IMAGE | PEDIATRICS

Juvenile Polyposis of Infancy Presenting as Protein-Losing Enteropathy

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CASE REPORT

A 20-month-old female was admitted with rectal bleeding and rectal prolapse with extrusion of a polyp. Her medical history was relevant for chronic diarrhea, abdominal distension since birth, delayed neuropsychomotor development, and refractory iron deficiency anemia. Physical examination revealed macrocephaly, skin pallor, severe malnutrition with significant muscle atrophy, and generalized edema. Laboratory workup demonstrated severe anemia and hypoalbuminemia. Subsequent abdominal scintigraphy with Tc99m marked albumin revealed intestinal protein loss to some degree in the small bowel and severe colonic loss. Endoscopic examination showed diffuse severe polyposis throughout the colon (Figure 1). The patient underwent total colectomy with the creation of an ileostomy which significantly improved the protein-losing enteropathy and allowed for nutritional rehabilitation (Figure 2).
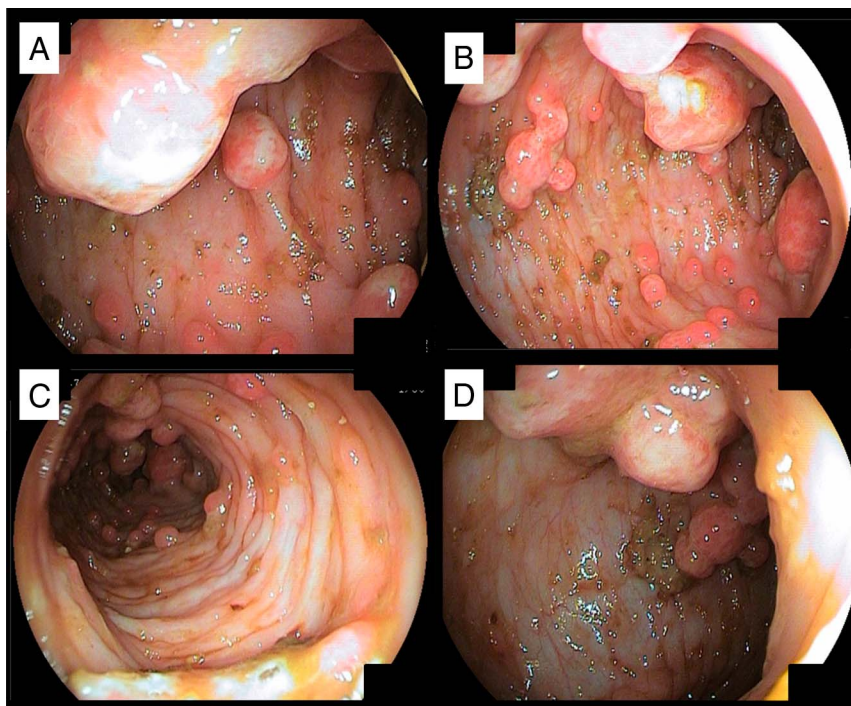

Figure 1. Severe diffuse colonic polyposis (A and B) ascending colon, (C) transverse colon, and (D) descending/sigmoid.
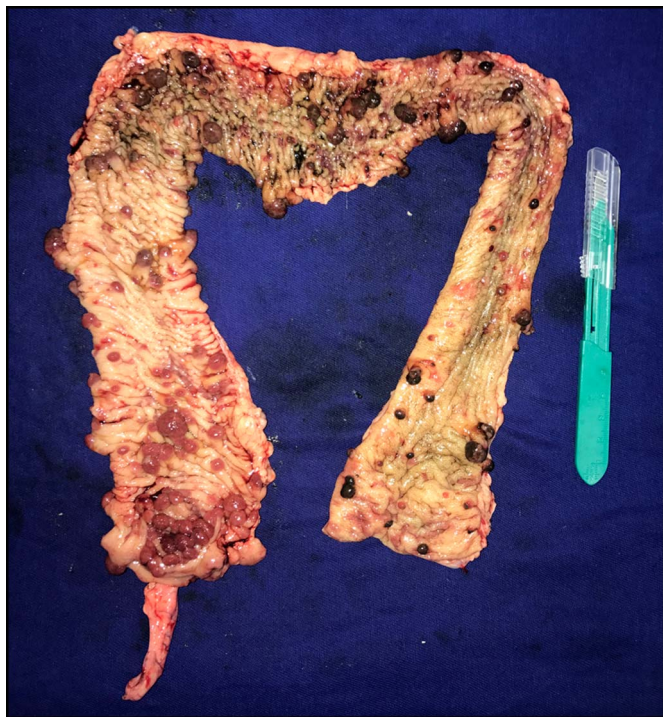
Figure 2. Macroscopic view of the colon after total colectomy.

Juvenile polyposis syndrome (JPS) is a rare autosomal dominant condition with an estimated prevalence of 1/100,000.[1] Juvenile polyposis of infancy is considered the most severe form of JPS. This generalized, early-onset form of JPS manifests with diarrhea, anemia, gastrointestinal bleeding, rectal prolapse, intussusception, and protein-losing enteropathy—as illustrated in this case.[1,2] Depending on the magnitude of protein loss, colectomy may be required to treat protein-losing enteropathy.

The use of sirolimus has also been reported as a management strategy for juvenile polyposis of infancy.[3]

DISCLOSURES

REFERENCES

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