Peutz-Jeghers Syndrome in Children and Adolescents
Almoutaz A El Tayeb, Naglaa H. Ibrahim, Azza A. El Tayeb, A.E. Essa
Departments of Paediatric Surgery and Pediatrics, Faculty of Medicine, Assiut University

Abstract
Background/Purpose: The syndrome is characterized by multiple polypi in the small intestine, and may be the colon and the stomach, in addition to melamn pigmentaion of the lips, oral mucosa and possibly other areas of the skin. These hamartomatous polypi are liable for malignant transformation and may cause intestinal obstruction. This characteristic pigmentation is of a diagnostic importance when associated with abdominal pain, bleeding per rectum, anemia or evidence of intestinal obstruction due to intussusception. Most cases are inherited in an autosomal dominant pattern. Also, the syndrome may be associated with malignancy in other sites of the body away from the gastrointestinal tract (GIT) which should be looked for. The aim of this study is to highlight the importance of the oral pigmentation in the diagnosis of this syndrome and the need for investigating the patient and his family for the possibility of presence of gastrointestinal polypi or malignancy in or outside the gastrointestinal tract.

Materials & Methods: Three new cases of Peutz-Jeghers syndrome were reported. All presented with recurrent abdominal pain, sometimes with manifestations of incomplete intestinal obstruction and oral pigmentation. Intestinal resection was done in one case, endoscopic removal in another and resection through multiple enterotomies was performed in the third.

Conclusion: Every patient suffering from recurrent abdominal pain and/or bleeding per rectum in presence of this characteristic pigmentation should be screened clinically, endoscopically and radiologically. Family screening for all suspected members in his family is recommended because the syndrome may affect more than one member in the family. The bowel resection should be conservative or better avoided because the polypi are usually widespread and recurrent.

Index Word: Peutz-Jeghers syndrome, intestinal polyposis.

INTRODUCTION

The association of intestinal polypi with mucocutaneous pigmented spots of the mouth, hands, feet and possibly other areas was first reported by Peutz in 1921.1 Jeghers2 reported two cases and in 1949 added 8 more in addition to 12 cases he had collected from personal communication. In 1954, Bruwer et al4 were the first to use the term Peutz-Jeghers syndrome. Essa and others5,6 reported five cases at Assiut University Hospitals. Reid and Pertelli7 collected 492 reported cases in the literature till December 1974.

The syndrome is characterized by melanotic spots ranging in color from brown to black, occurring on the lips, around the mouth and on the oral mucosa. The spots may also be found in hands, feet, nasal mucosa, forehead, conjunctiva and rectum. The polyps occur most commonly in the small intestine
but may be found in the stomach and duodenum (30%) or colon and rectum (15%).

The syndrome is rare, has an equal sex distribution and most of the cases are inherited as in autosomal dominant pattern, but some develop do novo, most likely representing new, spontaneous mutation.

The usual clinical presentation is recurrent abdominal pain due to transient intussusception of the polyp. Anemia resulting from occult blood loss and malignant conditions in the GIT or away from it is other presentation. Thirty percent of patients with syndrome presents in the first 10 years of life, 50% presenting by 20 years of age. About 22% of the cases develop cancer (13% gastrointestinal or pancreatic and 9% were extra intestinal). The chance of dying from cancer in patients with Peutz-Jeghers syndrome by the age of 60 years is about 60% compared to 25% in an age matched general population.

In recent years, the reports of intestinal and extra intestinal cancers developing with the syndrome have led to a reassessment of the management of these patients. Extra intestinal tumors associated with Peutz-Jeghers syndrome include ovarian, uterine cervix, testicular, breast, thyroid, bile duct, pancreas and gall bladder.

**PATIENTS AND METHODS**

Eight patients Peutz-Jeghers syndrome were treated at Assuit University Hospital over a 30 year period. The first five patients were reported earlier by Essa et al. Three more new cases are presented in this work and the relevant literature is reviewed (Figs. 1-6). All patients presented with recurrent abdominal pain and recurrent incomplete intestinal obstruction in addition to oral pigmentation. Limited intestinal resection was done in one case where the polyps were localized to a small segment of jejunum, and due to possibility of malignancy because of the presence of enlarged mesenteric lymph nodes. In another case endoscopic removal of the polyps from the colon was accomplished. In the third case, abdominal exploration and ligature excision of the polypi through multiple enterotomies was done.

**DISCUSSION**

Polypoid diseases of the gastrointestinal tract (GIT) may be restricted to the intestine (juvenile, lymphoid and familial adenomatous polyps (FAP), or involving other parts of the body such as uncommon syndromes mainly Peutz-Jeghers syndrome, Gardner and Turcot syndrome.

Most polypli of the GIT are benign and result from hamartomas of the mucosa or lymphoid hyperplasia of the submucosa. Adenomatous polypi, however, occurs as a disturbing alteration in the mucosa that has substantial malignant potential.

GIT polyps are common during childhood occurring in about 1% of preschool- and school-aged children. They represent the most common cause of bleeding per rectum in this age group. The juvenile polyps accounts for 80%, followed by lymphoid polyps (15%) and adenomatous polyps (<3%) of all children with polypli. The polypli occurring with syndromes mentioned above (Peutz-Jeghers and Gardner and Turcot) accounts only for 1-9% of polypli.

**The juvenile polypi are classified into:**

- The isolated juvenile polyps (constitute about 80% of all polypli in children 1-5 in number) and they are hamartomatous polypli and are not premalignant.
- The juvenile polyposis syndromes which is rare and have a malignant potential.

The lymphoid polypi are benign, self-limiting and tend to regress spontaneously.

The adenomatous polypli of the FAP are premalignant in all cases. The polypli with Peutz-Jeghers, have a malignant potential in about 22% of cases. In Gardner's and Turcot syndromes the polypli are adenomatous polypli and have the malignant potential of FAP. The number of accepted cases of cancer in Peutz-Jeghers syndrome was 33 from the 492 cases reported by Reid and Pertelli (1974) (i.e. about 6%). Of these 33 cases, 16 (49%) involved the colon and rectum, 5 cases (15%) involved the jejunum, 4 cases (12%) occurred in the duodenum, 3 cases (9%) affects the stomach and duodenum in continuity and 5 cases (15%) affects the stomach.
Fig 1. Six-year old boy with oral pigmentation and small intestinal polyps presented as intermittent intestinal obstruction and bleeding PR.

Fig 2. Eleven-year female child with oral pigmentation and intestinal polyposis. Her brother 19 years old has oral pigmentation but no confirmed intestinal polyps.

Fig 3. Multiple polypi 1-3 cm removed through multiple enterotomies.

Fig 4. H. & E stain for one of the polyps showing its hamartomatous nature.

Fig 5. Contrast enhancement CT scanning showing intussusception caused by intestinal polyp.

Fig 6. Brownish pigmentation on the lips, hard and soft palate with intestinal polyposis.
As an unusually high proportion of these cases involves parts of intestine which are ordinarily least affected by cancer and as the mean age in these patients is significantly lower than that ordinary cancer, there is thus a strong evidence of the small risk of malignant transformation in Peutz-Jeghers cases.

So, any child presenting with abdominal pain and melonic pigments should be suspected to have Peutz-Jeghers syndrome and should be examined, investigated and followed up together with his suspected family members. Essa et al found that 60% of the patients family members have the characteristic pigmentation and 44.6% have increased mutant P53 gene on chromosome 17. In the present work 50% of the patients family members were affected by this melanotic pigmentation. The management protocol proposed by Philips and Spigelman include the following annual evaluation for suspected cases:

- Observation of symptoms related to polyps.
- Blood count to detect anemia caused by blood loss.
- Breast and pelvic examination with cervical smears and pelvic ultrasonography in girls.
- Testicular examination with ultrasonography in boys.
- Pancreatic ultrasonography in addition esophagogastroduodenoscopy, colonoscopy are recommended on biennial basis, along with small intestine contrast studies.
- Mammography is recommended at 25, 30, 35 and 38 years of age, biennially until 50 years of age, and then annually.

It is to be noted that visualization of the polypi in the small intestine is not easy and repeated barium meals and barium follow through, barium contrast enema and endoscopy is essential for definite diagnosis.

All polyps larger than 0.5 cm in diameter found at endoscopy should be removed. Laparotomy with intraoperative enteroscopy or enterotomy which may be multiple, and removal of all small intestinal polyps larger than 15 mm in diameter should be done. The old practice of radical intestinal resection should be avoided because of the recurrent nature of the polyps and the ensuing short-bowel syndrome that can occur. Any intestinal and extra intestinal cancer should be treated aggressively.

**CONCLUSION**

Investigation and follow up of cases with oral pigmentation together with the suspected cases of their families for evidence of intestinal polyposis and for the danger of intestinal obstruction and/or malignancy in the GIT or outside is highly recommended. Resection of the polypi should be conservative because they are multiple, recurrent and benign in the majority of cases.

**REFERENCES**


