

FROM THE RADIOLOGY- PATHOLOGY TEACHING FILE

Peutz-Jeghers Syndrome

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A 26-YEAR-OLD MAN WAS REFERRED TO MEMORIAL HOSPITAL for evaluation of gastrointestinal polyposis. He gave a history of episodes of crampy abdominal pain since childhood and recurrent upper and lower gastrointestinal bleeding since age 12 for which he required multiple transfusions. At ages 12 and 16, small intestinal polyps causing intussusception were resected. Family history was negative for gastrointestinal polyposis. The only pertinent findings on physical examination were macular, hyperpigmented areas on the face and lips (Figure 1). Laboratory studies revealed an iron deficiency anemia and intermittent occult blood in the stool.

Radiographic examination disclosed multiple polypoid lesions, both sessile and pedunculated, in the stomach, duodenum, mesenteric small intestine and colon. These varied in size and distribution, some appearing multilobulated, others as conglomerate masses, and were most numerous in the duodenum and jejunum (Figure 2). Four polyps were identified in the colon (Figure 3). None appeared ulcerated or umbilicated. There was no evidence of intestinal obstruction and the bowel between the lesions was normal.

Due to recurrent bleeding and suspicion of a malignant tumor, two subsequent surgical procedures were performed, at which time polyps were removed from the stomach, duo-

-denum, small intestine and colon.

The association of mucocutaneous pigmentation and gastrointestinal polyposis has become known as the Peutz-Jeghers syndrome. About half of all affected patients have a family history of the disease with an autosomal dominant inheritance pattern; the remaining cases are sporadic and non-familial. The mucocutaneous lesions appear as brown or black macules, usually 1-5 mm in diameter, and are seen most commonly on the lips and buccal mucosa. They are also frequently noted on the face and volar aspects of the palms and soles. Most patients become symptomatic during adolescence or early adulthood, presenting with pain secondary to intussusception or signs of gastrointestinal blood loss. Chronic blood loss anemia is common, massive hemorrhage rare.

The polyps, from a fraction of a millimeter to many centimeters in size, are small intestinal in 95 per cent of cases, colorectal in 30 per cent and gastric in 25 per cent. They are often innumerable in the small intestine; this results in an appearance which is described as "carpeting."

Histologically, the Peutz-Jeghers polyp is of the hyperplastic variety and is not adenomatous. There is no atypia or nuclear hyperchromatism such as is seen in the conventional intestinal polyp. The Peutz-Jeghers polyp (Figure 4) is an overgrowth of normal mucosa, often with prominent strands of muscularis mucosa pulled up with the glands. The involvement of the muscular wall, as in the illustration, suggests a hamartomatous process and the polyp, because of the apparent infiltration, is sometimes ill advisedly considered a malignant growth. These glands in the muscle, however, are typical and benign.

Gastrointestinal cancer, however, occurs with greater incidence in patients with this syndrome than in the general population, with carcinomas of the stomach, duodenum and colon accounting for most of the reported cases. An increased incidence of ovarian neoplasms has been found as well. Thus, management of these patients requires a high degree of vigilance for early detection of superimposed malignant change.

Since this paper went to press, a more detailed report on this case has been published in the New York State Journal of Medicine. See Paglia, M.A., Ghosh, B., Sherlock, P. and Kurtz, R.: Peutz-Jeghers Syndrome: surgical challenge, NY State J Med, 75:3, 402-405, 1975.

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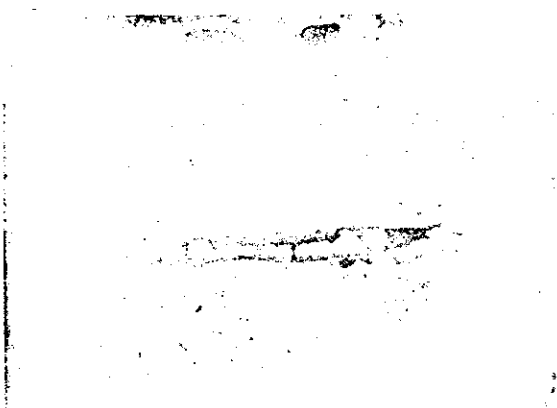


Figure 1. Hyperpigmented macules are present on the lips and circumorally.



Figure 2. Upper gastrointestinal and small intestinal examination reveals multiple polyps (some indicated by arrows) in the stomach, duodenum and jejunum.



Figure 3. Four polyps (arrows) are present in the colon.



Figure 4. A Peutz-Jeghers polyp in the jejunum, the commonest site of involvement in the intestinal tract. Note the presence of glands deep in the muscle and having the appearance of a hamartoma. The more peripheral part is simply an irregular overgrowth of benign mucosa.