Puetz-Jeghers Syndrome Involving Appedix

*Dr.Raysan F. Al-Fayadh, M.D, F.R.C.S.E, F.A.C.S **Dr.Ali Jaliel Awad, C.A.B.S

Abstract

This is an autosomal dominant disease. The gene STK11 on chromosome 19 has been found in proportions of patients with this condition, this consists of:

A- Intestinal hamartomatosis.
B- Melanosis of the oral mucous membrane and the lips.

Introduction:

Puetz-Jegher's syndrome is an autosomal dominant hereditary disease, which is characterized by hamartomatous polyps and mucocutanous pigmentation mainly over the circum-oral region. Patients with Puetz-Jegher's syndrome seek medical attention whenever there are complications such as intussusception, bleeding from the polyps, etc.

Occasionally, gastrointestinal tract malignancies have been reported in Puetz-Jegher's syndrome. In this paper, we report a patient with Puetz-Jegher's syndrome who had multiple complications and polyposis involving the appedix, because involvement of the appedix is extremely rare in Puetz-Jegher's syndrome.

Key words: Puetz-Jegher's syndrome-polyposis-appedix.

Case report:

A 15-year old male with pigmentation in the circum-oral region tips of the fingers and toes presented with lower gastrointestinal bleeding and rectal prolapse. The patient underwent surgery 3 months earlier (by another surgical team) for ileo-ileal intussusception, the lead point of which was Puetz-Jegher's polyp.

Upper and lower gastro intestinal endoscopies revealed two small polyps at the antral region of stomach and three large sessile polyps at the rectosigmoid junction, which were responsible for the lower gastrointestinal bleeding. The lower gastrointestinal polyps could not be removed by polypectomy snare because they were too large and sessile. This operation was performed on November 2, 2008, and we found three large polyps at the rectosigmond region and moderiately enlarged and elongated appendix. No other polyp was palpable in the rest of the intestinal tract. We did a localized resection of the rectosigmoid region with primary anastomosis, Well's rectopexy, and appedicectomy. Histopathological examination revealed Puetz-Jegher's polyp in the appendix. The patient was doing well, no longer has bleeding from the rectum, and his rectal prolapse had been fixed.

Discussion:

The Puetz-Jegher's polyps are hamatomas that are mainly found in the jejunum and the ileum, but they can occur anywhere in the gastrointestinal tract or extraintestinal.

Polyps in the absence of pigmentation have also been reported. Usually patients with this syndrome seek treatment only

when there are complications such as intussusception, bleeding or rarely, malignant transformation. Those polyps situated in the stomach and duodenum have a small potential for undergoing malignant change. The incidence of intestinal malignant transformation in Puetz-Jegher's syndrome has been reported to vary from 2% to 3% and those of nongastrointestinal malignancies about 29%.[3]

Our patient had been operated on for lower gastrointestinal bleeding due to the three polyps at the rectosigmoid junction. Because the patient had an appedix that was moderately enlarged and elongated and contained fecalith-like swelling inside, we performed an apperdectomy in addition to localized resection of the rectosigmoid region having the polyps. The patient may develope appendicitis if we had left the appedix. At no stage during the surgery did we doubt that the appedix would eventually also be affected. To our surprise, the histopathology revealed Puetz-Jegher's polyps in the appedix in addition to the similar lesion at the rectosigmoid junction, both show similar picture comprising of benign glands of variable sizes and are supported by broad bands of smooth muscle fibers.[4]

Conclusion:

A very rare case of Puetz-Jegher's syndrome involving the appedix is reported. We have not found this reported elsewhere. Our case may be the first report of Puetz-Jegher's syndrome involving the appedix.

From this report we conclude that the appedix is also one of the sites of polyposis in Puetz-Jegher's syndrome and should be examined during operations performed for complicated Puetz-Jegher's syndrome.

References:

1- Taylor TV, Polyps of the large intestine, Surg Gastroentrol, 1985, 1: 399-401.

2- Bansekupin LA, Douglass MC, Localisation of Puetz-Jegher's macules to psoriatic plagues, Arch Dermatol, 1986, 122, 679-683.

3- Singh LJ, Moirangthem GS, Singh LP, Singh SI, Malignant jejunal polyp in Puetz-Jegher's syndrome, Trop Gastroenterol, 1998, 19: 24-25.

4- Reid ID, Intestinal carcinoma in Puetz-Jegher's syndrome, JAMA, 1974, 229: 833-834.