FAMILIAL ADENOMATOUS POLYPOSIS: NOT ALWAYS A FAMILIAL CONDITION

A CASE STUDY

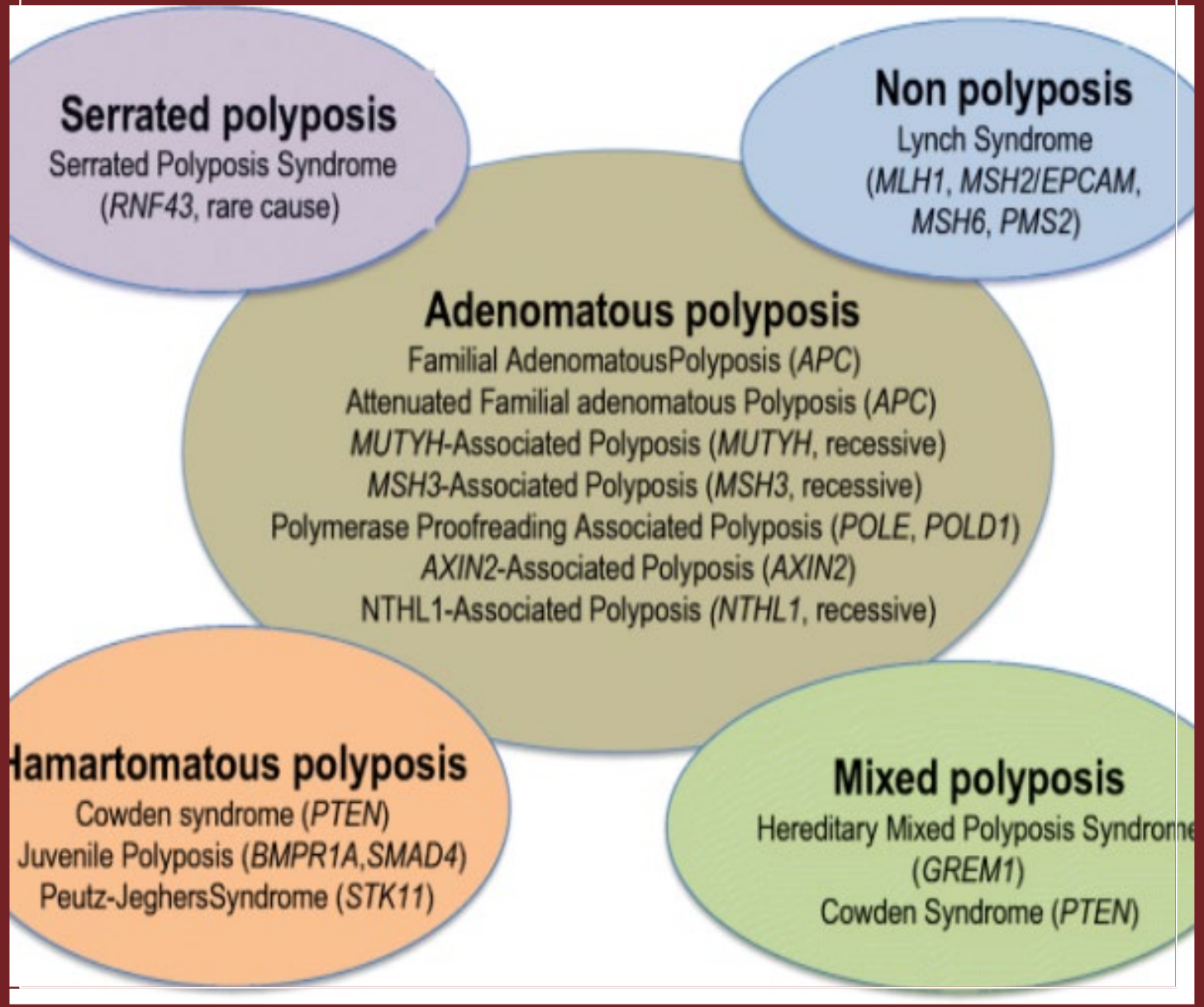
OBJECTIVES

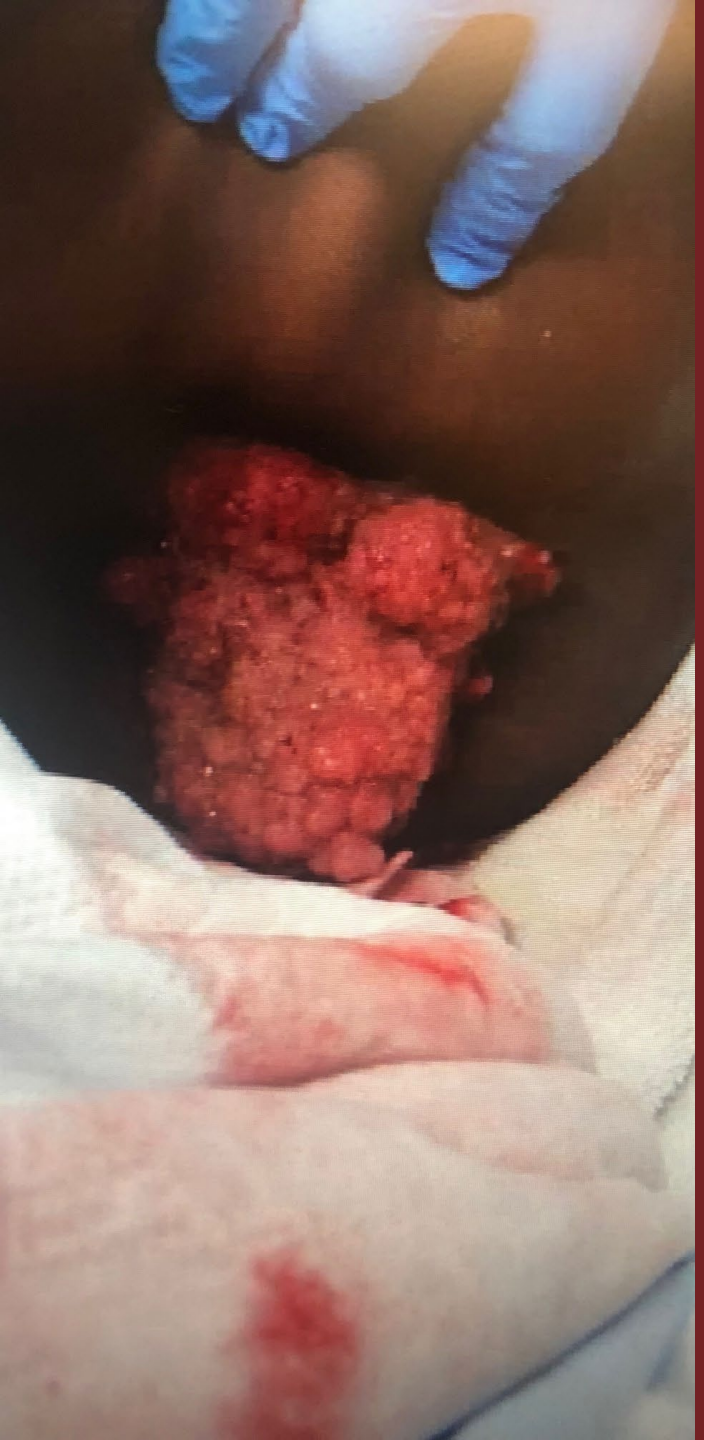
- Recognize presenting symptoms and clinical presentation of FAP patients
- Discuss the epidemiology for the disease for the disease process and association with other neoplastic processes
- Describe the long-term impacts of this disease on the patient and affected family
- Review surgical treatment and surveillance options

DISCLAIMER

I am indeed a redneck from NC. I have no financial interest or professional relationships with any healthcare or instrument organization and do not discuss specific products.

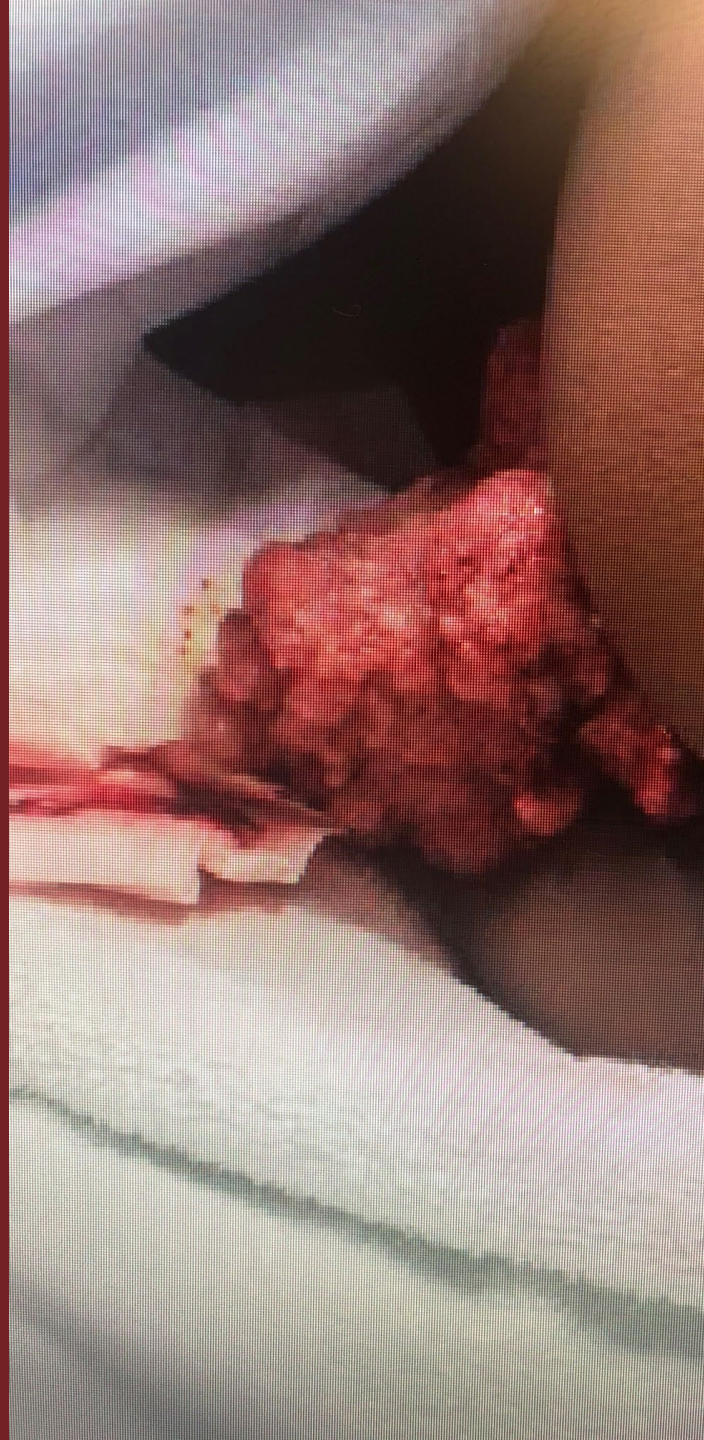
HEREDITARY POLYPOSIS SYNDROMES





CASE STUDY

22 year old male with bright red blood per rectum



FAP – CLINICAL PRESENTATION

- BLEEDING (80%)
- Diarrhea (70%)
- Abdominal pain
- Mucous discharge
- CRC at time of diagnosis (27%)
  - Mt. Sinai, NY
  - 12-43% in the literature
- Weight loss (usually in the setting of malignancy
- Upper GI adenomas (Duodenum)
- Osteoma (Jaw)
- Congenital hypertrophy of retinal pigment epithelium - CHRPE (Eyes)
- Epidermoid cysts
- Unerupted or additional teeth
- Desmoid tumors
- Adrenal gland tumors

FAP QUICK HITS

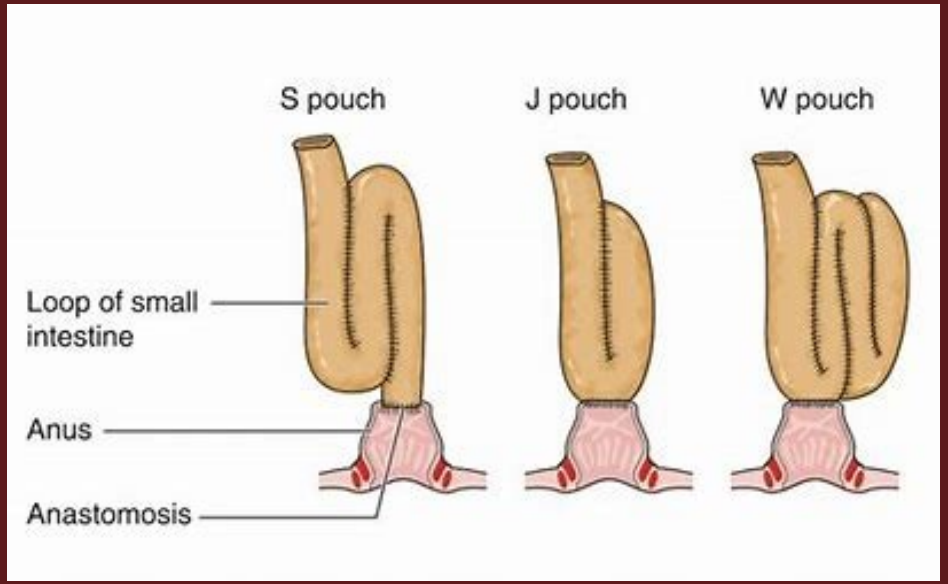
- Incidence 0.0001% (1:10,000)
- Symptoms in early adolescence
  - *Polyposis ~ 16 yo with >100 polyps (attenuated 30s with < 50 polyps)*
- Cancer risk ~ 40 yo (attenuated 50-60)
- APC gene (yMUTYH)
  - *ß-catenin downregulation*
- Familial (20-25% spontaneous)
- Desmoid tumors, duodenal cancer, biliary tract carcinoma, Hepatoblastoma, Medulloblastoma

SURGICAL OPTIONS

WHAT TO DO WITH OUR 22-YEAR-OLD??

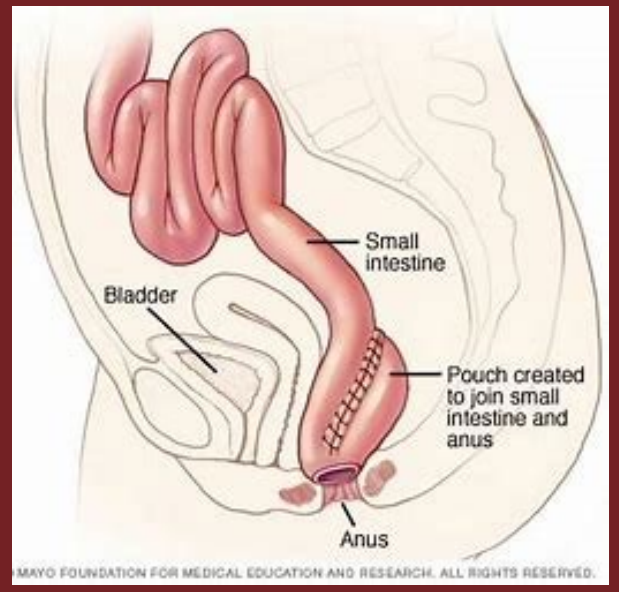
SURGICAL OPTIONS

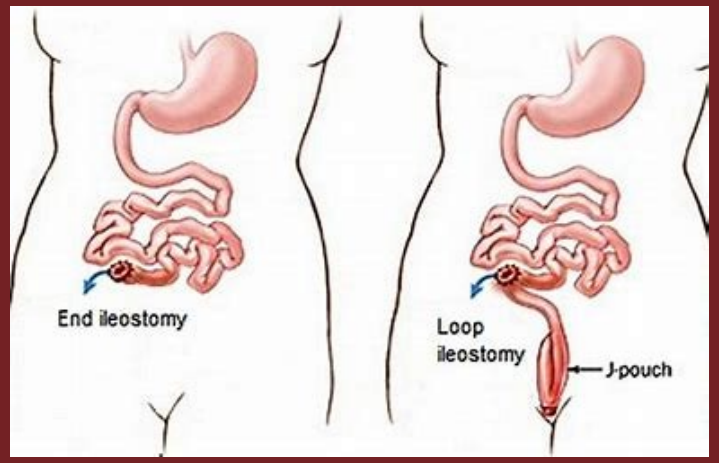
- Total abdominal colectomy with ileorectal anastomosis
  - *Requires surveillance*
  - *Future risk of cancer*
  - *Bowel function closer to baseline*
- Total proctocolectomy with IPAA
  - *+/- mucosectomy*
    - Surveillance of anal mucosa
  - *Staged procedure*
  - *More frequent bowel movements*
- Total proctocolectomy with end ileostomy
  - *Easier to control succus*

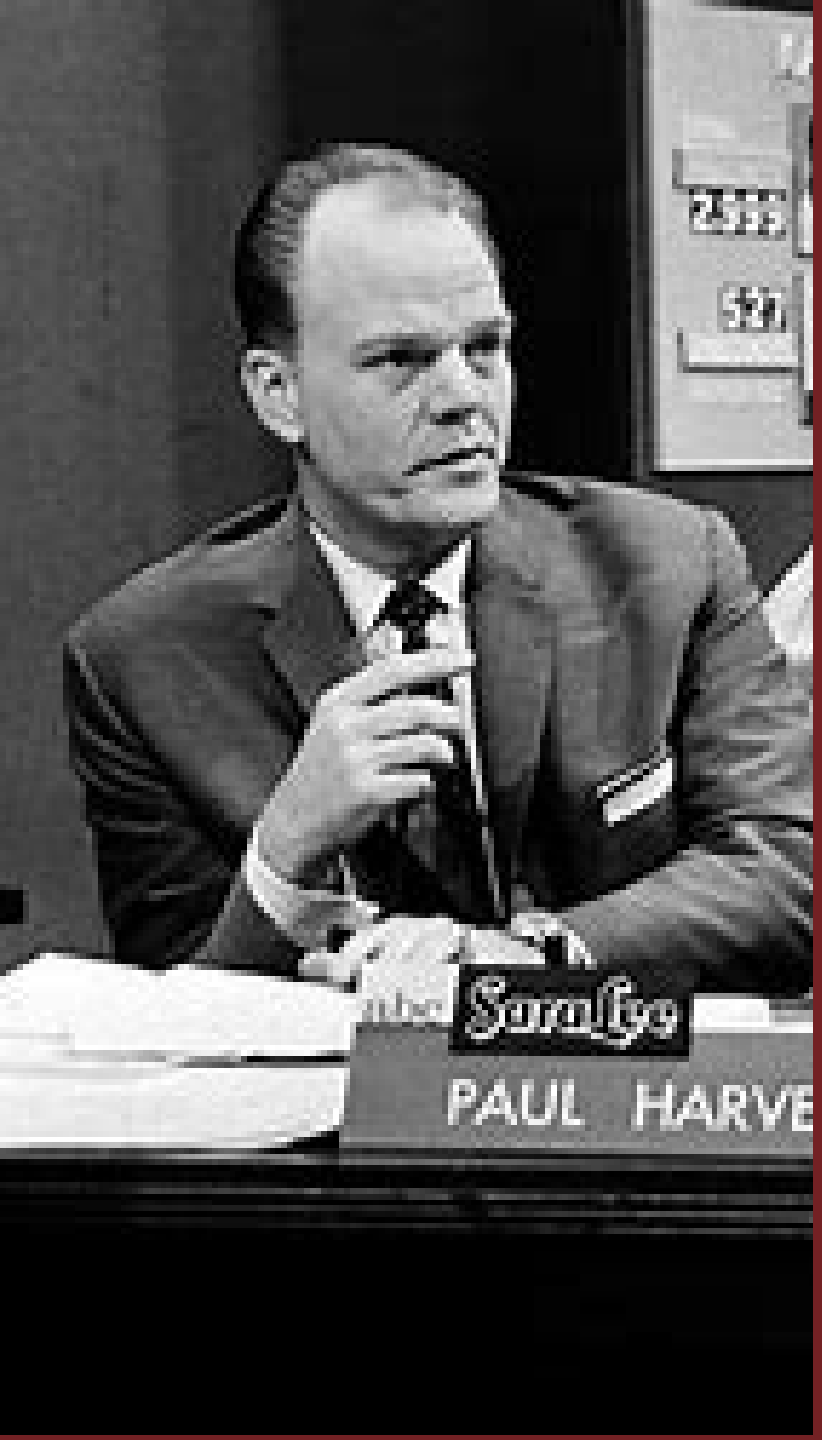


IMPACTING FACTORS

- Diagnosis and workup in adolescence
  - Surgery can be delayed until patient reaches appropriate intellectual and physical maturity unless disease or symptoms are more severe
- Surveillance for extracolonic manifestations
- Alterations of bowel habits in youth
  - Can be socially isolating
  - Many centers include therapy sessions and support groups.
- Physical, financial, and psychological stressors on parents/siblings
  - Threat of malignancy
  - Staged surgery
  - Multiple multidisciplinary appointments







AND NOW…THE REST OF THE STORY

- Prolapsing mass was reduced
- Total abdominal colectomy distal to mass (proximal rectum) with end ileostomy
- Pathology multicentric areas of high-grade dysplasia, no adenocarcinoma
- Staged completion proctectomy with IPAA – J pouch
- Improved bowel function with fiber and anti-diarrheal medications
- Duodenal adenomas resected with surveillance every 1-2 years

QUESTIONS??