

Bloody Diarrhea

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Case Presentation

- 61yr-old-woman, Libyan origin
- Past medical history
 - B12 deficiency
- Family history
 - unremarkable

Clinical Presentation

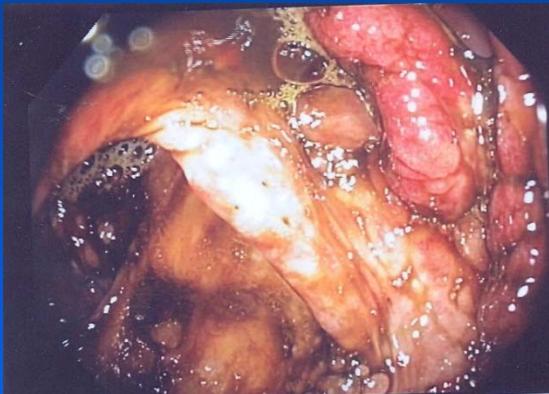
- Bloody Diarrhea
- Vomiting
- Physical examination
 - Unremarkable

Labs

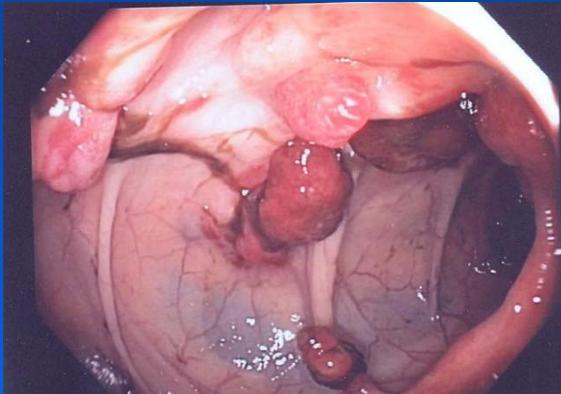
Hb (14-18g/dL)	10.7
Ferritin (30-400ng/ml)	6
Iron (40-145mcg/dL)	43
Transferrin (200-360mg/dL)	261

Colonoscopy

- Multiple huge polyps



Right colon

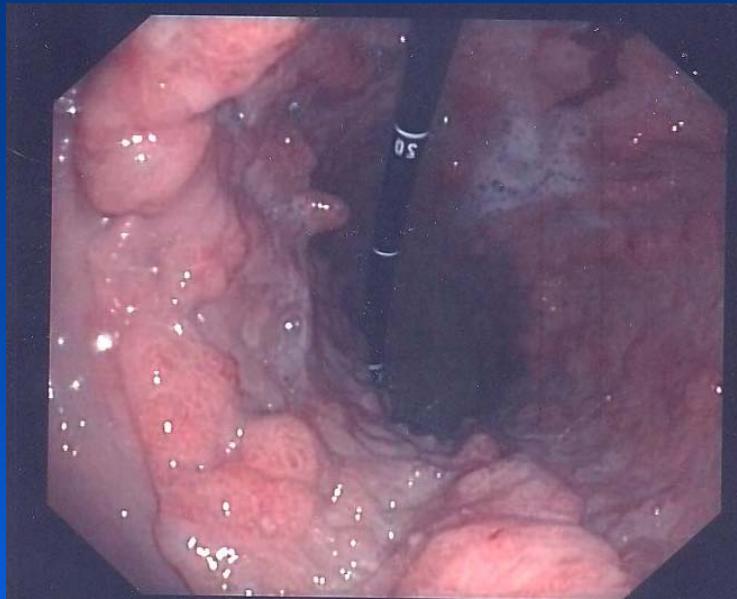


Splenic flexure



Rectum

Gastroscopy

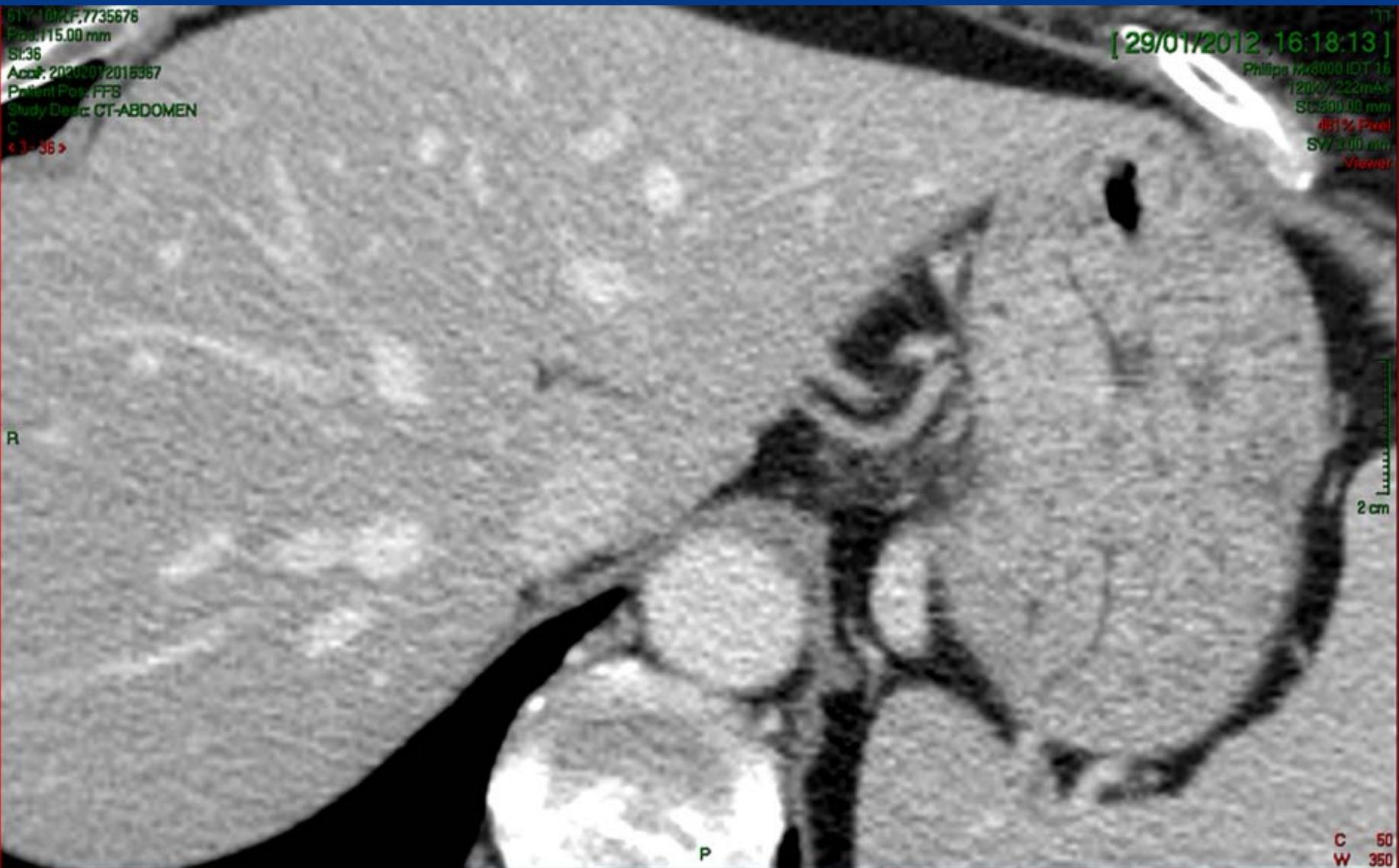


CT

61Y-10M/F,7735676
Fidc:15.00 mm
SI:36
Acq#:2020012015367
Patient Pos: PFS
Study Desc: CT-ABDOMEN
C
<3-36>

[29/01/2012, 16:18:13]

Philips MX3000 IDT 16
120kV, 222mAs
Sc300.00 mm
481 % Pixel
SW:300 mm
Viewport



VCE

00:00:00

10 Nov 11

GE



PillCam® SB

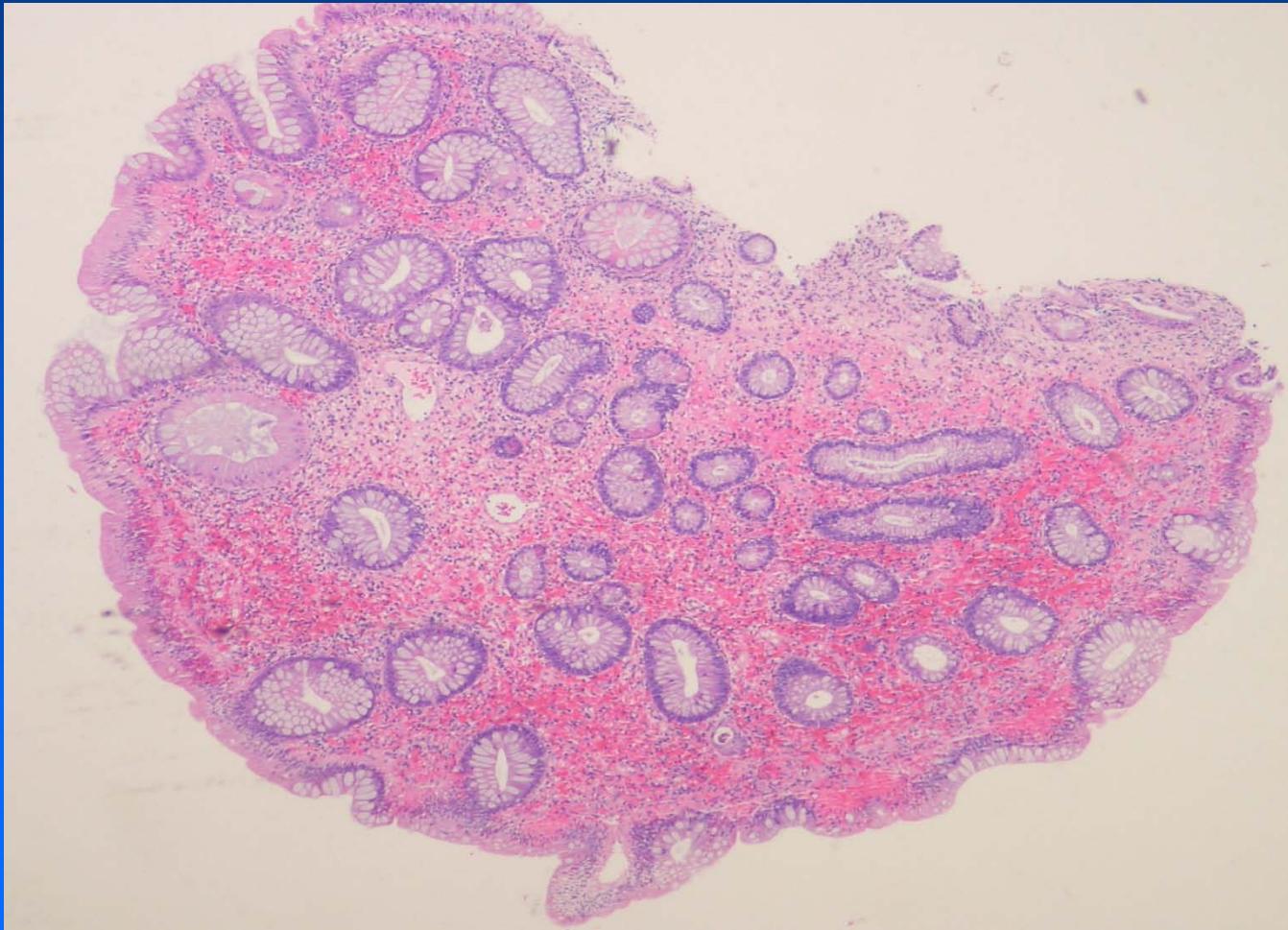
VCE



Summary

- 61yr-old woman
- Bloody diarrhea
- Iron deficiency anemia
- Carpeting gastric polyposis
- Huge multiple colonic polyps
- One jejunal polyp

Pathology

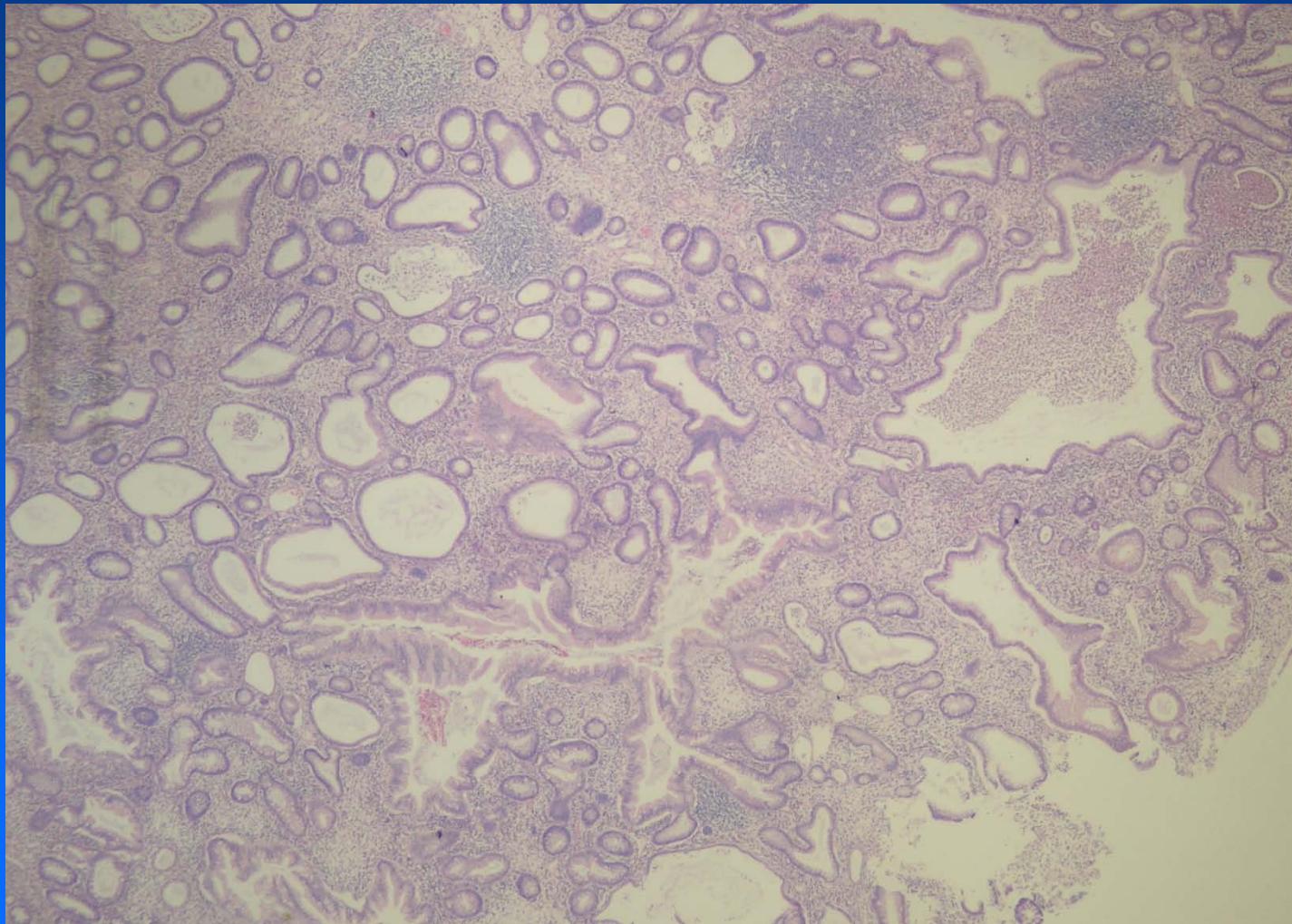


Pathology



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Pathology



Juvenile Polyposis Syndrome

- Incidence: 1:100,000
- Autosomal dominant
- 75% - a family history
- 25% - *de novo* mutations

JPS - Genetics

- 60% - germline mutation:
 - *SMAD4 / BMPR1A*: 18q21.1 / 10q22.3
 - BMP/TGF-beta pathway
- 40% - no mutation:
 - PTEN?
 - Endoglin?

JPS – Clinical Diagnosis

- Any of the following three criteria are met:
 - 1. Multiple colonic hamartomatous polyps
 - 2. Hamartomatous polyp + family history
 - 3. Extracolonic hamartomatous polyps
- phenotype-genotype correlation:
 - *SMAD4* - massive gastric polyposis

Giardiello FM, et al. Arch Dis Child 2001

JPS – Clinical Manifestations

- Often silent
- Age dependent
- Co-occur with HHT
- A risk of an associated malignancy

JPS – Malignancy Risk

- A cumulative life-time risk for CRC - 39%
 - R.R – 34 *Brosens LA et al , Gut 2007*
- The incidence of gastric adenocarcinoma – 21%
- Gastric & SB ca. – 1/5 the frequency of CRC

Brosens LA et al, World J Gastroenterol 2011

What is The Clinical Management?

- Polypectomy?
- Colectomy?
- Gastrectomy?
- Only surveillance?

Treatment

- Total proctocolectomy with IPAA
- Frequent post-operative endoscopic surveillance



Future management

- Subtotal gastrectomy
- Endoscopic surveillance
- Sulindac?
 - COX-2 expression is higher in JPS polyps
 - Sulindac had prevented polyp development in the pouch

Take Home Message

- One intact polyp can be more informative than multiple biopsies
- VCE - has a role in SB disease evaluation

Thank you