A Case of Peutz-Jeghers Syndrome Presenting as a Rectal Mass

Sumanth Subhramaniyam¹, Peter Manoharan², Aju James Ashok³, Amena Feroz⁴

¹Postgraduate, Department of General Surgery, Pondicherry Institute of Medical Sciences, Kalapet, Puducherry.
²Professor, Department of General Surgery, Pondicherry Institute of Medical Sciences, Kalapet, Puducherry.
³Senior Resident, Department of General Surgery, Pondicherry Institute of Medical Sciences, Kalapet, Puducherry.
⁴CRRI, Department of General Surgery, Pondicherry Institute of Medical Sciences, Kalapet, Puducherry.

PRESENTATION OF CASE

A 14-year-old boy presented with complaints of mass descending per rectum for 10 months which was manually reducible and non-progressive in nature. It was not associated with bleeding per rectum or pain or pus discharge. No history suggestive of alteration in bowel habits. No history of any abdominal pain or discomfort, vomiting. He also gives history discoloration around the lips and toes for past one year. The patient also has past history of intussusception for which he underwent exploratory laparotomy. Patient was conscious, oriented. His vitals - BP: 130/90 mmHg, pulse: 84/minute Patient was afebrile at the time of examination. Multiple discrete hyperpigmented macules were present on both lips. Abdomen was soft, non-tender, non-distended. Previous surgical scar was present. Other systems normal. On digital rectal examination, no external fissures / fistula. A mobile growth was felt at around 5 cms from the anal verge which was mobile which does not bleed on touch.

DIFFERENTIAL DIAGNOSIS

- Colorectal polyp which can occur from Cowden’s syndrome, PJS, Cronkite-Canada syndrome
- Internal haemorrhoids
- Rectal mucosal prolapse
- Solitary rectal ulcer
- Laugier-Hunziker (based on mucocutaneous pigmentation)¹
PATHOLOGICAL DISCUSSION

Peutz-Jeghers syndrome is an autosomal dominant condition characterized by gastrointestinal hamartomatous polyps. It has an incidence of one in 25000 births. Polyps found in other polyposis syndromes may share features found in the polyps of PJS. Cowden's syndrome can have a variety of hamartomatous polyps such as juvenile and hyperplastic polyps. Clinical features such as macrophalhy, papillomatus lesions and developmental delay are typically associated with Cowden's syndrome and help differentiate the syndromes Cronkite-Canada syndrome polyps can be differentiated from PJS as they are more inflammatory or hyperplastic.

Laugier-Hunziker is associated with macules in a similar location and with a very similar appearance to JPS. In contrast to PJS the macules seen in Laugier-Hunziker develop throughout adulthood. On the basis of clinical history and examination Internal haemorrhoids and rectal mucosal prolapse was ruled out. There were no features suggestive of solitary rectal ulcer Hence with the above clinical features and examination the diagnosis was more in favour of colorectal polyp. Hence colonoscopy was done to rule out colorectal polyps which showed large multihed polyps in rectum, sigmoid and descending colon from which random biopsies was taken which showed features of Peutz-Jeghers syndrome. Since the polyps were large and not pedunculated and as they cannot be removed endoscopically, surgical resection of the polyp was done. Intraoperatively multiple polyp was found in the sigmoid colon for which polyectomy was done and bowel walk was done there was no other polyp identified.

After one year he came for follow, patient came for follow up and was found to have another polyp in the colon, which was removed endoscopically, and he is on regular follow up. Histopathological examination of the specimen: Excised multiple polyps from sigmoid colon- features of hamartomatous polyp consistent with Peutz-Jeghers polyps.

DISCUSSION

Peutz-Jeghers syndrome (PJS) is usually associated with gastrointestinal polyposis, mucocutaneous pigmentation, and cancer predisposition. This gastrointestinal polyp usually predominates small bowel most commonly, but they can also develop in large bowel and stomach. Mucocutaneous hyperpigmentation may range from dark blue to dark brown macules. These mucocutaneous pigmentation occurs in infancy and diminishes in later adolescence. Pigmentation is seen in 95% of PJS patients. Most of pigmentation is found on the lips but can also be seen on the buccal mucosa, hands and feet. For paediatric age group, they may present with small bowel intussusception5, secondary to polyp and Emergent laparotomy for intussusception has been reported to occur in 30% of patients by 10 years of age. Other presentations include abdominal pain, rectal bleeding and rarely rectal prolapse of a polyp. The characteristic mucocutaneous pigmentation may be absent since this finding is most prominent during infancy. Individuals with Peutz-Jeghers syndrome are at increased risk for developing wide variety of epithelial malignancy with the lifetime risk of 95% and relative risk of 15.2%. The diagnosis of Peutz-Jeghers syndrome is based on clinical findings. The standard methods used to screen, and survey JP patients are colonoscopy and UGI scopy. Although capsule endoscopy has the potential for evaluation of the entire GI tract, its usefulness is primarily for evaluation of the small bowel. Routine endoscopic surveillance with polypectomy decreases the frequency of laparotomy and bowel loss due to intussusception. Diagnosis and management of small bowel polyp is complex which requires capsule endoscopy and MR enterograph. Occasionally intraoperative endoscopy and enterotomy is needed for removal of large distal small-bowel polyps. Patient with PJS are at increased chance of developing malignancy9 hence the require routine screening and surveillance and various protocols have been suggested to monitor stomach, small and large bowel, breasts, testicles, ovaries, uterus, and pancreas by various procedures as frequently as once a year.
**FINAL DIAGNOSIS**

Peutz-Jeghers Syndrome.

**REFERENCES**


