Peroperative enteroscopy and polypectomy in Peutz-Jegher syndrome

Mukesh Kalla, Lalit Bharadia, Tridiv Madhok, Komal Kalla, Rajesh Bhojwani, Ravi Saxena

S R Kalla Memorial Gastro and General Hospital, Jaipur, Rajasthan

We report a 32-year-old who lady when presented with anemia and was detected to have Peutz-Jegher syndrome. She had malignancies of the colon and ovary over a 2-year follow up and was successfully managed. On screening the family two more members were confirmed to have Peutz-Jeghers syndrome and have been put on surveillance. [Indian J Gastroenterol 2006;25:162-163]

Peutz-Jegher syndrome (PJS) appears to be inherited as a single pleiotropic autosomal dominant gene with variable and incomplete penetrance.1 PJS is characterized by gastrointestinal polyposis and mucocutaneous pigmentation. Individuals with PJS are at increased risk for intestinal and extraintestinal malignancies.

A 32-year-old lady was referred for work-up of iron-deficiency anemia. She had stool occult blood positive on many occasions. On examination she had hyperpigmented purplish macular lesions in the perioral area apart from moderate anemia. Upper GI endoscopy was normal. Colonoscopy revealed a single 5-cm sessile polyp at the splenic flexure; biopsy revealed moderately differentiated adenocarcinoma. Rest of the colon was normal. CT scan abdomen confirmed resectability of the tumor. Barium meal follow-through and mammogram, which were done as recommended in patients with PJS,2 were normal. She underwent subtotal colectomy with ileo-colic anastomosis.

On follow up after a year, she was asymptomatic with normal hemoglobin and stool occult blood negative. After 2 years she presented with menorrhagia and lump in the lower abdomen. She was anemic, with stool occult blood positive. Upper GI endoscopy and colonoscopy were normal. CT scan showed left turbo-ovarian malignancy with multiple intramural rounded masses in the jejunum and ileum, probably polyps. Barium meal follow-through corroborated the CT findings in the duodenum and jejunum. She underwent laparotomy, hysterectomy with bilateral salpingo-oopherectomy; histology was reported as mucinous adenocarcinoma. In the same sitting an enterotomy and peroperative enteroscopy were done and 5 pedunculated polyps, >1.5 cm each, in the jejunum and duodenum were identified and polypectomy done; histology revealed adenomatous polyps.

On probing family history, her mother and maternal grandmother had had surgery for hematochezia before their demise. Her sister (age 27 years) and one daughter (age 6) also have characteristic perioral hyperpigmentation. The sister was also subjected to screening and was
found to have a single colonic polyp, which was removed; biopsy revealed adenomatous polyp with no dysplasia.

Classically the polyps in Peutz-Jegher syndrome are hamartomatous, but adenomas and adenocarcinomas are also reported. Hizawa et al described his experience with 75 GI polyps from 7 patients with PJS where 71 had hamartomatous polyps, 2 adenoma, 1 cancer in adenoma, and 1 pyogenic granuloma.

Small bowel polyps are treated with laparotomy with enteroscopy and polypectomy. Our patient had developed colonic and ovarian malignancies along with small bowel polyps; over just a two-year follow up, all of these were successfully treated. A hamartoma-adenoma-carcinoma sequence as been described in PJS.

References