Case Report

A bizarre presentation of Peutz – jegher's syndrome in a 2 year old

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Abstract

Peutz - jegher's syndrome (PJS) is a rare autosomal dominant disorder with gastro intestinal and mucosal pigmentations.

We present a 2 y/o boy with a lesion prolapsing from his rectum that could not be reducted. We found him to have PJS and a familial history of PJS in his parents.

Keywords

- Peutz - jegher's syndrome
- Pediatric
- Rectal prolapse

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Case Report

The patient was a 2 y/o boy, referred to us with a complaint of prolapsed mass from rectum.

Parents stated that the problem had occurred 2 months ago but was resolved with manual reduction. Since then the patient had no complaints such as constipation, bowel habit change or bleeding.

On examination under anesthesia in the operation room a very large mass prolapsed from the rectum just above the anal verge.

Reduction was done without any problem and biopsy was taken.

The next day he again experienced prolapsed of a mass during defecation and manual reduction was not successful so the patient was scheduled for surgery.

A large vegetative mass with a small pedicle was found and excised and the rectal mucosa was repaired.

The mass was also sent for pathologic examination, and a more thorough family history was obtained in which it was discovered that the father suffered from PJS.

In histopathological examination hamartoma was reported which also pointed to PJS and the

patient was referred to a pediatric gastroenterologist for further study and follow up.

Discussion

The first article about PJS was published by peutz in 1921 and Jegher in 1949 reported this disease in 10 females. PJS transmission is autosomal dominant and is seen in a sporadic or familial pattern.[1] One in four of PJS patients present with no familial history, in a sporadic pattern. PJS has 2 important component: 1) Gastro intestinal hamartoma and 2) mucosal pigmentation. Mucosal pigmentation which is due to melanocytic accumulation can be seen in most patient and may be painful in child hood or infancy. Location of pigmentation is in the mouth or nasal mucosa or on the lips. Fingers, palm and sole are spared. Hamartomatous polyps are usually found in the small intestine but may involve the GI from the oral cavity to the rectum. The first symptoms of these polyps usually present during the second decade of life and almost always before the age 20. Intermittent abdominal pain is one of the most common symptoms that may occur due to invagination.[2]

Patient with PJS are prone to different malignancies (GI and non GI). Pancreas, lung, breast, uterine, cervix, ovary and testis are non GI organs which are susceptible to malignancy. Polyps have a Christmas tree pattern and differ with invasive adenocarcinoma. As such assessment of polyps is important, especially when the diagnosis is not certain.

Imaging studies include: GI series, GI endoscopy and some authors recommend sonography and MRI. Laparoscopic or open surgery are useful in emergency setting. During surgery, intraoperative endoscopy and entroscopy with capsule may be used. Now a days, double balloon endoscopy with capsule entroscopy are gold standard for diagnosis.[3]

References

1. Hemminki A, Markie D, Tomlinson I, et al: Peutz – jeghers syndrome. Nature 1998; 391:184-187.

2. Zivkovic V, Pejović S, Nagorni A, et al: Hereditary hamartomatous gastrointestinal polyposis syndrome. Scientific Journal of the Faculty of Medicine in Nis 2010;27(2):93-103.

3. Manfredi M:Hereditary hamartomatous polyposis syndromes: Understanding the disease risk as children reach adulthood. Gastroentrology & Hapatology 2010; 6(3), 185-196.