aims of presenting this case here is that, the polyps in (PJS) could cause gastric outlet obstruction and intestinal obstruction.

Case report:

An 18 years old female patient, single, has four brothers and four sisters, both father and mother are alive and healthy. She presented to the hospital, complaining of recurrent vomiting and abdominal pain, for ten days. She had no malaena or haematemesis, no diarrhoea, fever, drugs intake, or trauma to the abdomen. On examination: B.P was 90/60, pulse 104/min, dehydrated, but she was not pale, there were pigmented spots over the lips (figure 1), buccal mucosa, fingers and toes (figure 2, 3). Abdominal examinations, revealed tender abdomen with no palpable mass, bowel sounds were exaggerated, respiratory, cardio-vascular and, nervous systems were all normal. The thyroid gland was not enlarged, no lymph node enlargement.

Investigations: Hb 12gm./dl, WBCs 7600/mm^3, Platelets 280,000, ESR was 30mm in the first hour, urinalysis was normal, liver and renal functions tests were normal. Her abdominal ultrasound reported a big mass 70×41mm occupying the lumen of the stomach in the pyloric area partially

1. Assoc. Prof. of Medicine
2. Assistant Prof. of Surgery
3. Professor of Medicine Sana"a Med College Yemen Republic.
*Correspondence to: Abdul Hafeez A al Selwi
PO box 22589 Sana'a Republic of Yemen
E- mail hafeezali42@yahoo.com

obstructing the lumen. Gastroscopy showed a big polyp blocking the pyloric canal. Colonoscopy showed normal large bowel.
The patient was operated. A big polyp 7×4cmm obstructing the pylorus, which wasresected. The patient ran uneventful post operative period discharged in good condition. Four weeks later, she returned complaining of abdominal pain colicky in nature associated with vomiting and constipation. She was diagnosed as a case of intestinal obstruction, and hence re-operated. Three polyps were found in the jejunum, the biggest was 2.5×1.5cms, and an ileo-ilealintussusception due to a big polyp in the middle of ileum. All polyps were resected and about 20cm of the middle ileum was resected end to end anastomosis.
Histopathology revealed hamartomatous polyps. The patient was discharged after one week in good condition and was scheduled for regular follow-up.

Fig1: Melanotic spots in the lips.

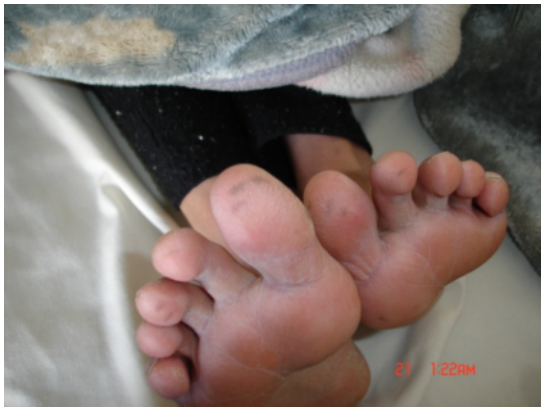

Fig2: Melanotic spots in the toes

Discussion:
PJS is an autosomal dominant disease characterized by hamartomatous polyposis throughout the gastrointestinal tract and melanosis on the lips, oral and gingival mucosae[24-26]. Also, melanosis occur in eye-lids, fingers hands, back, toes, and sole, and less frequently the lumbo-sacral areas, and the perineum[2].

Fig3: Melanotic spots in the tip of the index.

The morbidity of this syndrome is due primarily to lesions of small intestine[6], that generally demand repeated enterectomies leading eventually to short bowel syndrome [27,28], but endoscpic polypectomy, is the treatment for the polyps in the stomach, duodenum and colon [29,30,31]. The polyps are hamartomatous associated with hyperplastic and adenomatous polyps[32]. Ultrasonography is useful for follow up of these patients along with endoscopy[33,34,22]. The risk of cancer of (GIT) associated with this syndrome is higher than general population and range from 3% to 48%[34].
Other cancers related to (PJS) includes bilateral breast carcinoma[35],cervix tumours[36], ovarian tumours[11], testicular tumours[37], carcinoma of gall-bladder[38], pancreatic adenocarcinoma[39], gastric[40], duodenal[41], jejunal[42], illeal[43], rectal[44], and male sex-cord tumors[45].
Our patient did not have any malignancy neither in gastrointestinal tract nor in other sites, The other point of interest is that she had no family history of (PJS). Her family members have no any mucocutaneuos

pigmentations and no history of abdominal pain, bleeding per rectum, or anaemia, but unfortunately the father refuses any endoscopic screening of any other family member.

Conclusion:

Any patient with muco-cutaneuos pigmentations should be screened with abdominal ultra-sonography and endoscopy for polyps, and this syndrome should be thought of in any case presented with intestinal obstruction. These patients should be screened regularly for malignancies. If a polyp is found to be the cause of obstruction in any site of the GIT, all other parts should be carefully looked for.

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