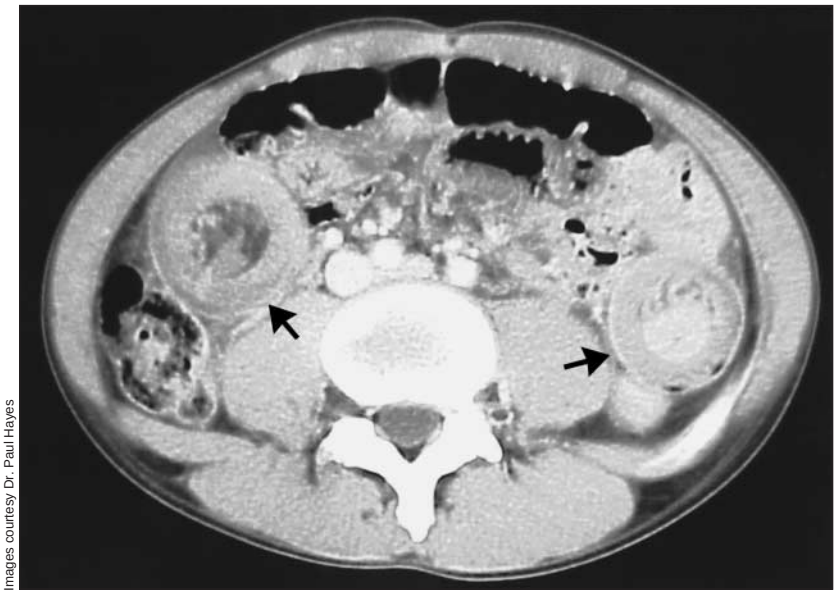
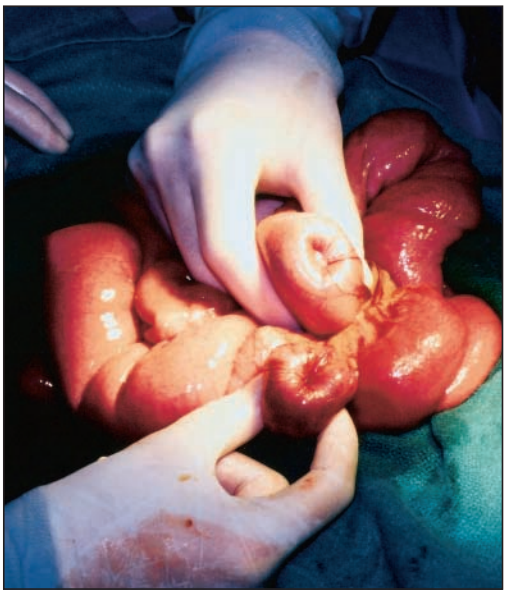
Intussusceptions in a man with Peutz–Jeghers syndrome





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A 27-year-old man presented with a 12-hour history of vomiting and nonradiating midabdominal pain. He had not undergone abdominal surgery and was previously healthy except for Peutz–Jeghers syndrome (PJS), diagnosed in his late teens after hamartomatous polyps were detected during a screening colonoscopy. His abdomen was diffusely tender, and a mass was felt in the right lower quadrant. Perioral pigmentation of his skin and oral mucosa was noted incidentally. His vital signs were normal except for a slightly elevated temperature (37.8°C) and leukocyte count (12.5×10^9/L). A CT scan of the abdomen showed 2 target lesions (Fig. 1, arrows), pathognomonic findings of intussusception.[1] At surgery 2 enteroenteric intussusceptions (ileoileal and jejunoileal) were discovered (Fig. 2). Both were resected with primary anastomoses. Pathological analysis of the resected specimens revealed characteristic branching hamartomatous polyps functioning as the lead points for each intussusception. The patient recovered and had no significant abdominal complaints in the following year.

PJS is an autosomal dominant inherited disorder characterized by mucocutaneous pigmentation, hamartomatous gastrointestinal polyps and cancer predisposition. Although Peutz first recognized the combination of pigmented lesions and gastrointestinal polyps in 1921, Jeghers is credited with providing the precise clinical description of the syndrome in 1944.[2] PJS is relatively rare, with an estimated incidence of 1 in 29 000 to 1 in 8300 live births.[3] Mutations in the *STK11/LKB1* serine /threonine kinase gene, located on chromosome 19p, have been identified in more than 50% of patients with PJS.[4] *STK11/LKB1* gene mutations are often unique to a particular family, making the routine identification of mutations difficult for families newly diagnosed with PJS.

For patients with PJS, the lifetime risk of mucocutaneous lesions approaches 100%, and the risk of polyps in the small intestine and colon is about 50% and 25% respectively. Histologically the pigmented lesions are characterized by increased melanocytes at the epidermal–dermal junction and increased melanin in the basal cells.[5] No malignant transformation has been attributed to these lesions,[5] which are predominantly located around the mouth, nostrils, eyes and perianal region.

Gastrointestinal manifestations of PJS include polyposis and recurrent abdominal pain secondary to intermittent intussusceptions and, less commonly, gastrointestinal bleeding. In the past, treatment focused on the removal of only those polyps causing recurrent intussusceptions, but current recommendations advocate prophylactic endoscopic removal of all polyps.[5] Recent guidelines support complete colorectal surveillance with either colonoscopy or flexible sigmoidoscopy and barium enema at 18 years of age and every 3 years thereafter.[6] Upper gastrointestinal surveillance by means of endoscopy is recommended every 1–2 years from age 25.[6] Others have advocated routine small bowel surveillance, including small bowel follow through every 2 years, with laparotomy and resection reserved for polyps greater than 1.5 cm in diameter.[5]

PJS predisposes to gastrointestinal carcinomas as well as cancer of the pancreas, breast, lung and reproductive organs.[5,7] In a series of 34 patients with PJS followed for a median of 20 years, one of these forms of cancer developed in 53% by 39.4 years of age on average.[3] The mean interval from initial diagnosis of PJS to the diagnosis of cancer was 19.8 years.[3] In another series,

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among 72 patients with PJS the relative risk of dying from a gastrointestinal cancer was 13-fold greater than that in the general population and the relative risk of dying of any cancer was 9-fold greater, with a 48% chance of dying of cancer by the age of 57.[7]

Recent advances, including genetic testing of family members to identify those who will require endoscopic surveillance,[6,8] and the introduction of guidelines for gastrointestinal surveillance should result in improved management of patients with PJS.

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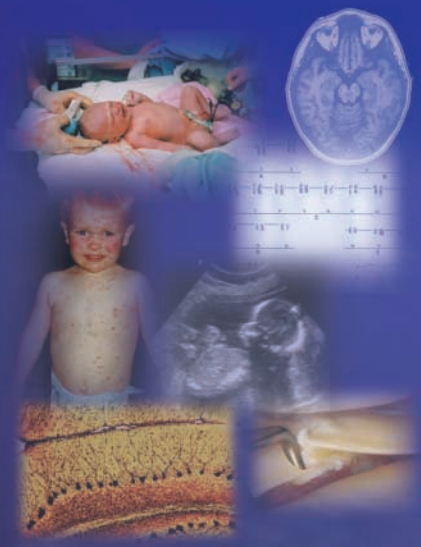

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