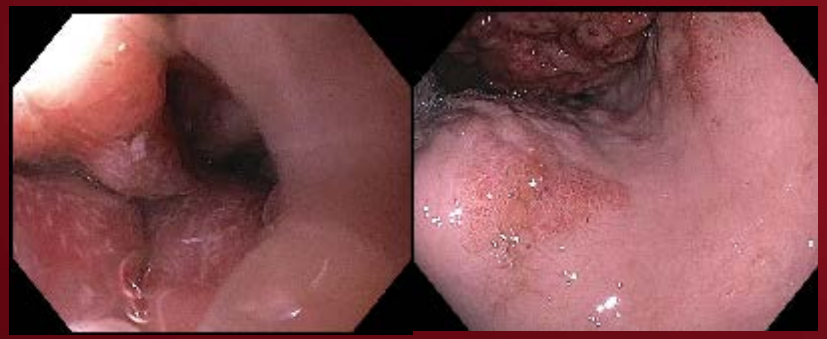
Gastrointestinal Bleeding In The Setting Of Juvenile Polyposis Syndrome Due To *SMAD4* Mutation

Paula Marincola Smith[1] *MD, PhD,* Marcus C.B. Tan[2] MBBS

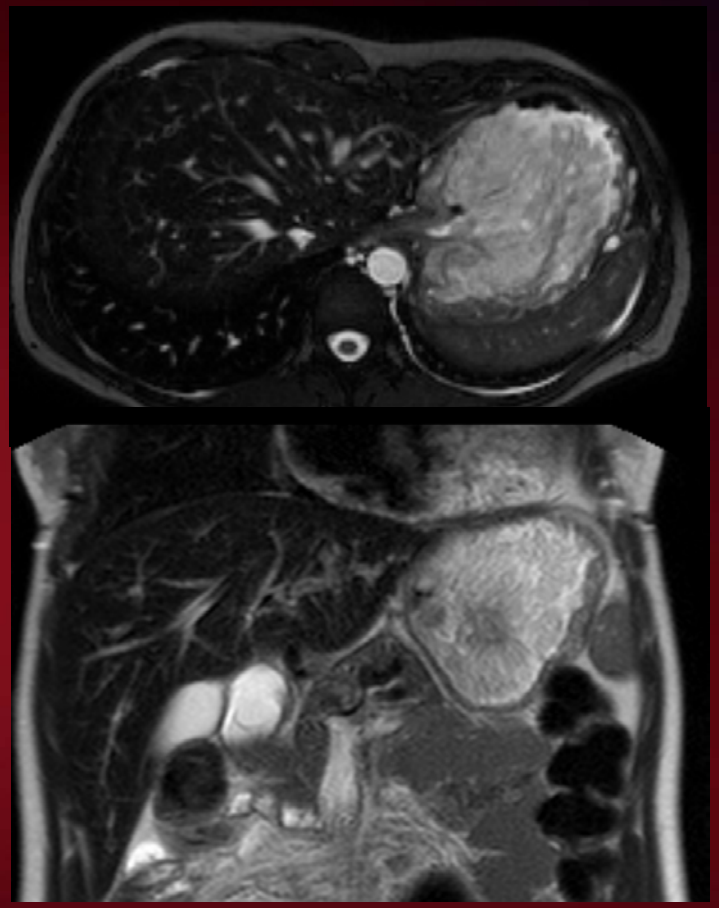
VUMC Departments of [1]*General Surgery,* [2]*Surgical Oncology & Endocrine Surgery*

A 27-year-old female presented at 13 weeks' gestation with epigastric pain and anemia.

- Upper endoscopy revealed a giant circumferential polyp in the proximal stomach.
- MRI showed a 9.6 x 11.6cm mass without evidence of locoregional lymphadenopathy or metastatic tumors.
- Biopsies revealed hyperplastic mucosa with frequent eosinophils in the lamina propria

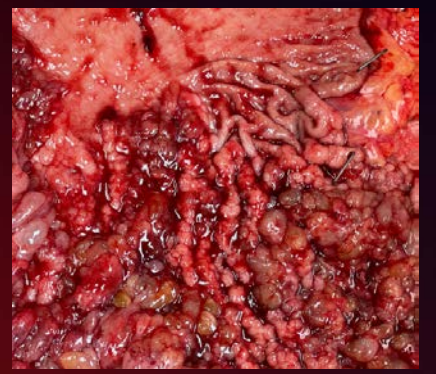


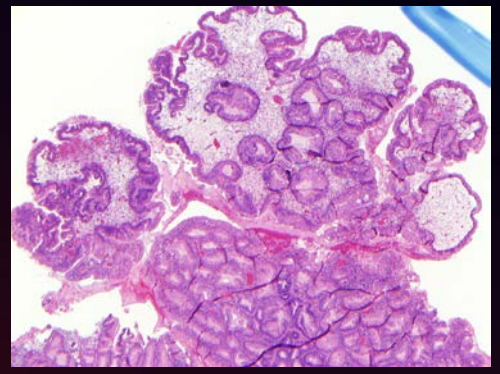
She was supported with intermittent blood transfusions until 34 weeks' gestation, then labor induced. Six weeks after uneventful delivery, she underwent total gastrectomy with jejunal pouch reconstruction.



Final surgical pathology revealed multiple hyperplastic polyps, negative for dysplasia or invasive cancer.

Germline testing revealed monoallelic mutation of the SMAD4 gene and a diagnosis of Juvenile Polyposis Syndrome (JPS) was made.





JPS is associated with

- Colorectal cancer (70% lifetime risk)
- Gastric cancer (20% lifetime risk)
- Hereditary Hemorrhagic Telangiectasia (HHT)

Follow-up colonoscopy revealed multiple polyps (both hyperplastic and adenomatous), for which she will need lifelong surveillance.

This case highlights the importance of coordination of care and genetic testing for unusual tumors in young adults.