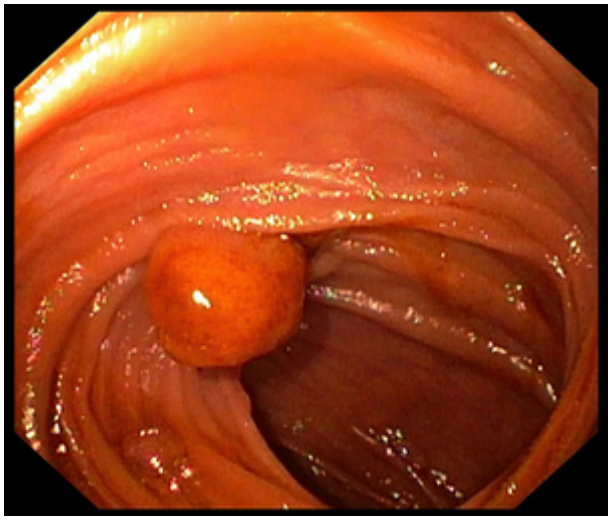
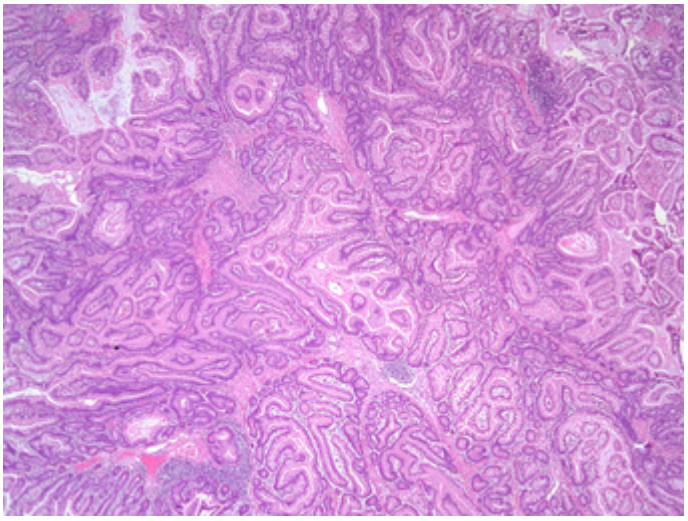
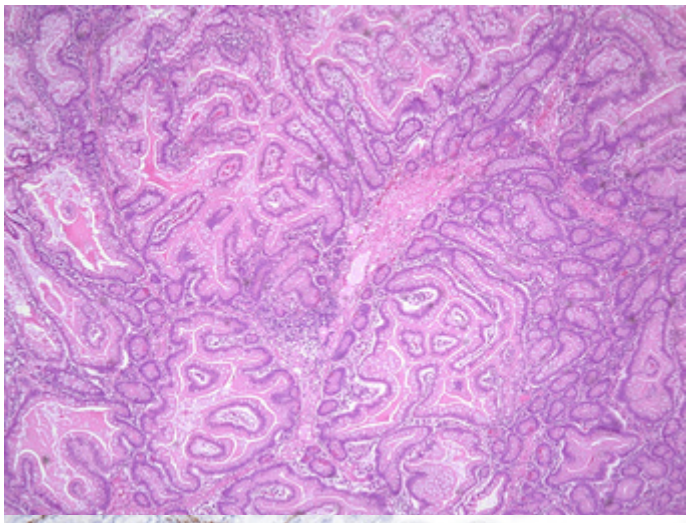
July 2019

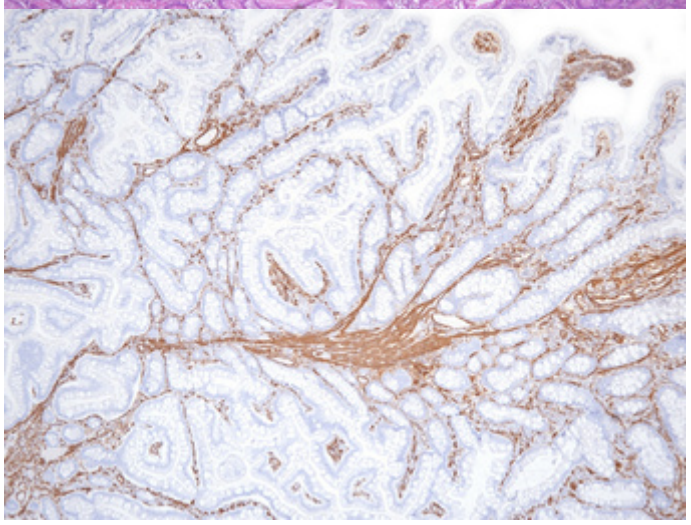
Duodenal polyp in a 59-year-old female.

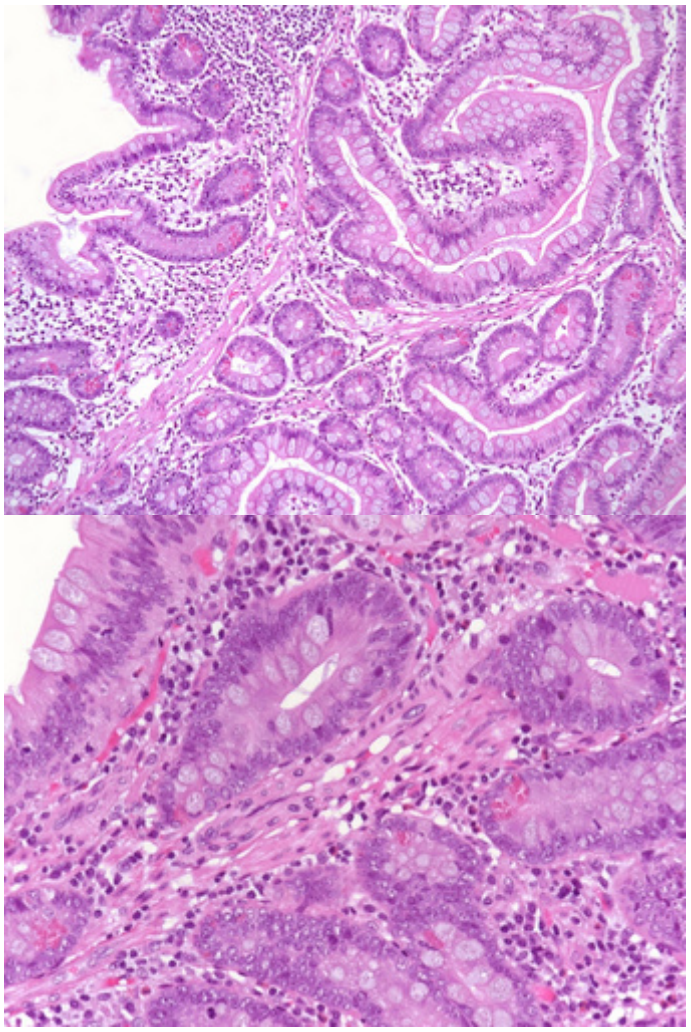
What is your diagnosis?











Diagnosis:

Peutz-Jeghers polyp.

Comment:

A solitary pedunculated polyp with coarsely lobulated nodular surface was removed from the duodenum by snare polypectomy (Panel A). At low magnification, histology discloses a complex villous architecture with arborizing smooth muscle core (Panels B and C), which is thicker centrally and can be highlighted by smooth muscle actin (SMA) immunohistochemistry (Panel D). On high power, the epithelium appears slightly hyperplastic, yet not dysplastic, the lamina propria being normal, that is, not expanded and without inflammation (Panels E and F). A final diagnosis of Peutz-Jeghers polyp was made.

Peutz-Jeghers polyps may arise sporadically or within Peutz-Jeghers syndrome (PJS), an autosomal dominant syndrome that is characterized by melanocytic mucocutaneous hyperpigmentation and distinctive hamartomatous gastrointestinal polyposis. The hyperpigmentation is present in childhood as brown macules around the mouth, eyes and nostrils, but also in the perianal area, on the buccal mucosa and on the fingers. Lesions tend to fade in puberty and adulthood. Polyps may occur throughout the entire gastrointestinal tract but are most common in the small bowel (65-90%), followed by colon (60%) and stomach (20-50%).

According to the WHO classification, diagnostic criteria for PJS are the following:

1. ≥ 3 histologically confirmed Peutz-Jeghers polyps
2. Any number of Peutz-Jeghers polyps with a family history of PJS
3. Characteristic, prominent mucocutaneous pigmentation with a family history of PJS
4. Any number of Peutz-Jeghers polyps and characteristic, prominent mucocutaneous pigmentation

The role of the pathologist is to make the diagnosis of Peutz-Jeghers type polyp and to raise the possibility of Peutz-Jeghers syndrome if the clinical context is compatible.

In the small bowel, the histological diagnosis is usually straightforward, because polyps in this location usually show the distinctive histological features. However, gastric Peutz-Jeghers polyps often lack specific histology and are often not readily distinguishable from gastric juvenile polyps or sporadic hyperplastic polyps. Without knowledge of the clinical context, one should therefore be cautious about establishing a new diagnosis of PJS on the basis of gastric polyps alone. In the large bowel, Peutz-Jeghers-type polyps resemble mucosal prolapse polyps. The context is essential to favor a hamartomatous polyp over a prolapse polyp (age, polyp location, number of polyps, and other pathology).

Pathologists need to be aware of the following important diagnostic pitfall: Epithelial misplacement, due to prolapse and peristaltic kneading, is relatively common in Peutz-Jeghers polyps (particularly in small bowel lesions) and may extend throughout the muscularis propria into the serosa, mimicking a well-differentiated invasive adenocarcinoma.

A broad range of malignancies has been associated with PJS. Cumulative cancer risk has been estimated to be 76% to 93%, with a median age at first cancer diagnosis of 45 years. Cancer risks by organ are 32-54% for breast carcinoma, 39% for colorectal carcinoma, 29% for gastric carcinoma, 13% for small intestinal carcinoma, and 11-36% for pancreatic carcinoma. Other well documented extraintestinal tumours include carcinomas of the lungs, uterus and ovaries, as well as otherwise rare gonadal lesions, including sex cord tumour with annular tubules of the ovary and Sertoli cell tumour of the testis.

For further reading:

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