

Case Report

A 16-Year-Old Female with Peutz-Jeghers Syndrome

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Abstract

Peutz-Jeghers syndrome is a rare autosomal dominant disorder of hamartomatous polyposis of the gastrointestinal tract, with pigmentation around lips and macules on the buccal mucosa that typically manifests itself as recurrent colicky abdominal pain and intestinal obstruction due to intussusception. Here we report a case of a 16-year-old girl who presented with abdominal pain, vomiting and previous history of laparotomy for intussusception. Multiple well demarcated black pigmented macules on lips, perioral region, buccal mucosa, digits, palms and soles were noted. She was diagnosed as a case of Peutz-Jeghers syndrome and managed conservatively.

Key words: Peutz-Jeghers syndrome; Intussusception; Polyps; Circumoral pigmentation

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Introduction

Peutz-Jeghers syndrome (PJS), also known as hereditary intestinal polyposis syndrome, is an autosomal dominant genetic disease characterized by the development of benign hamartomatous polyps in the gastrointestinal tract, presence of hyperpigmented macules on the lips and oral mucosa (melanosis) and susceptibility to multiple cancers.¹ It has an incidence of approximately 1 in 120000 births.²

A clinical diagnosis of PJS may be made when any one of the following conditions is present in a single individual: two or more histologically confirmed Peutz-Jeghers (PJ) polyps; any number of PJ polyps detected in an individual who has a family history of PJS in a close relative; characteristic mucocutaneous pigmentation in an individual who has a family history of PJS in a close relative or any number of PJ polyps in an individual who also has characteristic mucocutaneous pigmentation.³

The disease affects males and females equally. In addition to polyposis, the risk of gastrointestinal and extra-gastrointestinal malignancies is significantly

increased in PJS patients.⁴ The risk of any other malignancy (especially cancer of the reproductive organs, breast, pancreas and lung) is nine times greater than in the general population.⁵ A few years ago, two independent groups of investigators defined the mutated gene responsible for PJS.^{6,7} The gene was localized to chromosome 19p34-p36 and is known as STK 11, a serine-threonine kinase involved in growth control regulation.⁸ On the other hand, not all patients with PJS have a mutation in this gene.⁹ Here we report a case of a 16-year-old girl who presented with abdominal pain, vomiting and previous history of laparotomy for intussusceptions and subsequently was diagnosed as a case of PJS.

Case report

A 16-year-old girl presented in the Medicine OPD in Enam Medical College Hospital with the complaints of abdominal pain and vomiting. Multiple black pigmented macules on lips, perioral region, buccal mucosa, digits, palms and soles were noted (Fig 1, 2). Few months back she was admitted in the department of

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Surgery with same complaints and was diagnosed as a case of intussusception and exploratory laparotomy was done. Intraoperatively, jejuno-jejunal and ileo-colic intussusceptions were found and nobel multiple sites plication was done. No polyp was felt manually and no enterotomy or enteroscopy was done at that time.

The patient did not have positive family history of PJS or any other polyposis syndrome. Barium follow-through radiography was done and it showed multiple filling defects through entire small intestine (Fig 3). Endoscopy of upper gastrointestinal (GI) tract revealed several polyps of different sizes in stomach and duodenum (Fig 4, 5). PJS was diagnosed on the basis of presence of polyps and characteristic pigmentation. She was managed conservatively and counseled and advised to come for regular follow-up.



Fig 3. Barium follow-through X-ray showing multiple filling defects in small intestine



Fig 1. Hyperpigmentation in lips and finger pulp



Fig 4. Endoscopy of upper GIT showing multiple polyps in stomach



Fig 2. Hyperpigmentation in buccal mucosa



Fig 5. Endoscopy of upper GIT showing multiple polyps in duodenum

Discussion

PJS comprises multiple polyps of the stomach and bowel leading to transient intussusception and a brownish pigmentation around the mouth and lips. The melanin spots of PJS are present in more than 90% of patients and occur most commonly on the lips and buccal mucosa, but are found also on circumoral and facial skin, on the palms and soles and on the digits.¹⁰ The melanotic macules on the skin may not develop until after the intestinal polyps and may fade with age.¹¹ In this case, the patient had melanin spots in the circumoral region, digits, palms and soles.

Polyposis occurs most often in the intestinal tract, but can be found in nasal, bladder or gallbladder mucosa and in the pelvis or lungs.¹² In the intestinal tract, early polyposis leads to clinical signs. One-third of the time, first manifestations occur during the first decade of life; in one-half cases these occur before 20 years and sometimes it occurs in the neonatal period.¹³ This hamartomatous polyposis is located in the small bowel in 90% of patients, most often in jejunum, followed by ileum and duodenum.¹⁴ It is also found in the stomach (24%) and colon (9%). Most polyps are pedunculated, except on the gastric wall where they are sessile. They are mainly diagnosed by clinical manifestations secondary to complications, such as acute intestinal intussusception (43%), iron deficiency anemia (14%) linked with polyp ulceration, chronic abdominal pain (23%), anal prolapse of rectal polyp (7%), or transforming into gastrointestinal tract adenocarcinoma (2–3%).¹³

Follow-up of this polyposis disease raises the problem of its main localization in the small bowel, which is difficult to investigate. There are various endoscopic tools, but videocapsule endoscopy seems to be the easiest to use and of best quality for standard polyposis screening. Among endoscopic tools, double-balloon enteroscopy helps in proper diagnosis and location of polyps and permits the surgeon to resect or biopsy these polyps.¹⁵ Periodic endoscopic screenings are advocated every 2 years.¹⁶

Intussusception occurs when one loop of bowel telescopes into an adjacent segment. This has been observed in 47–69% of adult patients with PJS and

most of them were due to polyps located in the small intestine.¹⁷ The majority of intussusceptions reported in the literature are in ileal or jejunal regions.^{18,19} Colo-colonic intussusception is reported in only a few cases.²⁰ In this case, the patient developed jejuno-jejunal and ileo-colic intussusceptions.

Screening should not be restricted to the gastrointestinal tract. Because of the increased risk of malignancy, it is essential to set surveillance guidelines based on a high cancer risk adjusted for the patient's age and the organ involved. All reports recommend establishing a familial register and screening all possible patients with genetic analysis.

In conclusion, it is important to be vigilant and observant about the patients who have circumoral pigmentation and to go into the depth of history and physical findings. Patients with PJS should be regularly and closely monitored, so that it can reduce the risk of cancer and also reduce the number of laparotomies.

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