Case Report

Peutz Jeghers Syndrome Presented as intermittent gastric outlet obstruction

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Peutz Jeghers Syndrome (PJS), which was first described in 1921 by Peutz[1], followed by Jeghers et-al in 1949[2], is an uncommon but not a rare disorder characterized by mucocutaneous melanin pigmentation, gastrointestinal hamartomatous polyps and increased risk of gastrointestinal and other organs cancer [1, 3, 4].

The polyps vary in size from few millimeters to several centimeters, with lobulated surface, and could be pedunculated as in large polyps, or sessile as in the small ones[5].

The clinical symptoms of the disease are recurrent abdominal pain, intestinal intussusception and obstruction, gastrointestinal bleeding with symptoms of iron deficiency anaemia. The symptoms usually take place in the second and third decade of life[6].

The complications of PJS which brings the patient to the doctor is severe anemia due to blood loss from GIT, acute abdomen, intestinal obstruction due to tumor-mass obstruction or intussusception ,or rarely due to gastric outlet obstruction as in this presented case[7].

The treatment of polyposis is by endoscopic polypectomy, laprotomy and resection which may lead to complications as short bowel syndrome. The other difficulty which the doctor comes across while managing these cases is to follow-up the occurrence of malignant disease in these patients ,malignant changes of hamartomatus polyps and other organs cancers have been reported in patients with PJS[8].

Keywords: hamartomatus polyps, polypectomy, mucocutaneous.

Case report:

AN 18 years old female patient - unmarried, has four brothers and four sisters, both father and mother are alive and healthy, with no family history of any medical or surgical illness – had presented to the hospital, complaining of repeated vomiting and abdominal pain, for a period of ten days. There was no melena or hematemesis, diarrhea, fever, drugs intake or trauma to the abdomen and her past medical history was unremarkable.

On examination: B.P 90/60, pulse was 104/min, dehydrated and was not pale. There were pigmented spots over the lips (figure 1), buccal mucosa, fingers and toes (figure 2, 3). Abdominal examination revealed tender abdomen and exaggerated bowel sounds on auscultation. Other systems were normal.

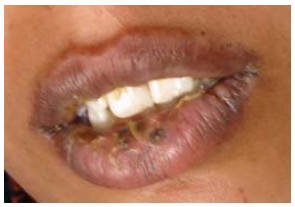

Figure1: Pigmented lip.

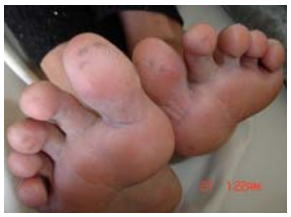

Figure2: Pigmented toes.

Figure3: Pigmented fingers.

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Her investigations showed: Hb.12gm./dl, WBCs.7600/mm^3, Platelet counts was 280,000, ESR is30/hour, urine analysis is normal, liver and renal functions tests are normal. Abdominal ultrasound reported a big mass 70×41mm occupying the lumen of the stomach in the pyloric area partially obstructing the lumen creating the possibility of a tumor mass or a giant polyp. Upper gastroendoscopy demonstrated a big polyp blocking the pyloric ring (figure 4), colonoscopy was normal.

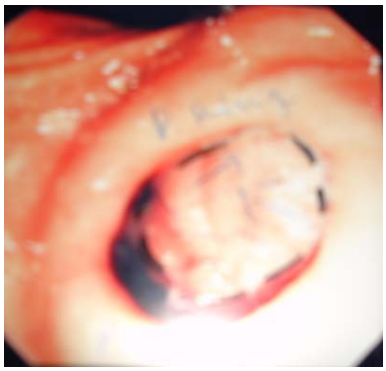

Figure4: Gastric polyp.

The patient was operated on and found to have a big polyp 7×4 cmm obstructing the pylorus, this polyp was resected through gastrotomy and was sent to histopathology for analysis, the patient ran uneventful postoperative days and discharged on the fifth postoperative day in good condition. Four weeks later, she returned complaining of abdominal pain around the umbilicus, it was colicky in nature and associated with vomiting and constipation. Clinical examination revealed intestinal obstruction. She was operated again three polyps were found in the jejunum. The biggest was 2.5×1.5cms. Another 4.5× 3cmms polyp causing an ileo-ileal intussusception was detected in the middle portion of ileum. Those polyps in the jejunum were resected by endo-luminal resection after enterotomy, while about twenty cmm. of the middle ileum was resected with the polyp, and end to end anastomosis was performed. The other parts of the gastrointestinal tract were normal.
The histopathology revealed hamartomatous polyps. The patient was discharged after one week in good condition without any complications and was scheduled for regular follow-up visits in the out patients clinic.

Discussion:

PJS is an autosomal dominant disease characterized by hamartomatous polyposis throughout the gastrointestinal tract as well as mucocutaneous lentiginosis ,mostly on the lips ,oral and gingival mucosae[9-11], however it can also attacks ,eye-lids ,fingers hands ,back ,toes ,and sole , and less frequently the lumbo-sacral areas ,and the perineum[2].
The morbidity of this syndrome is due primarily to lesions of small intestine[12], that generally demand repeated enterectomies leading eventually to short bowel syndrome[13, 14], but because of facilities of endoscpic polypectomy , the polyps in the stomach , duodenum and colon can be managed by this method without any complications[15]. The polyps that appear in the GIT are almost always hamartomatous and are of multiple sizes, sessile or pedunculated , however ,some of these are associated with hyperplastic and adenomatous polyps[16]. The polyps of PJS are generally not real neoplasms, but sometimes their large size can cause small intestinal obstruction and intussusception as in this case, so ultrasonography is useful for follow up of these patients along with endoscopy[17, 18].The risk of cancer of GIT associated with this syndrome is higher than general population and ranges from 3% to 48%[18].
Our patient has no malignancy, neither in gastrointestinal tract nor in other places. No neoplastic changes in the polyps removed , but her rare presentation is ,that she had a big gastric polyp causing intermittent gastric outlet obstruction ,which ,occludes the pyloric ring ,along with ileo-ileal intussusception ,due to a large polyp (4.5×3cmm) ,in the mid ileum , the jejunum containes three polyps the largest is (2.5×1.5cmm) . In spite of that she had no anemia and no any history of melena or bleeding per rectum, colonoscopy demonstrated no polyp in the colon.
The other point of interest is that she had no

family history of PJS. Unfortunately the father refused endoscopic screening of other family member for polyposis.

Conclusion:

Any patient with muco-cutaneuos pigmentations should be screened with abdominal ultrasonography and endoscopy for polyposis and this syndrome should be thought of in any case presented with intestinal obstruction. If a polyp is found then screening for other polyps is mandatory. The Hb level of these patients should be assessed as part of follow up to detect GIT bleeding. These patients should be screened regularly for malignancies.

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