Late-Onset Juvenile Polyposis and Hereditary Hemorrhagic Telangiectasia Overlap Syndrome

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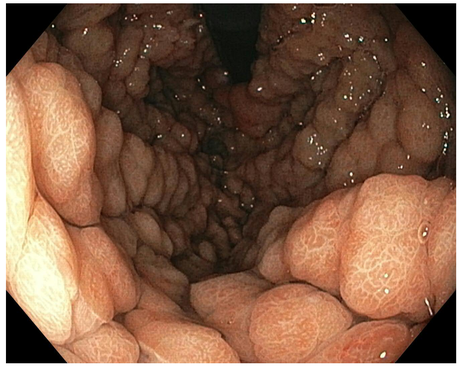
INTRODUCTION

- Juvenile Polyposis Syndrome (JPS) and Hereditary Hemorrhagic Telangiectasia (HHT) are rare disorders inherited in an autosomal dominant manner. The SMAD4 gene mutation causes a combined syndrome of JPS and HHT.
- We report a case of late-onset JPS-HHT overlap syndrome.

CASE PRESENTATION

- A 72-year-old man presented with orthostasis and melena for three days. He had a lifetime history of recurrent epistaxis.
- Family history revealed two daughters with a history of gastrointestinal (GI) bleeding due to arteriovenous malformations (AVMs) and had required surgical gastrectomies for polyposis due to SMAD4 gene mutation.
- Physical exam revealed mild tachycardia. Rectal exam showed melena. There were no mucocutaneous telangiectasias. Hemoglobin (Hb) level was 4.4 g/dL and he had an elevated BUN to creatinine ratio. He was resuscitated and transfused.
- Upper endoscopy revealed multiple non-bleeding semi-sessile polyps in the stomach (Figure 1A). Colonoscopy was unremarkable and CT angiography showed no active bleeding.
- Capsule endoscopy a week later displayed numerous small bowel angiectasias that were subsequently treated with argon plasma coagulation (Figure 1B).
- Gastric biopsies showed foveolar hyperplasia and edema of the lamina propria consistent with inflammatory polyp of JPS (Figure 1C).
- He met the diagnostic criteria for the JPS-HHT overlap syndrome. He declined genetic testing for SMAD4 mutation.



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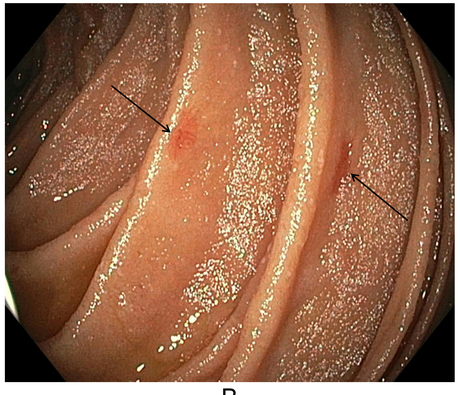

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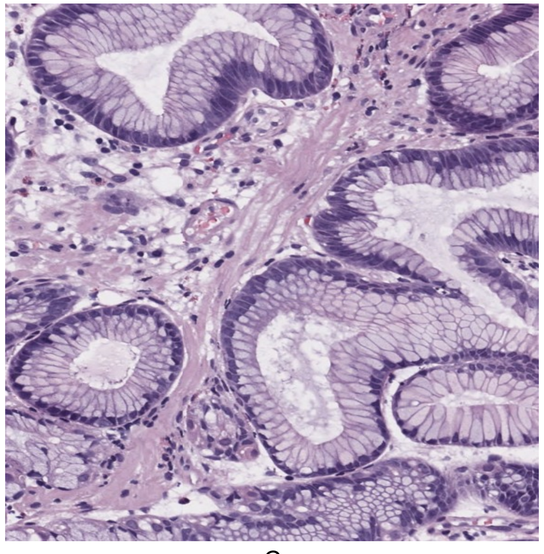

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Figure 1: A. Upper GI endoscopy showing multiple non-bleeding semi-sessile polyps in the stomach. **B.** Small Bowel Enteroscopy showing angiodysplastic lesions in the small intestine. **C.** Gastric biopsy showing foveolar hyperplasia and edema of lamina propria, consistent with inflammatory polyp of JPS.

CASE PRESENTATION

- Follow-up included oral iron supplementation, and Hb level monitoring.
- Screening for GI malignancy as well as pulmonary and central nervous system AVMs was recommended.

DISCUSSION

- JPS often comes to clinical attention by age 20 years with most presenting with bleeding or anemia due to GI polyps.
- Individuals with JPS due to SMAD4 mutations often exhibit features of HHT [1] with GI bleeding related to HHT often presenting in the 4th decade of life.
- Our patient had delayed onset penetrance of the GI manifestations of both HHT and JPS with a first GI bleed at age 72 years.
- It is important for gastroenterologists to be aware of the varied age-related GI manifestations of the JPS-HHT overlap syndrome.

REFERENCES

[1] Lin HC, Fiorino KN, Blick C, Anupindi SA. A rare presentation and diagnosis of juvenile polyposis syndrome and hereditary hemorrhagic telangiectasia overlap syndrome. Clin Imaging. 2015 Mar-Apr;39(2):321-4.

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No conflicts of interest to disclose