



DEPARTMENT OF PATHOLOGY

Short Report in Pathology

Organ system: Gastrointestinal Tract

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History:

A 67-year-old female presented to the emergency department with abdominal pain, vomiting and constipation for the past 5 days. Imaging study demonstrated a 4 cm mass causing small bowel obstruction. A small bowel resection was done.

Microscopic Images:

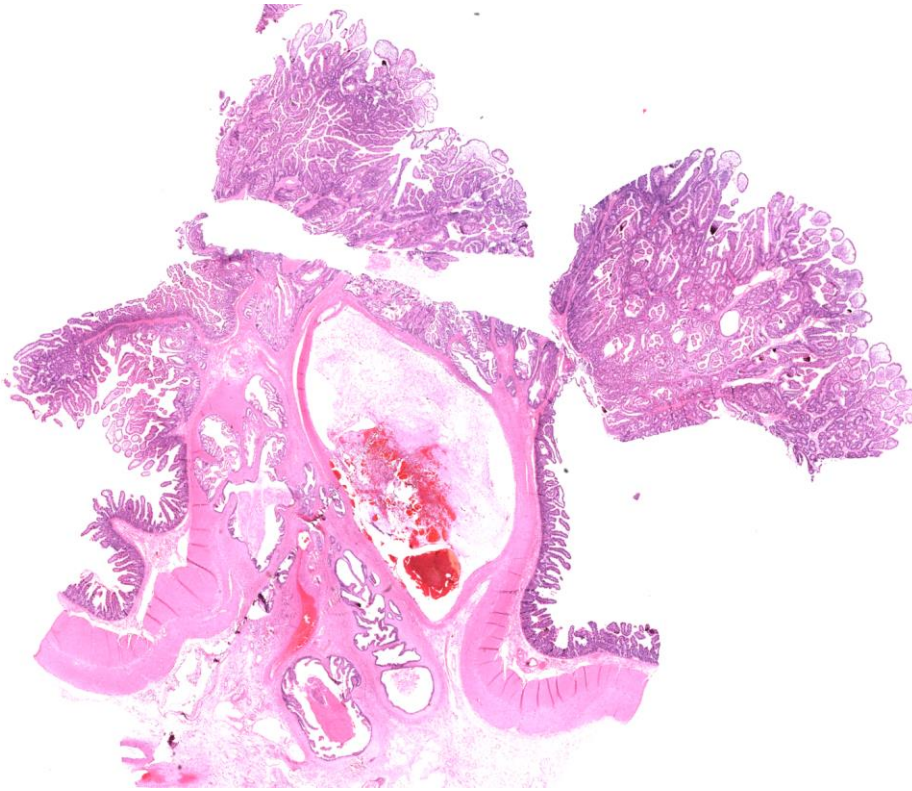


Figure 1. At low magnification, the tumor shows polypoid and pedunculated configuration with a multilobulated surface showing distinctive papillary villous architecture with tree-like arborization of smooth muscle bundles. Large and cystically dilated glands are noted at the base of this polypoid mass, intruding through the small intestinal wall into the sub-serosal adipose tissue (Hematoxylin and Eosin stain, 20x).

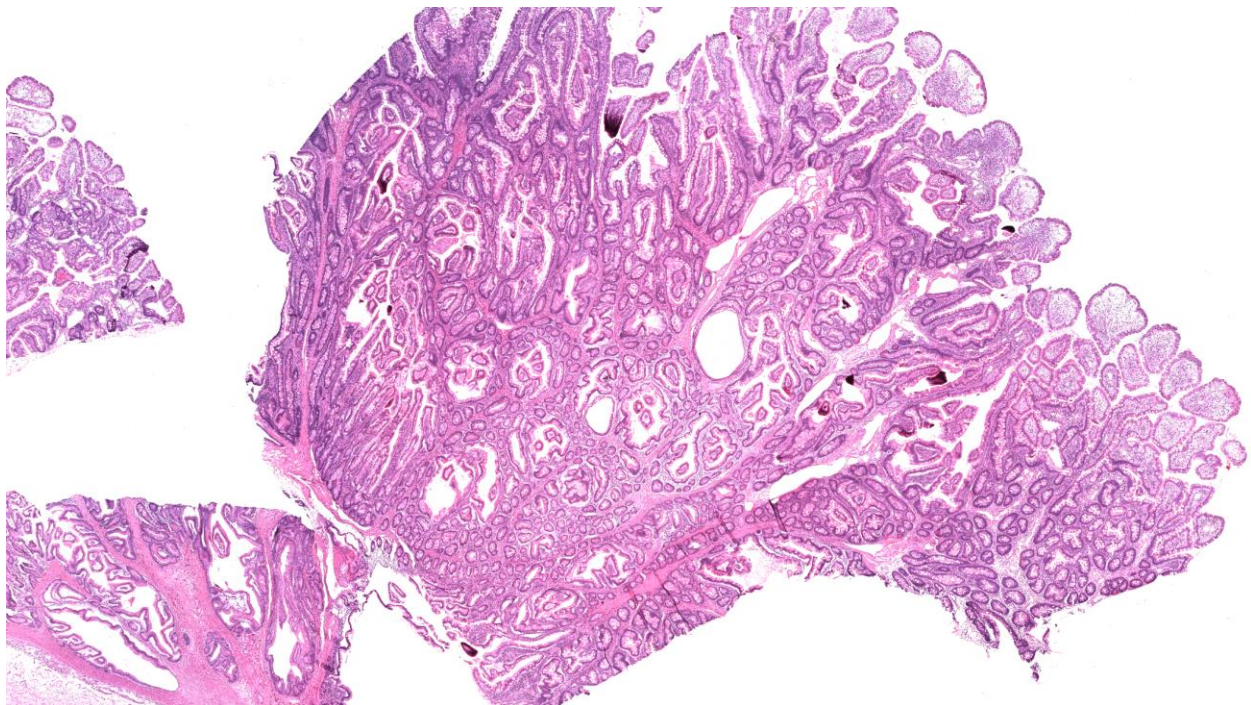


Figure 2. The surface portion of tumor shows epithelium with glandular dilatation and distortion. The epithelial component is arranged in a lobular configuration because of the separation by bundles of smooth muscles. There is no apparent dysplasia. (Hematoxylin and Eosin stain, 100x)

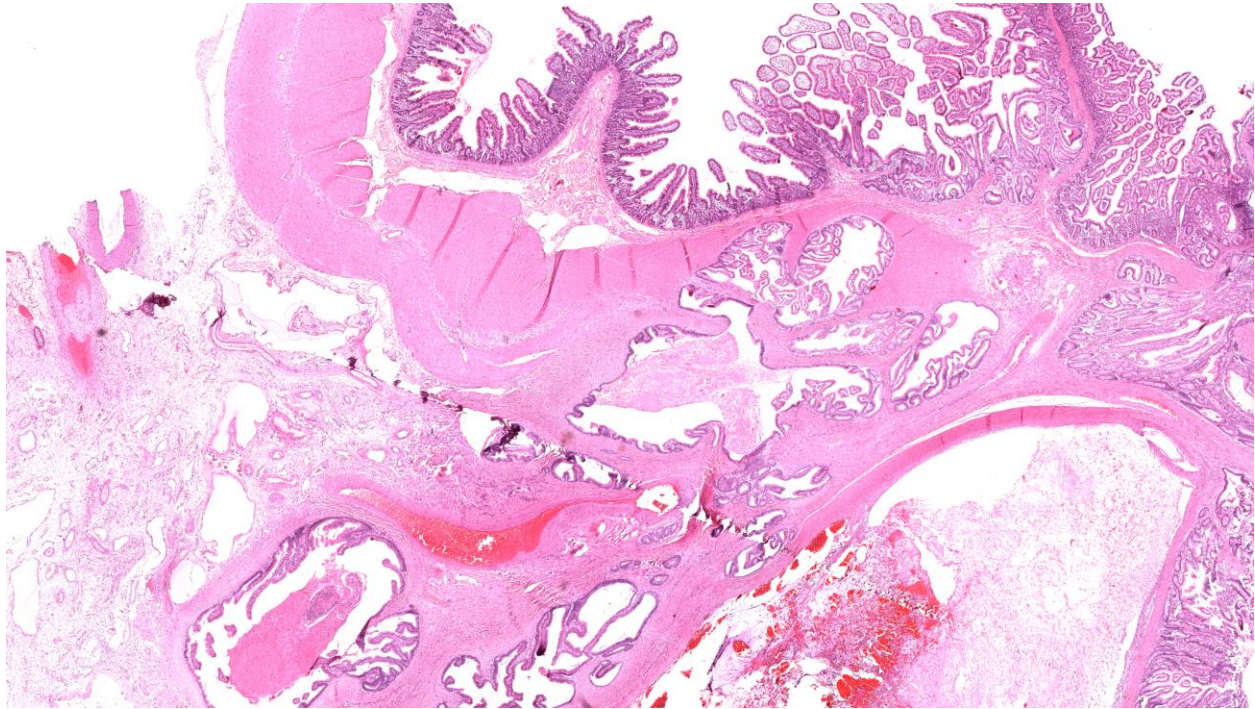


Figure 3. At the base of the tumor, there are irregular and cystically dilated glands intruding through the muscularis propria into the sub-serosal adipose tissue. The epithelial lining of these cystically dilated glands are relatively bland and have no apparent dysplasia (Hematoxylin and Eosin stain, 20x).

Immunohistochemistry:

Not done for this case

Differential diagnoses:

1. Invasive adenocarcinoma arising from tubulovillous adenoma of the small intestine
2. Tubulovillous adenoma of the small intestine with prolapse related changes
3. Invasive adenocarcinoma arising from Peutz-Jeghers polyp
4. Peutz-Jeghers polyp with pseudoinvasion due to prolapse related changes

Diagnosis:

Peutz-Jegher polyp with pseudoinvasion due to prolapse related changes

Discussion:

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant disorder characterized by the development of hamartomatous polyps, i.e. Peutz-Jeghers polyps (PJPs), in the gastrointestinal tract and mucocutaneous pigmentation. PJS is associated with a significantly increased risk of malignancy in other organ systems including gynecological system, pancreas and testis etc. WHO criteria for PJS includes: (1) ≥ 3 histologically confirmed PJPs, or (2) Any number of PJPs in a patient with family history of PJS, or (3) mucocutaneous pigmentation in a patient with family history of PJS, or (4) Any number of PJPs in a patient with prominent mucocutaneous pigmentation.

Approximately 75% of PJPs are syndromic and associated with autosomal dominant germline mutation of *STK11/LKB1*. Approximately ~25% are sporadic. Approximately 64% of PJPs occur in the small bowel, ~53 in the colon, and ~20% in the stomach. Histopathologically, PJP is characterized by compactly spaced glands supported by arborizing framework of well-developed smooth muscle that is contiguous with muscularis mucosae, resulting in a characteristic lobulated appearance. Dysplasia is rare, but syndromic patients have higher risk for dysplasia and malignancy elsewhere. Large PJPs in small bowel and colon are more characteristic, but gastric PJP are indistinguishable from hyperplastic polyps or other syndromic gastric hamartomatous polyps.

When PJPs are large, the prolapse related changes can cause glands misplacement, i.e. pseudoinvasion. The pseudoinvasion is characterized by the presence of irregular glandular structures surrounded by lamina propria. In contrast, true invasion is associated with desmoplasia and haphazard and infiltrative growth pattern. The distinction of pseudoinvasion versus true invasion in PJPs is crucial to prevent unnecessary radical surgeries or overtreatment.

References:

1. Tacheci I, Kopacova M, Bures J. Peutz-Jeghers syndrome. *Curr Opin Gastroenterol*. 2021 May 1;37(3):245-254
2. Giardiello FM, Trimbath JD. Peutz-Jeghers syndrome and management recommendations. *Clin Gastroenterol Hepatol*. 2006 Apr;4(4):408-15.
3. Latchford AR, Neale K, Phillips RK, Clark SK. Peutz-Jeghers syndrome: intriguing suggestion of gastrointestinal cancer prevention from surveillance. *Dis Colon Rectum*. 2011 Dec;54(12):1547-51.