Our patient is a 20-year-old white female who presented with a two-year history of intermittent rectal bleeding and iron deficiency anemia. She had no significant family history. A gastroenterologist initially evaluated her two years prior to the current presentation. In preparation for a scheduled colonoscopy at that time, she left the facility against medical advice because of difficulty in obtaining intravenous access. She presented herself more recently to the senior author at the behest of her mother. On examination in the office, bright red blood was found during the digital exam. Subsequent anoscopy revealed a probable villous adenoma. Colonoscopy revealed numerous colonic polyps, both pedunculated and sessile, ranging from 1.5–5 cm in diameter. The polyps were distributed along the entire length of the colon and they were separated by grossly normal appearing mucosa. A few of the polyps demonstrated active bleeding during the examination. Biopsy of two of the polyps showed moderate to marked dysplasia. The final pathologic examination revealed the diagnosis of juvenile polyposis. Esophagogastroduodenoscopy was negative for any abnormalities.

The patient was advised to undergo a hand-assisted laparoscopic subtotal colectomy with ileorectostomy (Figure 1). During the procedure, the patient was noted to have a Meckel’s diverticulum, which was removed, endometriosis of both ovaries, and uterine didelphica. The final pathology examination revealed dysplasia ranging from mild to severe in multiple polyps (Figure 2, 3). There was no evidence of adenocarcinoma. Ninety-six lymph nodes were negative for neoplasm. The Meckel’s diverticulum was found to contain ectopic gastric and pancreatic tissue.

The patient was discharged on the fourth post-operative day in excellent condition. Her final diagnosis was Juvenile Polyposis Coli. The sole sister and both parents were found to be negative for any polyloid disease. Her checkups at 3 and 12 months showed small rectal polyps, which were snared. Pathological examination again confirmed benign juvenile polyposis.

DISCUSSION

Juvenile Polyposis Syndrome (JPS) is uncommon and can be separated into three distinct entities: diffuse juvenile polyposis of infancy, diffuse juvenile polyposis, and juvenile polyposis coli, the diagnosis of our patient (1).

Juvenile Polyposis Syndrome was first described by McColl and colleagues in 1964 (2). It is inherited as
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Figure 1 (a, b). Gross examination of the resected colon revealed numerous polyps, like the ones pictured, mostly pedunculated and ranging in size from 1.5–5 cm. These polyps have an irregularity in their contour not consistent with Sporadic Juvenile Polyps.

an autosomal dominant trait. The incidence of JPS is estimated to be anywhere from one in 16,000 to one in 100,000 (3). Approximately 50%–80% of patients with this syndrome acquire it spontaneously (4). The syndrome begins in the first two decades of life, with a mean age of diagnosis at 18.5 years (5). It is characterized by four or five polyps to well over one hundred hamartomatous polyps throughout the gastrointestinal tract. Histologically, juvenile polyps are characterized by an abundant lamina propria that lacks smooth muscle proliferation and contains dilated mucin-filled glands lined by normal epithelium. While these polyps occur most often in the colon and rectum, they have also been described in the stomach and small intestine. Diagnosis is based upon one of three conditions: (a) more than five juvenile polyps of the colorectum, (b) juvenile polyps throughout the gastrointestinal tract, and (c) any number of juvenile polyps with a family history of juvenile polyps (6).

Patients with JPS usually present with anemia secondary to hemochezia. Bleeding most likely results from abrasion of the excess epithelium by the passage of stool. Other findings may include the passage of sloughed tissue, abdominal pain, prolapsed polyps, and intussusception. In addition, JPS can be associated with a number of extraintestinal manifestations. Reports describe congenital anomalies in 11% to 15% of JPS patients (5), with most of them occurring in the sporadic cases. These anomalies include ganglioneuromatous proliferation, arteriovenous manifestations, porphyria, psoriasis, mental retardation, congenital heart disease, cleft lip/palate, epilepsy, hereditary hemorrhagic telangiectasia, digital clubbing, hypertrophic pulmonary osteoarthropathy, and malrotation of the gut (5,7).

Families with JPS have a higher incidence of gastrointestinal cancers (8). Howe and colleagues reviewed the medical records of 133 familial JPS patients. Fifty-nine of the 133 cases developed GI malignancies: colorectal [42], gastric [15], pancreatic [1], and duodenal [1] (9).

Two chromosomes, 10q and 18q, have been shown to be linked to JPS. The SMAD4/DPC4 gene on 18q21 is thought to have a mutation hotspot where it undergoes a four base pair deletion in the SMAD4 exon 9 (10–12). The gene produces a protein that mediates in association with SMAD2 and SMAD3, signaling for transforming growth factor—beta (TGF-[B]) from the cell surface to the nucleus. This mechanism is vital for cell development and regulation of cell growth (13). Somatic deletions at 10q22 also have been found in over 80% of juvenile polyps in inheritable or sporadic conditions (14). PTEN (phosphate with tensin homology) on chromosome 10q22 also appears to play an important role. Unlike SMAD4, PTEN, a protein tyrosine phosphatase, is a tumor suppressor gene and has
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numerous roles (15). Some authors, however, believe that patients with PTEN deletion should be considered as having Cowden syndrome, with an increased risk of breast and thyroid cancer, and hamartomas being one part of the syndrome (16).

While JPS is a rare syndrome, it is necessary to appreciate its association with an increased risk of GI malignancies. It is also necessary to distinguish it from sporadic juvenile polyps, which are usually benign in nature and are of little clinical significance to the patient. It is the authors’ opinion that families with a history of JPS and cases of undetermined rectal bleeding should be carefully evaluated with colonoscopy. Furthermore, because of its association with upper gastrointestinal malignancies, patients should also undergo esophagogastroduodenoscopy.

References