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St Mark's Hospital, UK

GI Polyp syndromes in children

Screening and surveillance, surgery.

No conflict of interests to declare

Objectives

- Understand the reason for screening and surveillance
- Understand the genetic basis for different polyposis syndromes
- Update on how and when adolescents and children should undergo genetic testing and commence colonoscopic surveillance and surgery

Classification of Intestinal Polyps

- Common polyps
  - Juvenile polyps (hamartoma)
- Inheritable polyposis
  - adenomatous polyps
    - familial adenomatous polyposis coli (FAP)
    - MYH-associated polyposis (MAP)
  - hamartomatous polyps
    - Peutz-Jeghers syndrome (PJS)
    - PTEN hamartoma tumor syndromes
      - Cowden's disease
      - Bannayan-Riley-Ruvalcaba syndrome
    - Juvenile polyposis (JPS)

Journal of Pediatric Gastroenterology and Nutrition
48:S75–S78

Screening and Surveillance Recommendations for Pediatric Gastrointestinal Polyposis Syndromes

John Barnard

Research Institute at Nationwide Children's Hospital, Department of Pediatrics, Ohio State University College of Medicine, Columbus, OH, USA

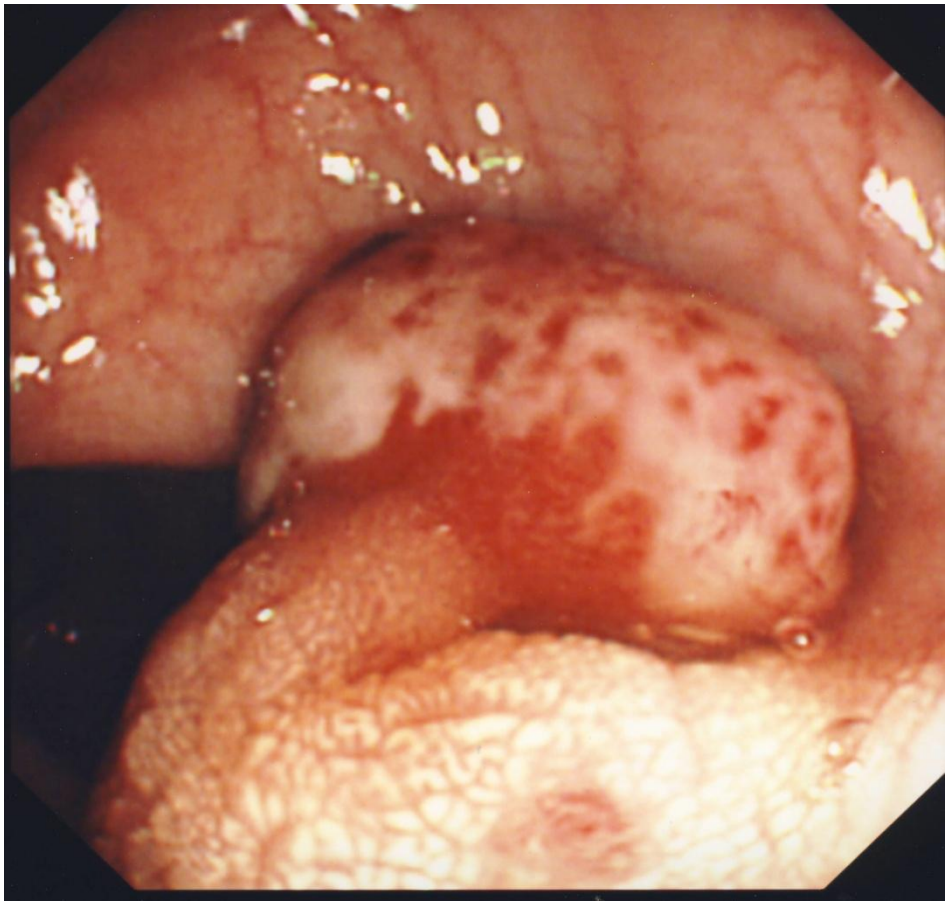
Peutz—Jeghers syndrome: a systematic review and recommendations for management

A D Beggs,[1] A R Latchford,[2] H F A Vasen,[3] G Moslein,[4] E Alonso,[5] S Aretz,[6] L Bertario,[7] I Blanco,[8] S Bülow,[9] J Burn,[10] B Capella,[11] C Colas,[12] W Friedl,[6] P Møller,[13] F Lesueur,[14] L Sävolainen,[15] J-P Mecklin,[16] E M Ngeow Joo,[17] J Parc,[18] R K S Phillips,[2] W Hyer,[19] M Ponz de Leon,[20] L Renkonen-Sinisalo,[15] J R Sampson,[21] A Stormorken,[22] S Tejpar,[23] A J W Thomas,[24] J T Wijnen,[14] S K Clark,[19] S Hodgson[1]

So why consider screening and surveillance?

- Solitary colonic polyp – to avoid missing others
- FAP – to prevent a CRC
- PJS – to prevent a mid gut intussusception and laparotomy
- JPS – to characterise the condition

The solitary isolated colonic polyp



Single “harmless juvenile” colonic polyp

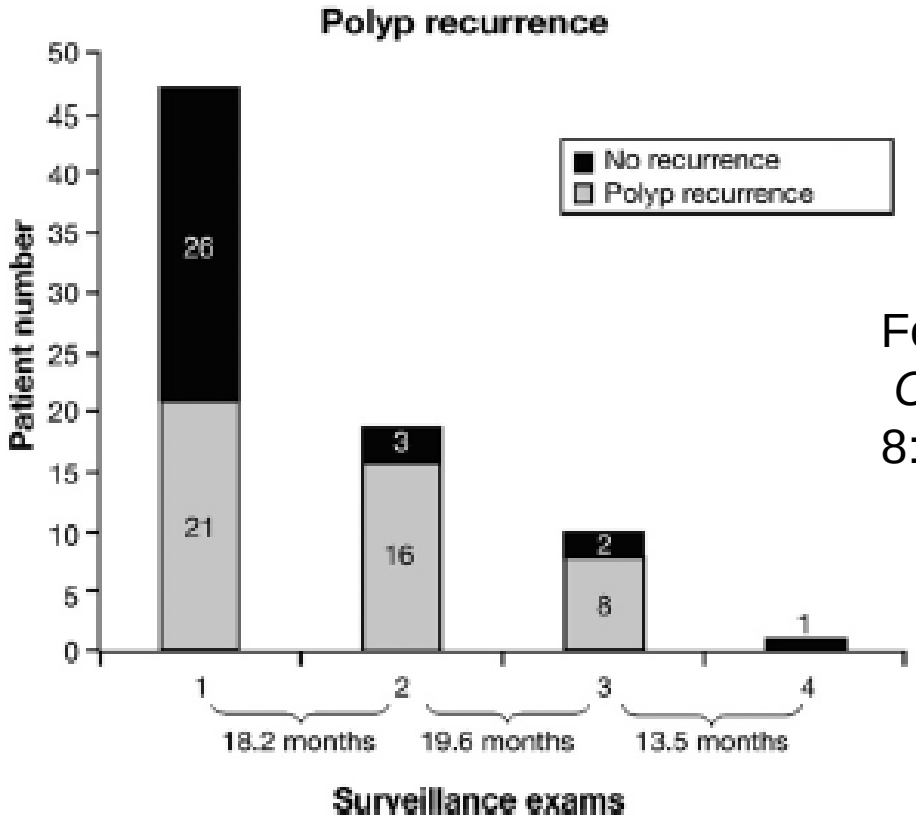
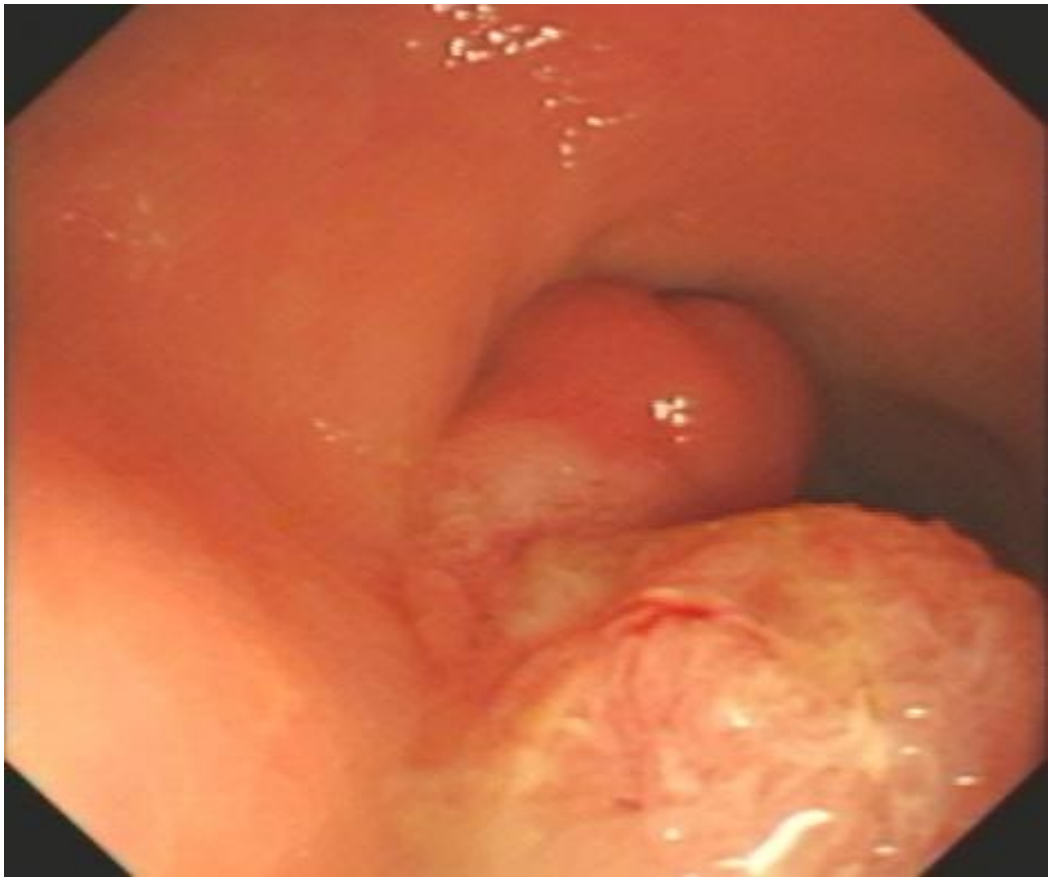


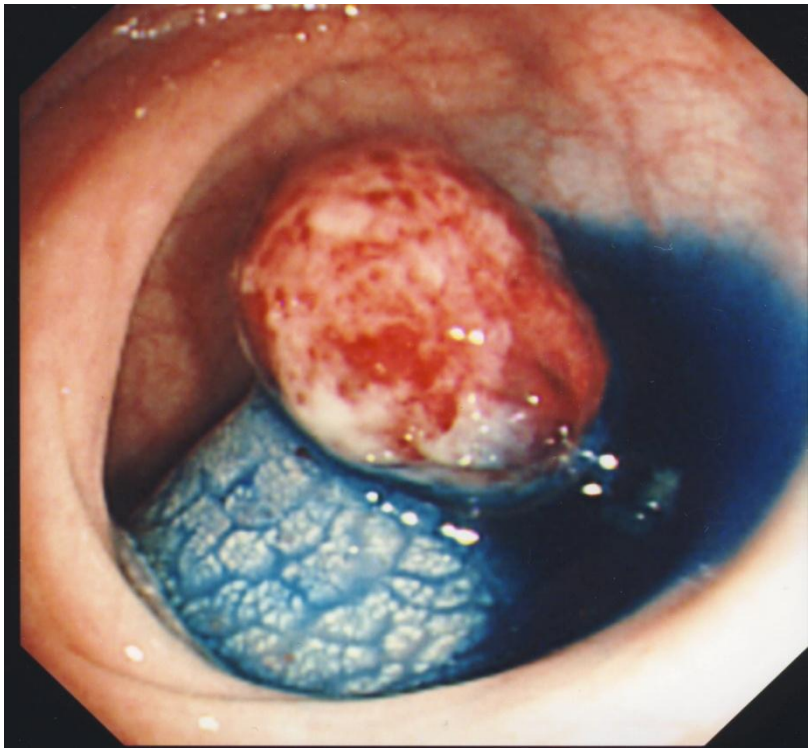
Figure 2. Polyp recurrence in patients returning for repeat surveillance colonoscopy after complete polyp removal or eradication during the preceding examination. Only patients with complete colonoscopy examinations are included. Median time intervals between successive examinations are shown.

Fox *at al*
Clin Gastroenterol Hepatol
8: 796, 2010

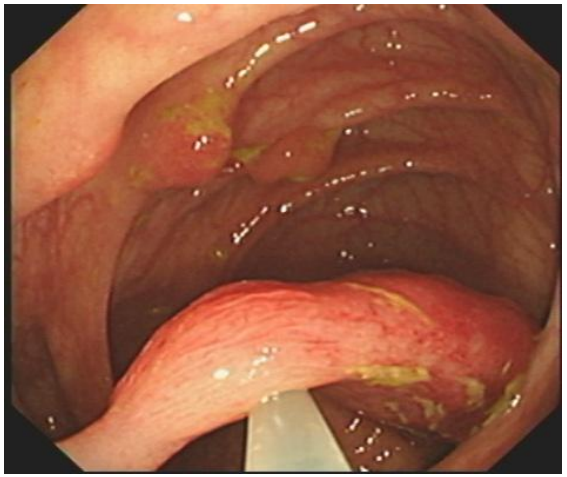
Not all single polyps are benign

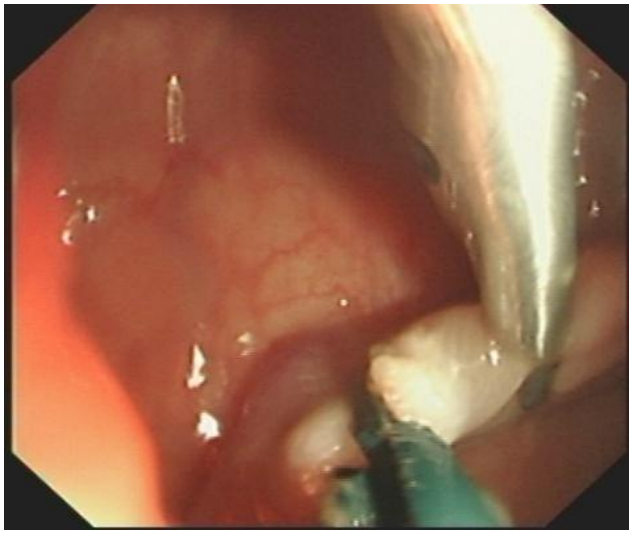


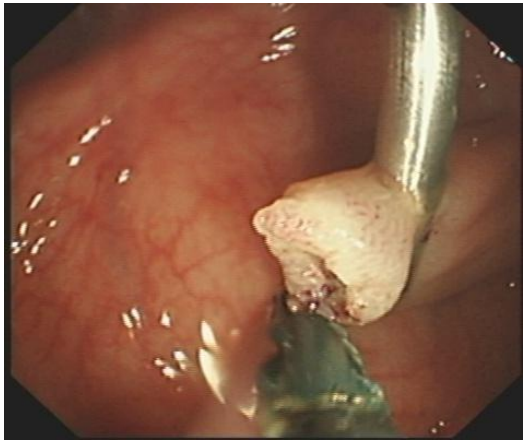
Preinjection

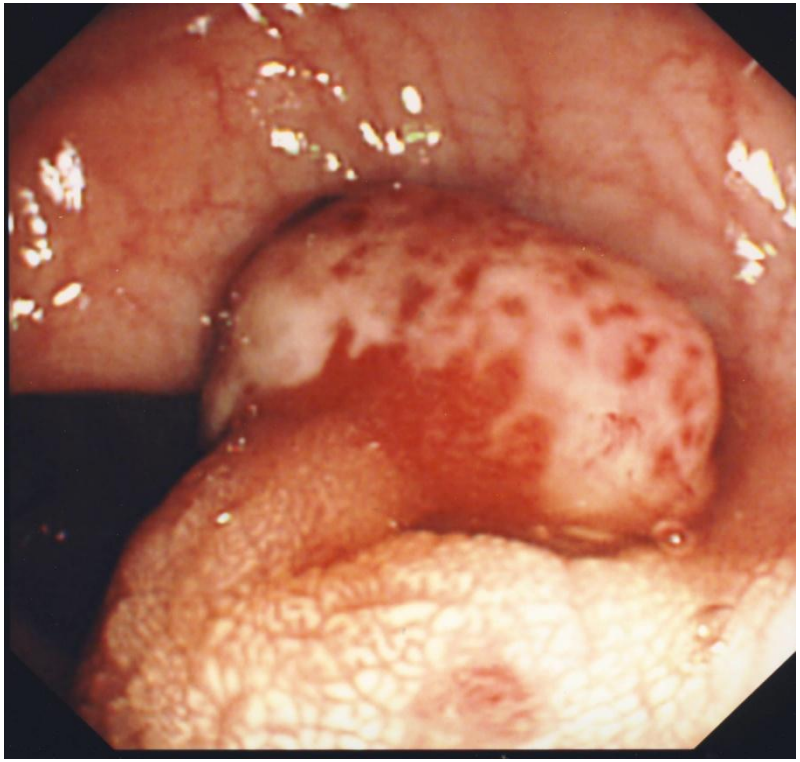


The surgeon is defunct









Bottom Line

They are benign – nearly always

Polypectomy needs waterproof pants

Rescoping to know if there are more

Consider FH

Rescope if bleed

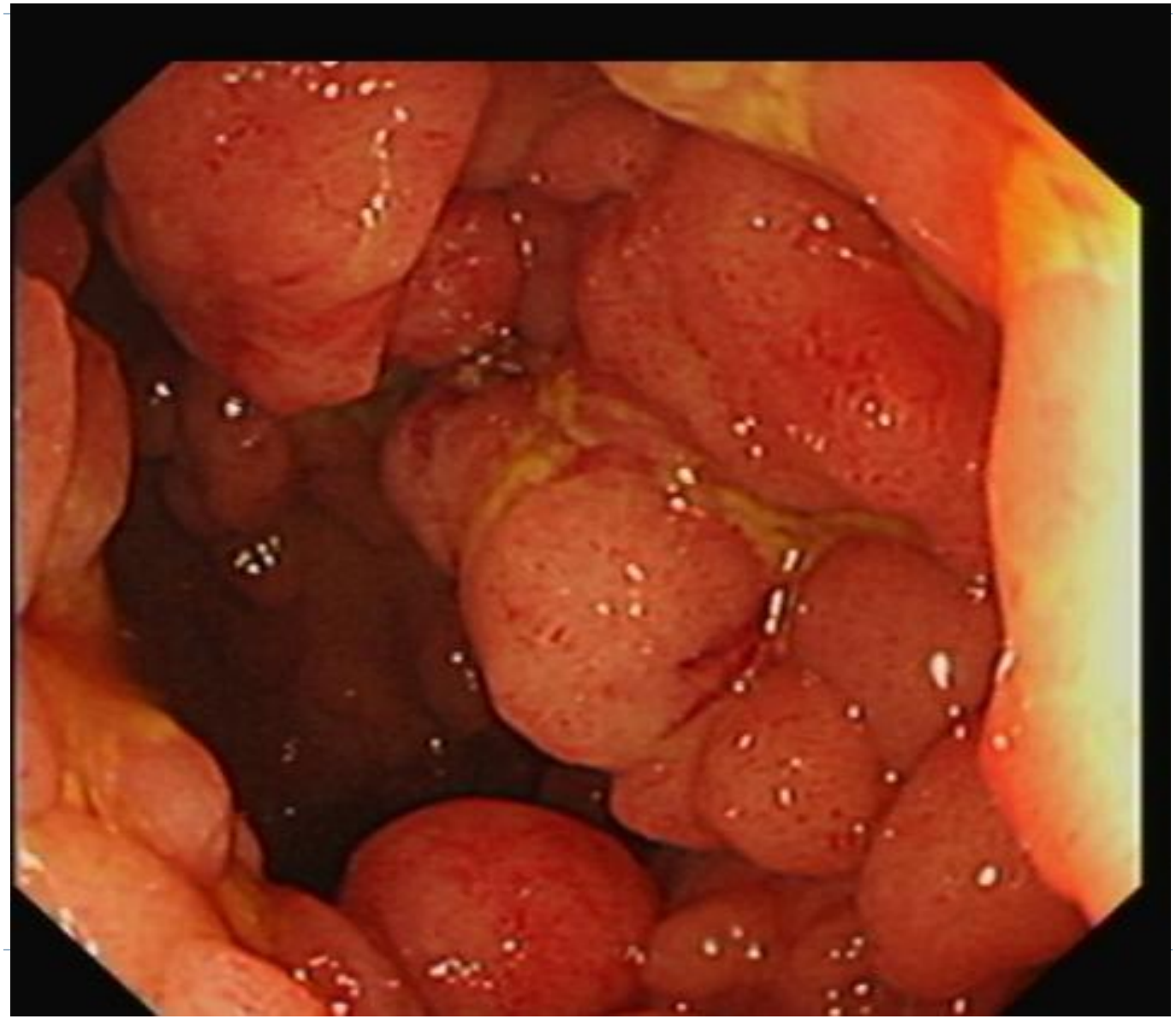
Perhaps rescope later if bleeding, FH, uncertain histology

FAP - Clinical scenario

- A 7 year old from a family known to be affected by FAP comes to your clinic with infrequent rectal bleeding.

  - Why and when to screen and start surveillance?
  - Should you perform genetic testing?
  - Should you undertake a colonoscopy?

Current practice, start screening in teenage years



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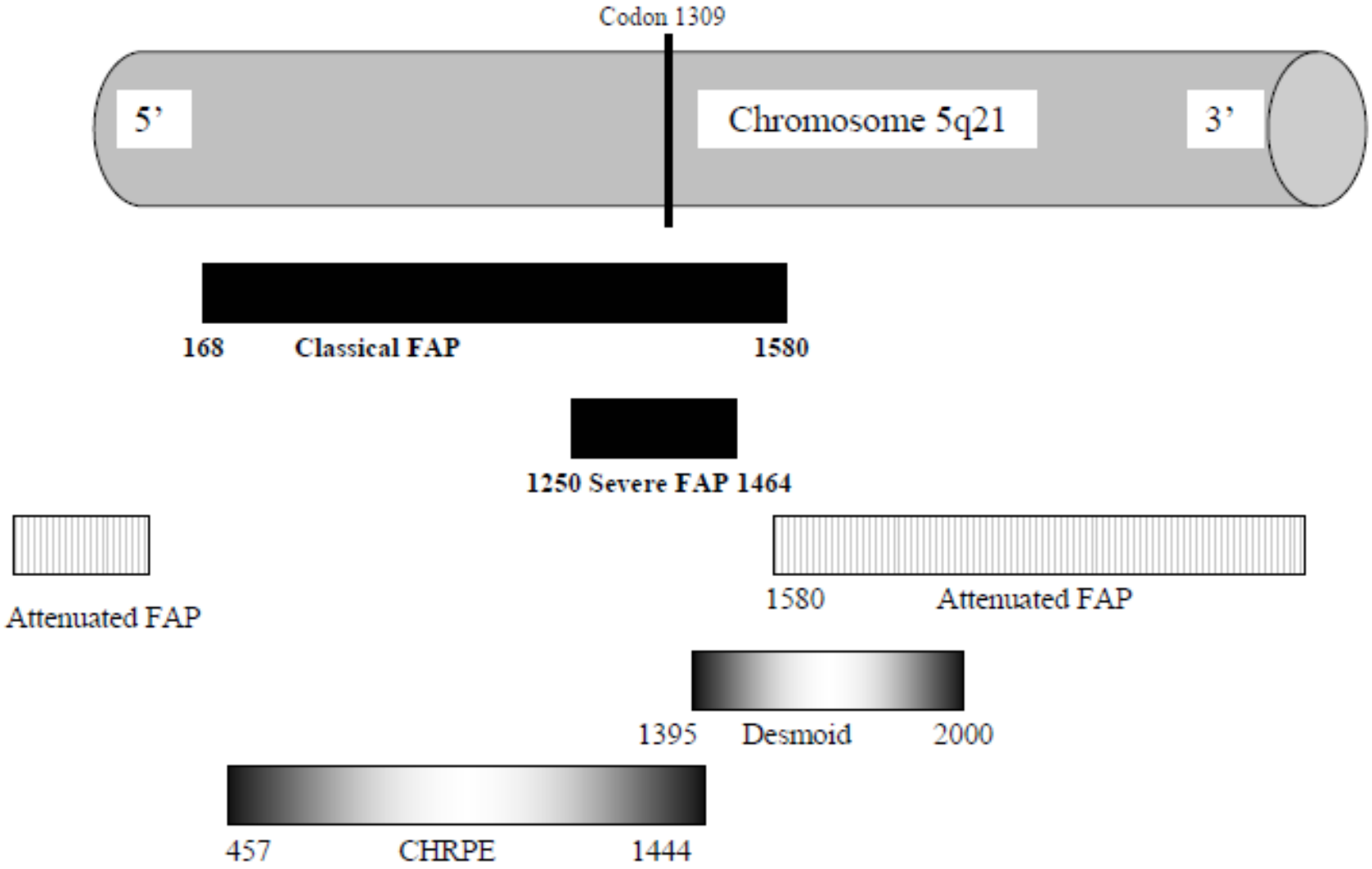
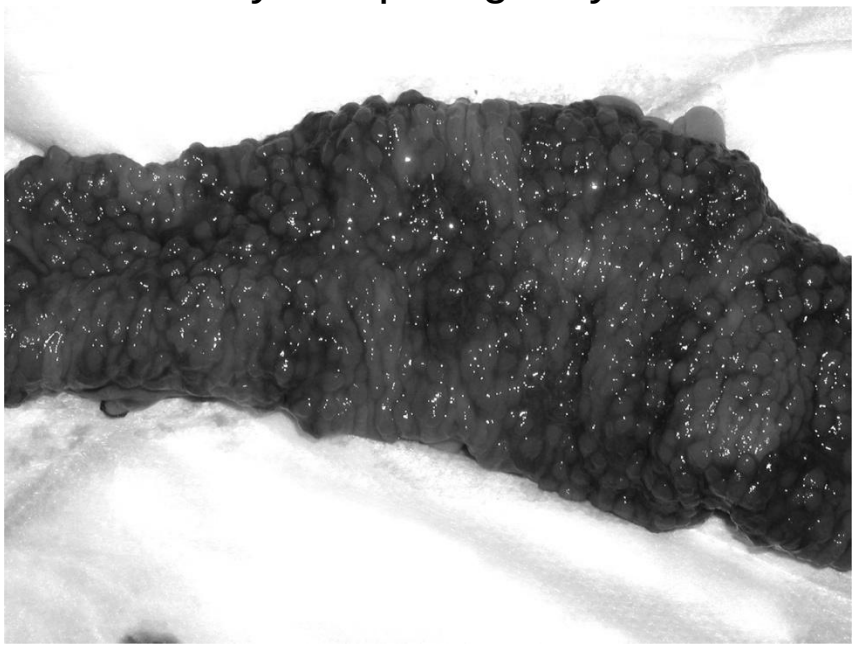

Codon 1309
5'
Chromosome 5q21
3'
168
Classical FAP
1580
1250 Severe FAP 1464
Attenuated FAP
1580
Attenuated FAP
1395
Desmoid
2000
457
CHRPE
1444

Early childhood presentation of FAP

- No FH
- Presents with rectal bleeding alone
- CHRPE
- Mutation codon 1309

- Youngest symptomatic FAP child

Colectomy sample age 4 years



Symptomatic Polyposis in a Four-Year-Old: The Exception Proves the Rule
Will, Phillips, Hyer, Clark.

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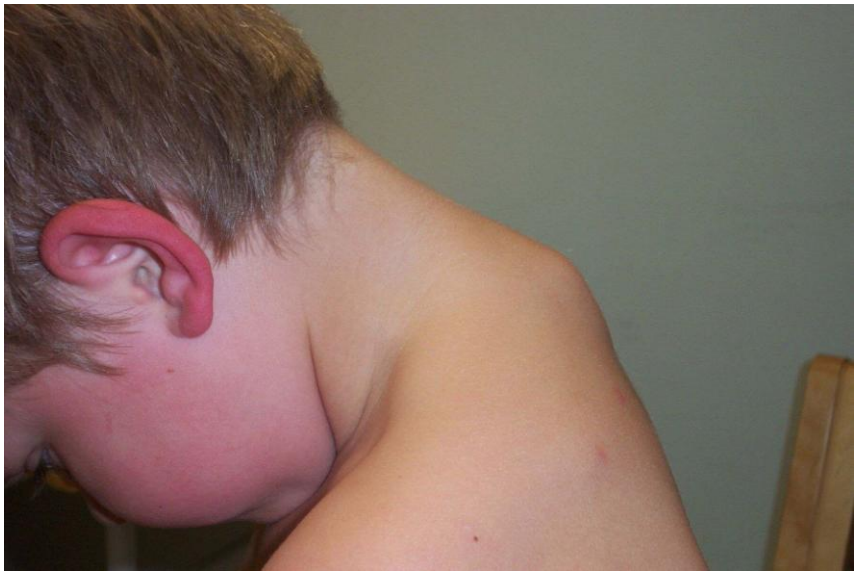
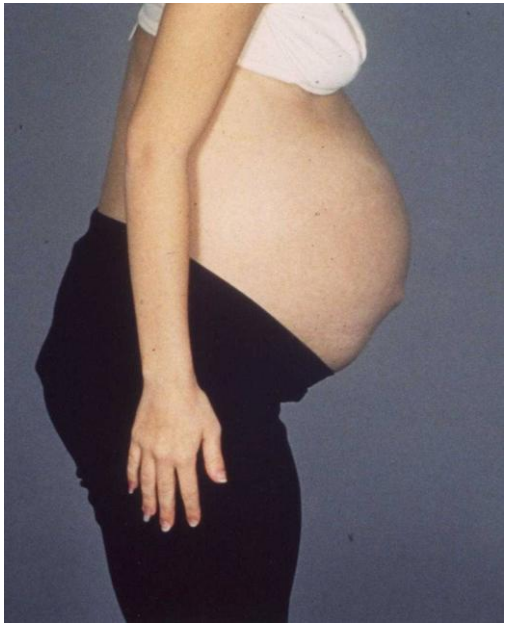
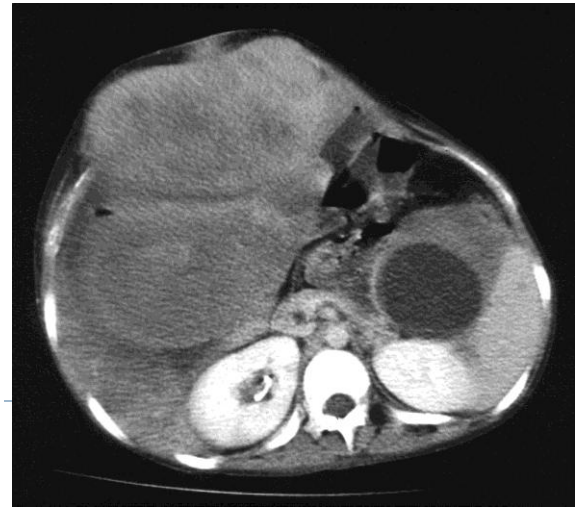
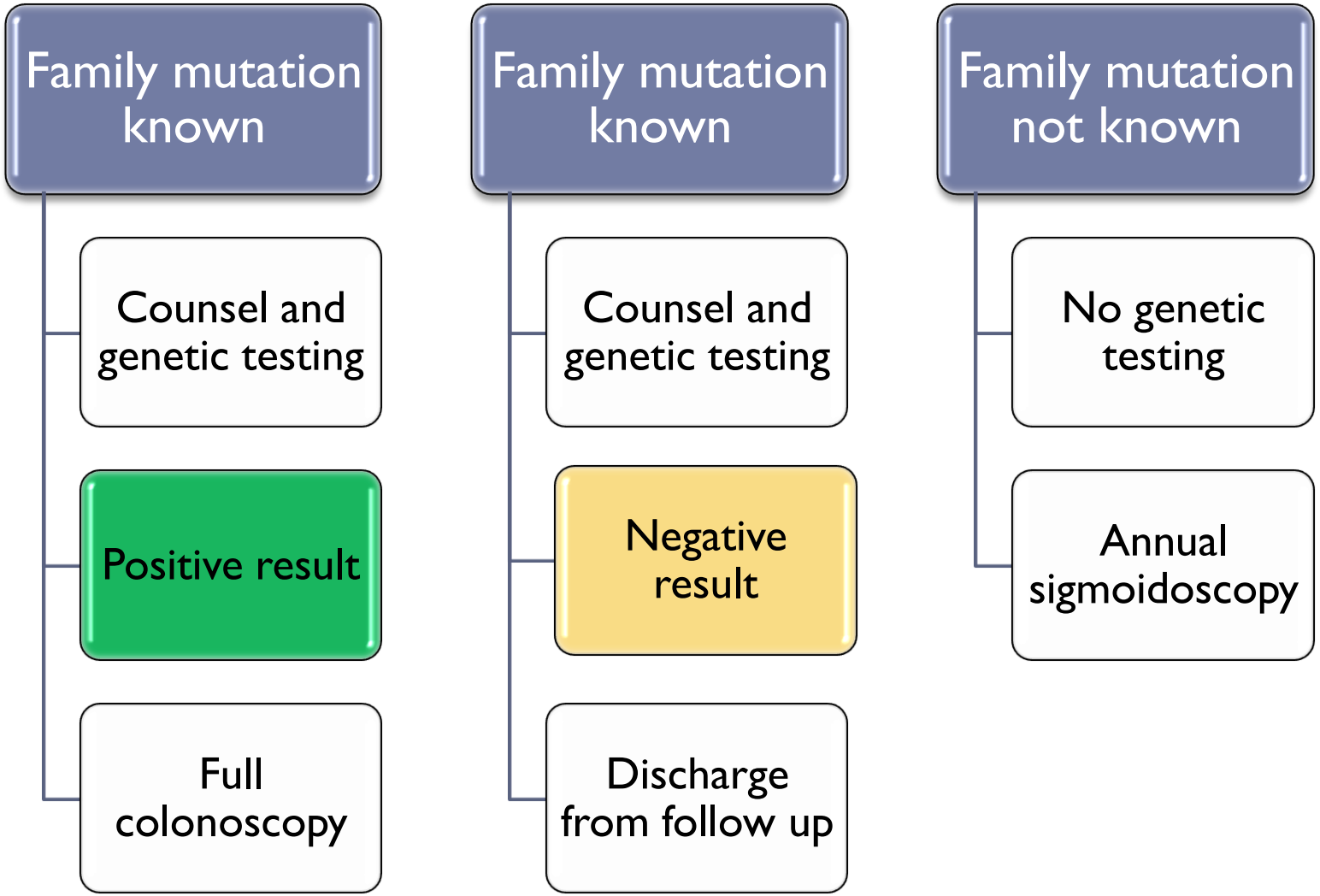
Desmoid disease- codon >1400







Commence genetic testing at age of earliest onset of disease



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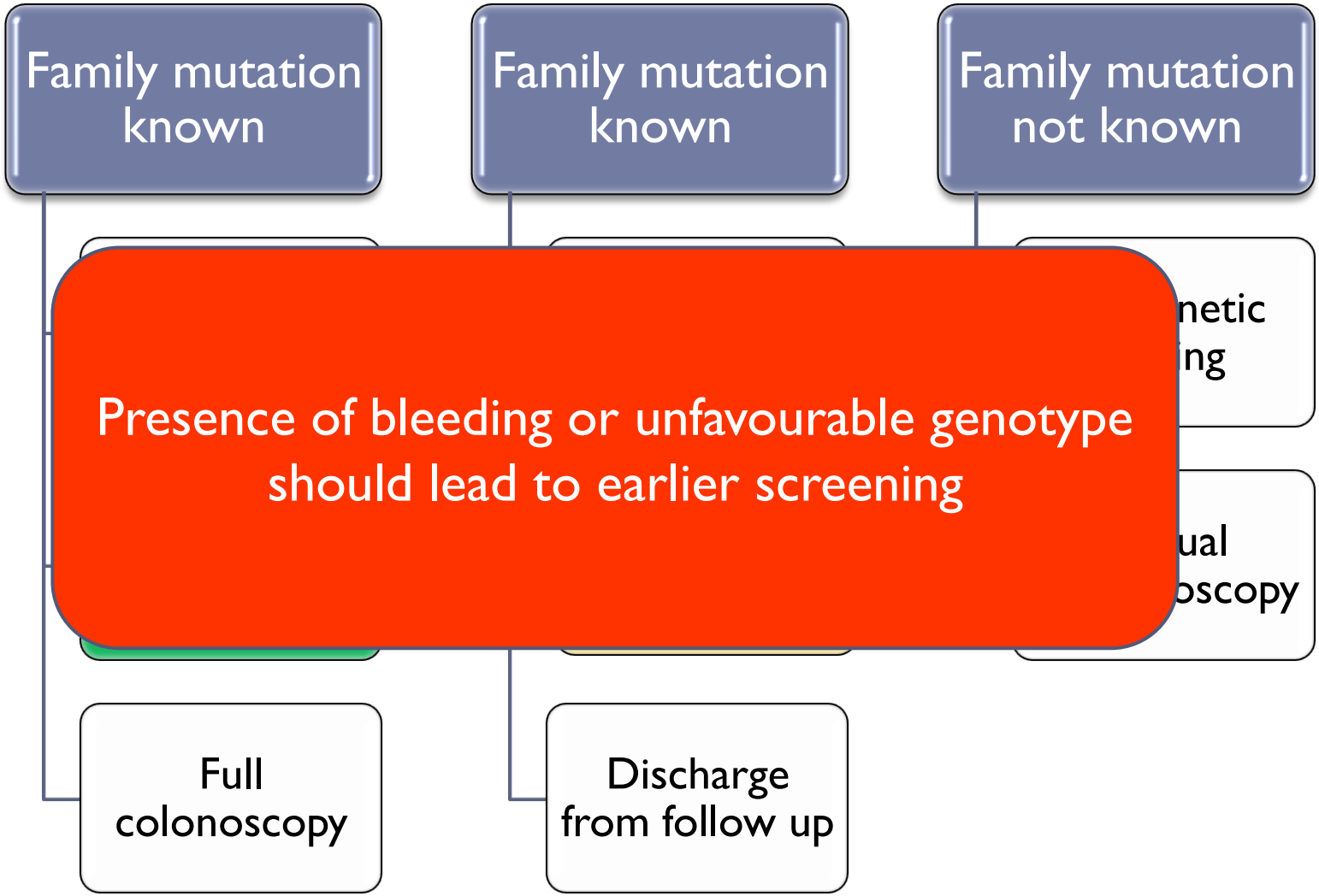

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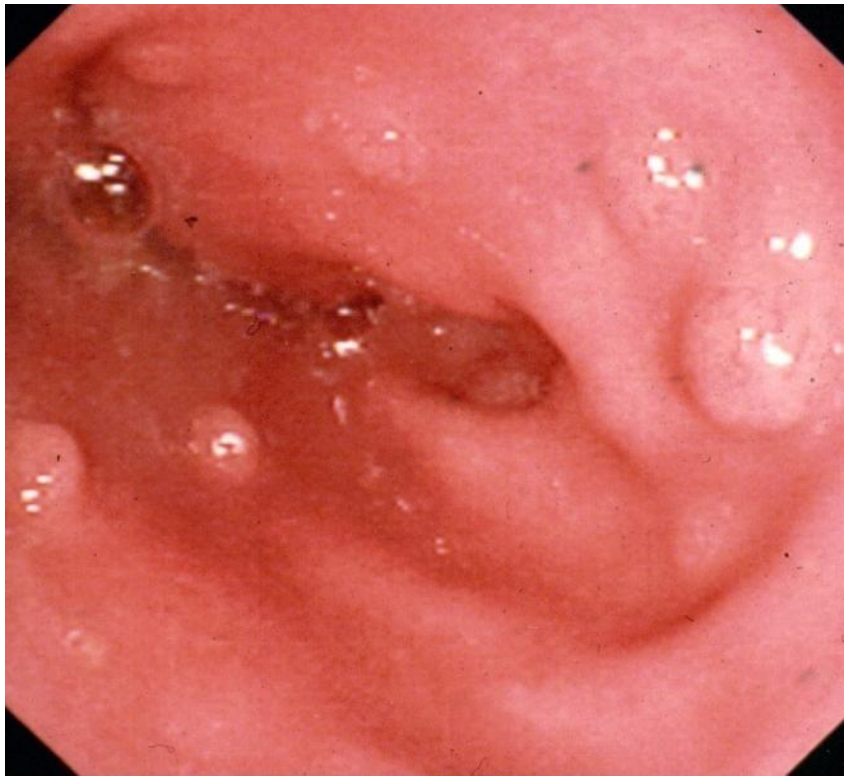
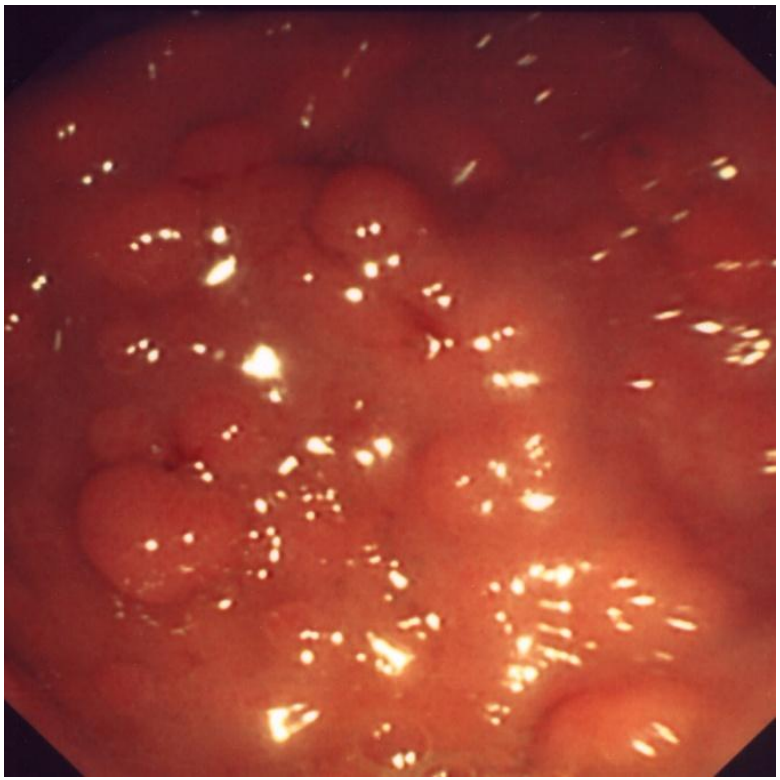
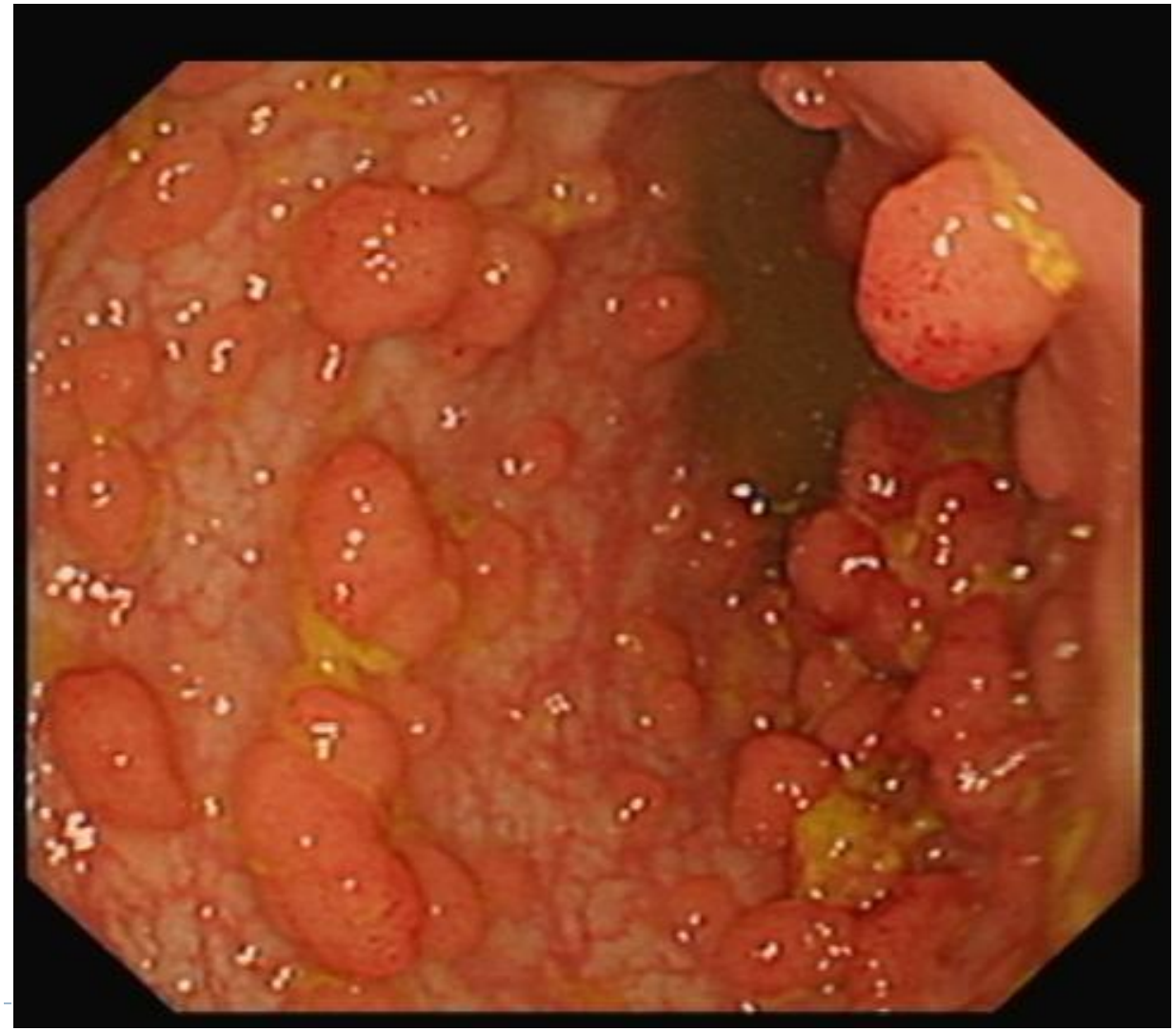
Assess adenoma burden in the rectum

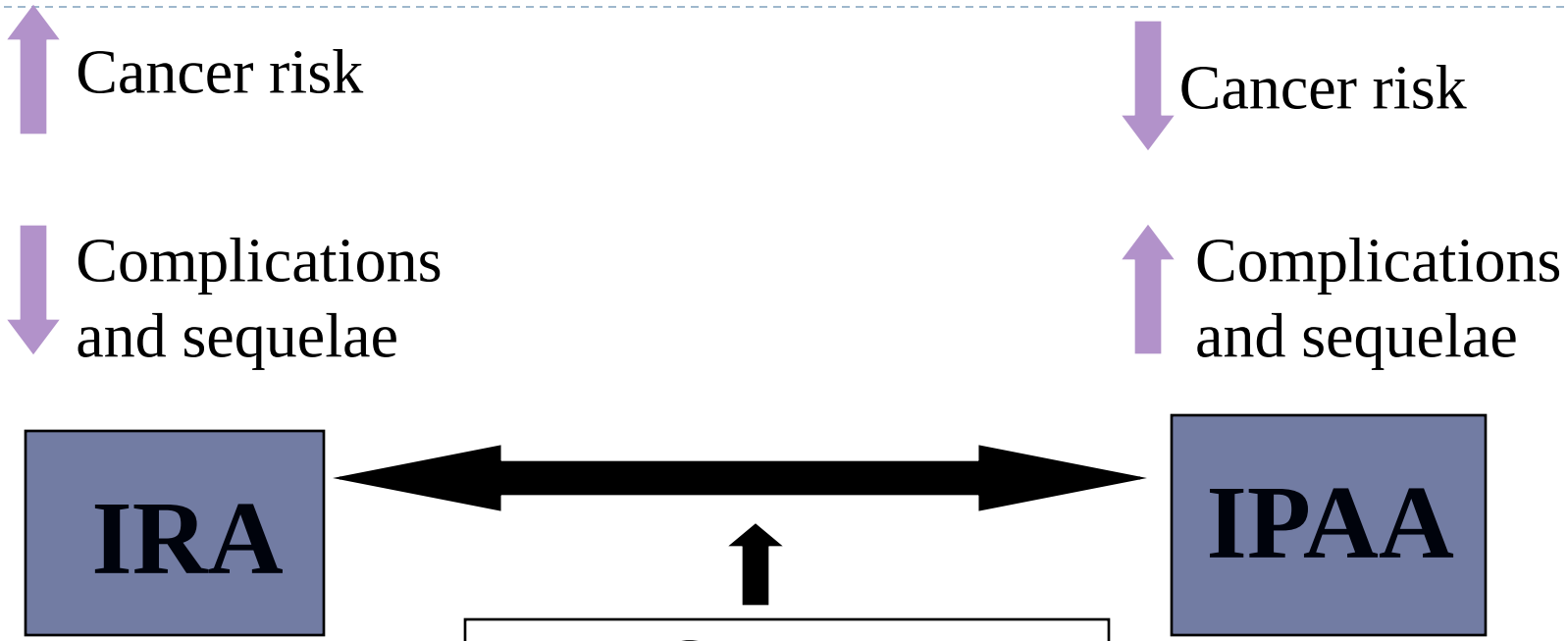




Assess polyp burden in the colon

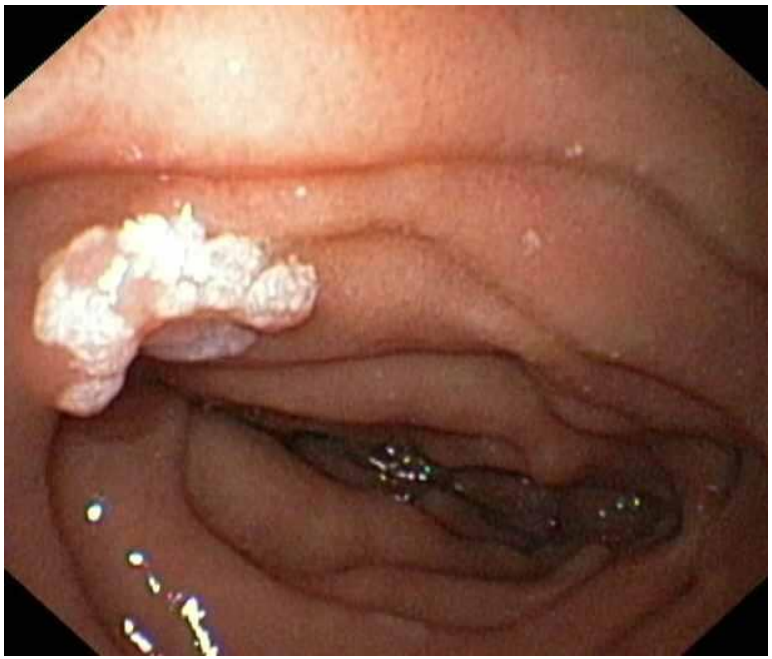


Annual colonoscopy impacts on surgical choices



Delay duodenal surveillance until > 20+

- Duodenal polyposis
  - Spigelman classification



Surveillance for a teenager does not stop after colectomy

FAP proband.
Genetic testing colonoscopy from age 10-12 years depending on symptoms.
Refer for surgery depending on adenoma burden

IRA if rectal adenoma burden permits

6 monthly sigmoidoscopy

IPAA if preferable

Annual pouchoscopy

Long term FU and commence duodenal surveillance (age 20+)

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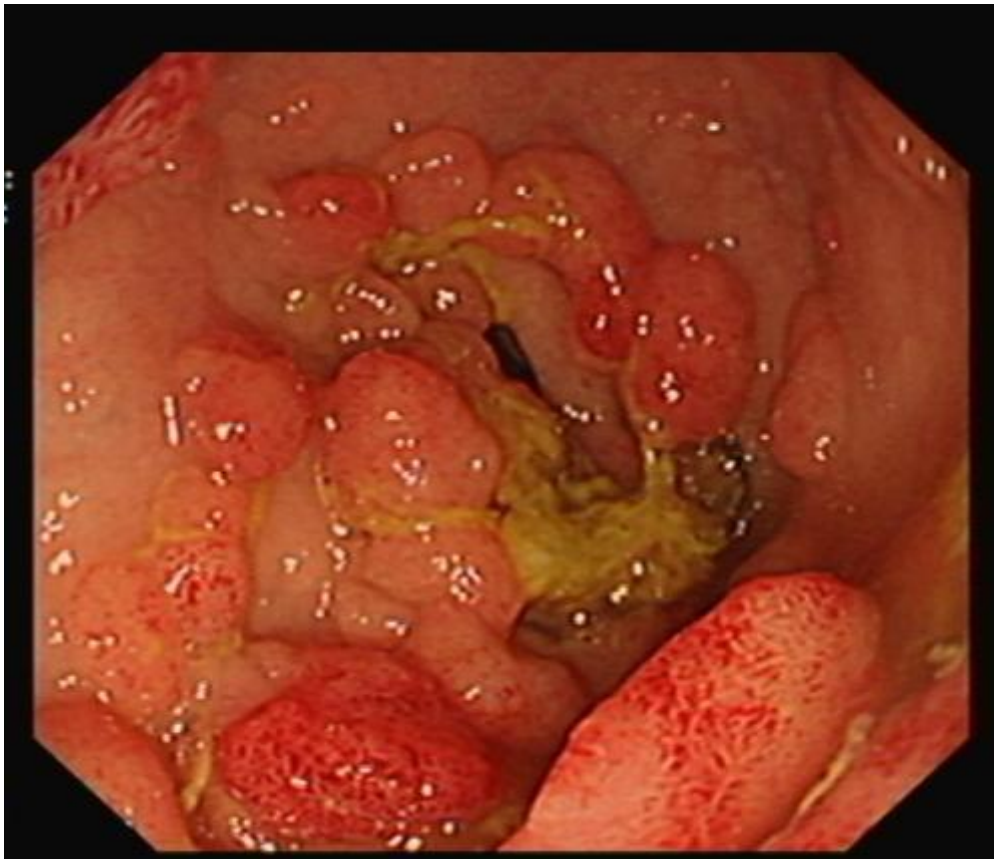


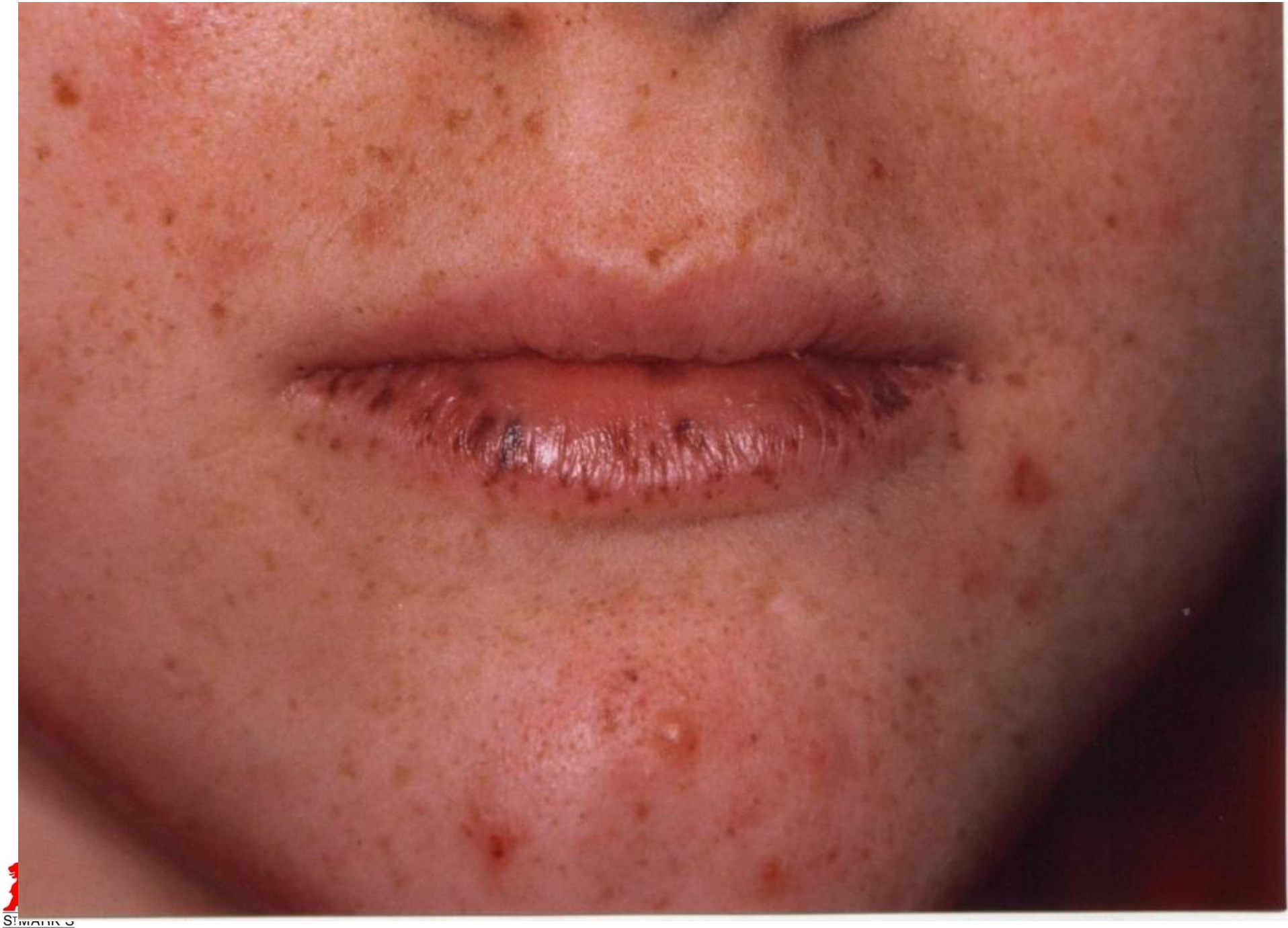
Bottom Line

Often wait until teenage years

Commence earlier screening if bleeding or symptomatic

Understand the gene, start earlier if high risk genotype

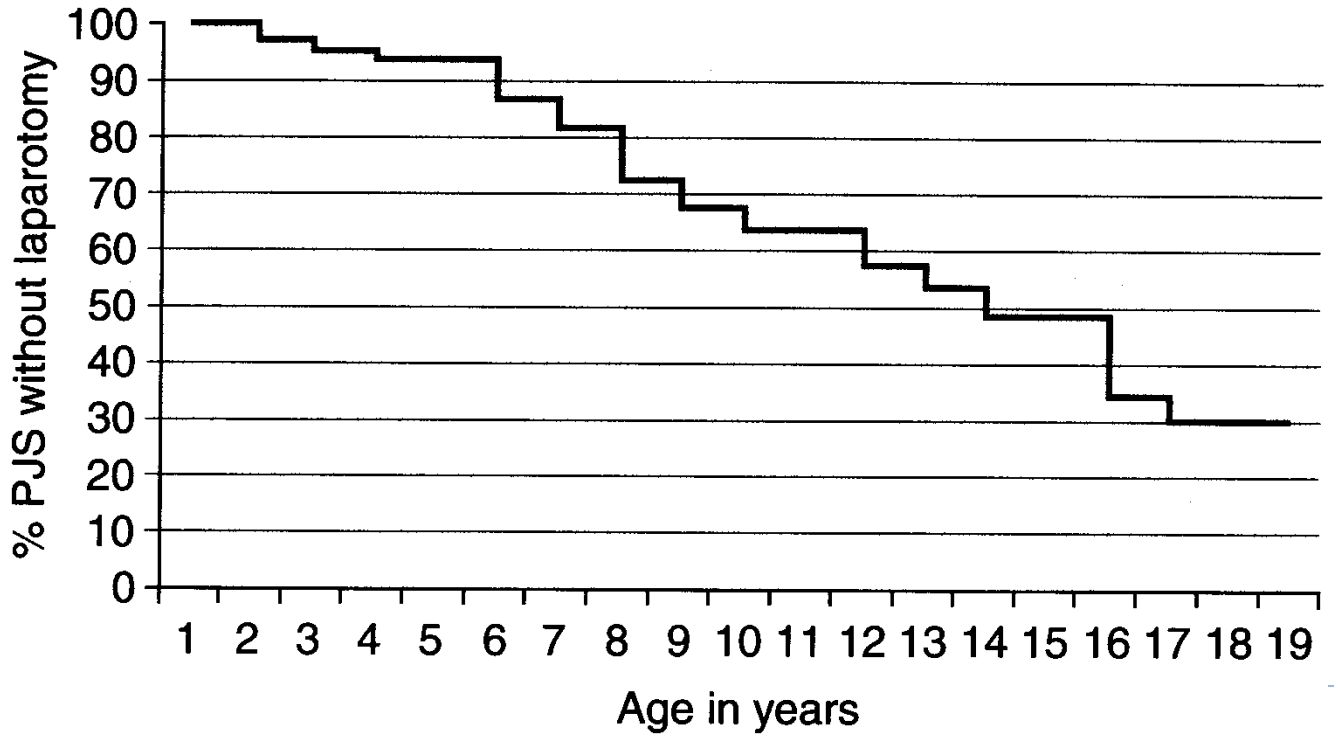
Chemoprevention does not alter timing nor surveillance



Complications of Childhood Peutz-Jeghers Syndrome: Implications for Pediatric Screening

*R. Hinds, †C. Philp, †W. Hyer, and *J. M. Fell

**Department of Paediatric Gastroenterology, Chelsea and Westminster Hospital, London; and the †The Polyposis Registry, St. Mark's Hospital, London, England*



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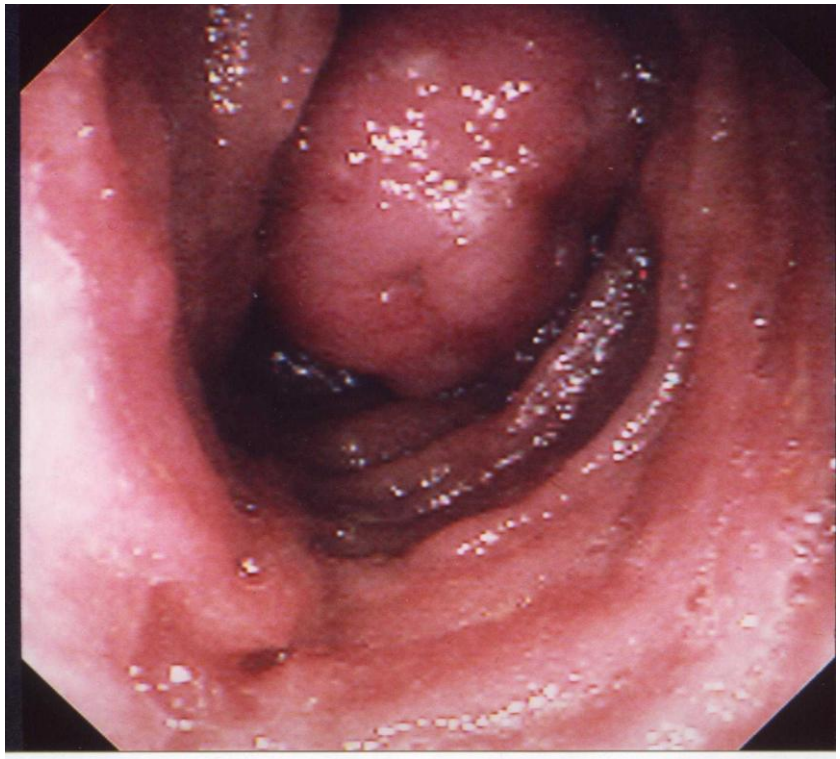
So at what age to undertake genetic testing?



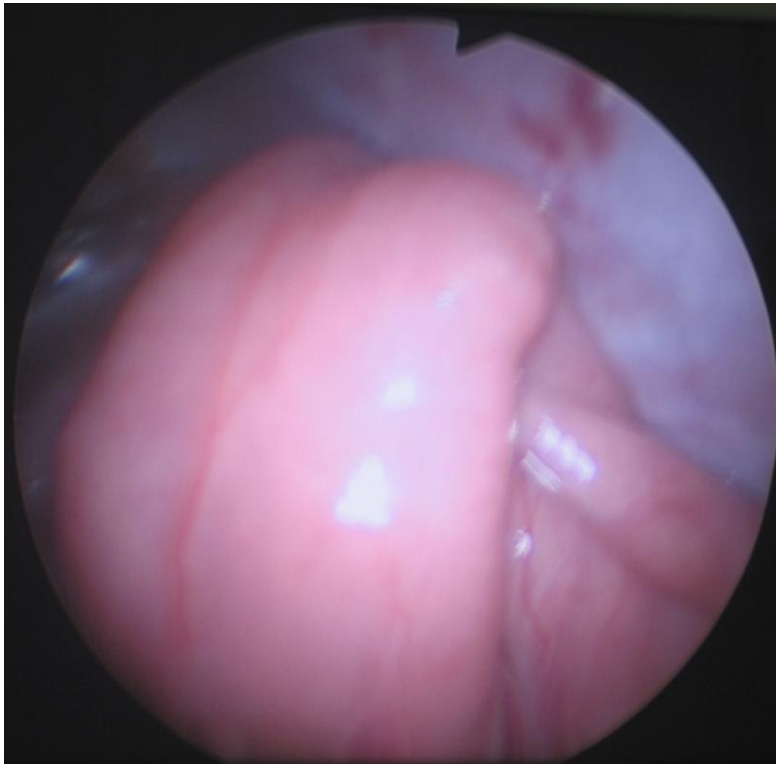
- If the kindred genetic mutation is known (STK11), offer genetic testing prior to age of earliest onset of complications eg infancy

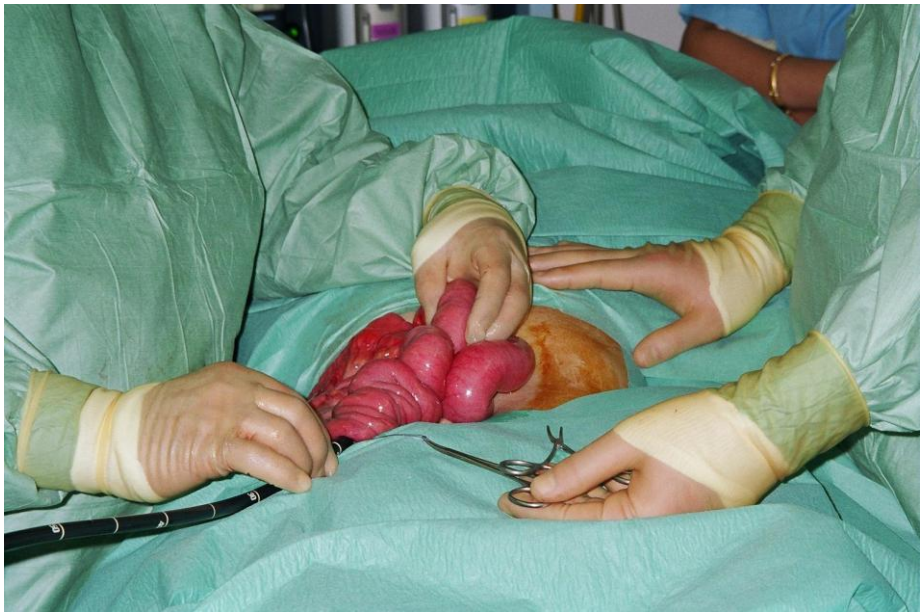
Diagnosis confirmed
eg mucosal pigmentation, FH, genetics

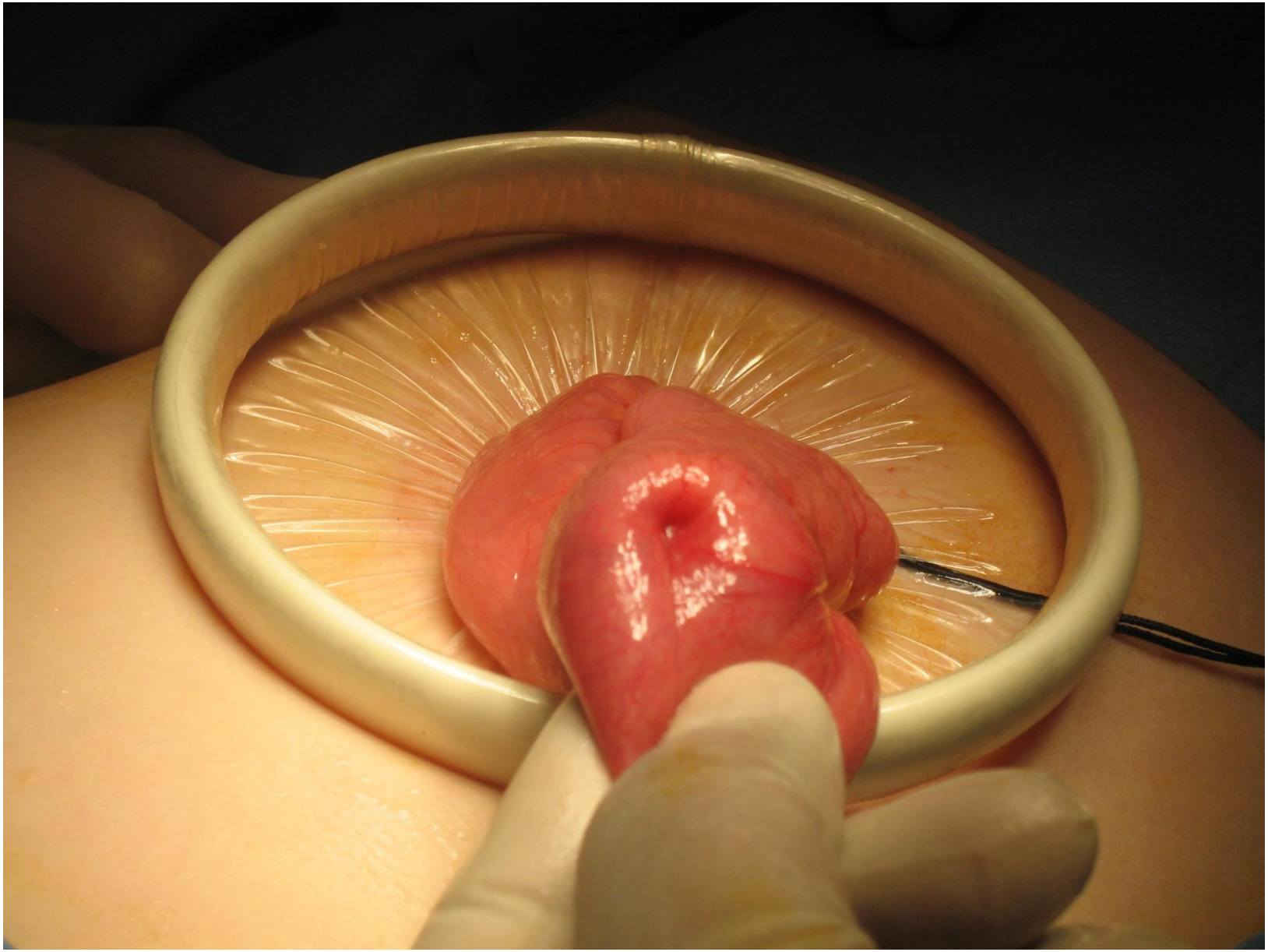
Symptomatic with polyp
leading intussusception

Asymptomatic affected child
Start screening age 5-8 years
OGD, VCE and colonoscopy

Symptomatic and obstructive symptoms with intussusception – get a surgeon







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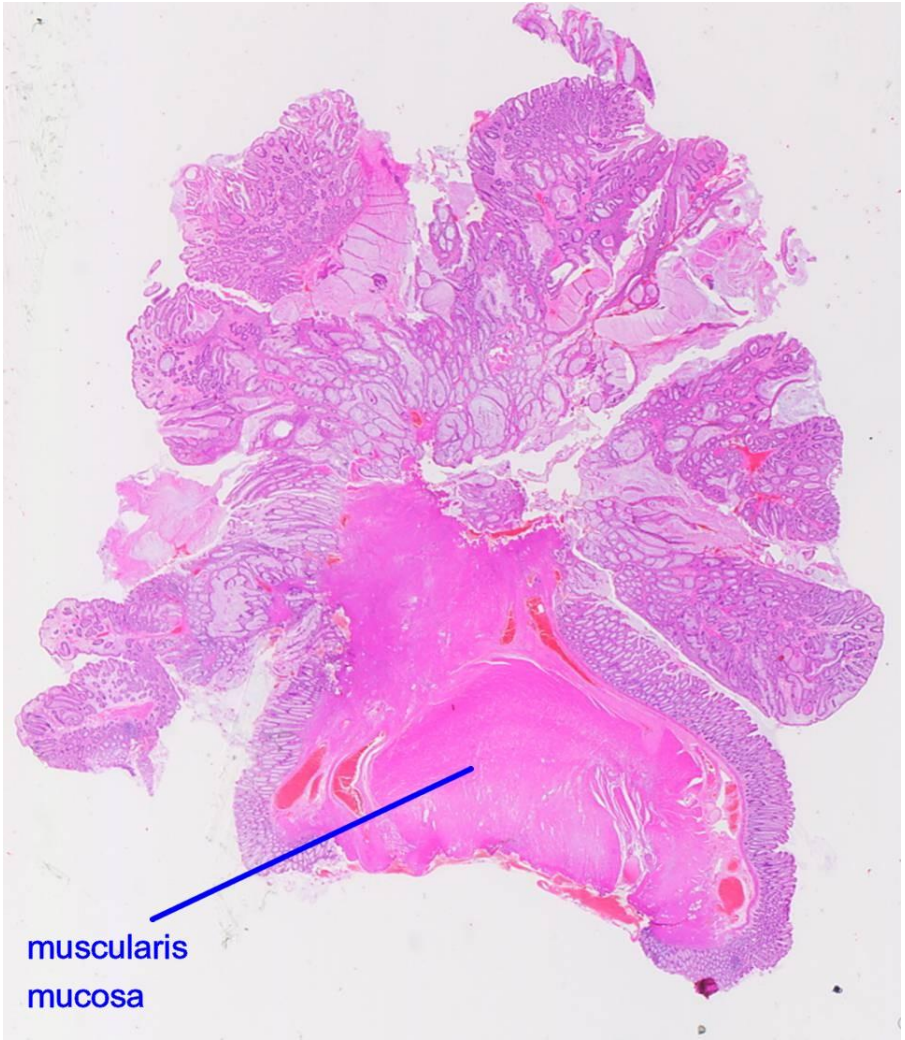
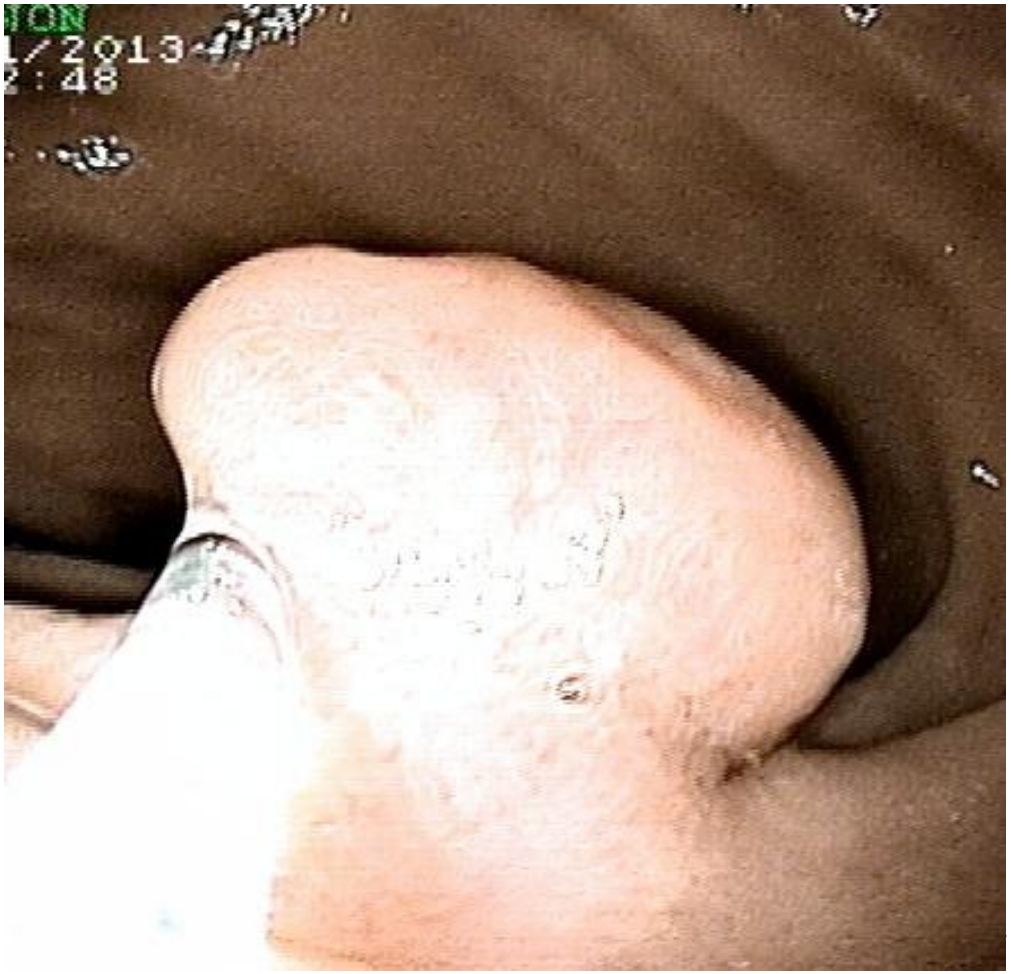

muscularis
mucosa

Diagnosis confirmed eg mucosal pigmentation, FH, genetics

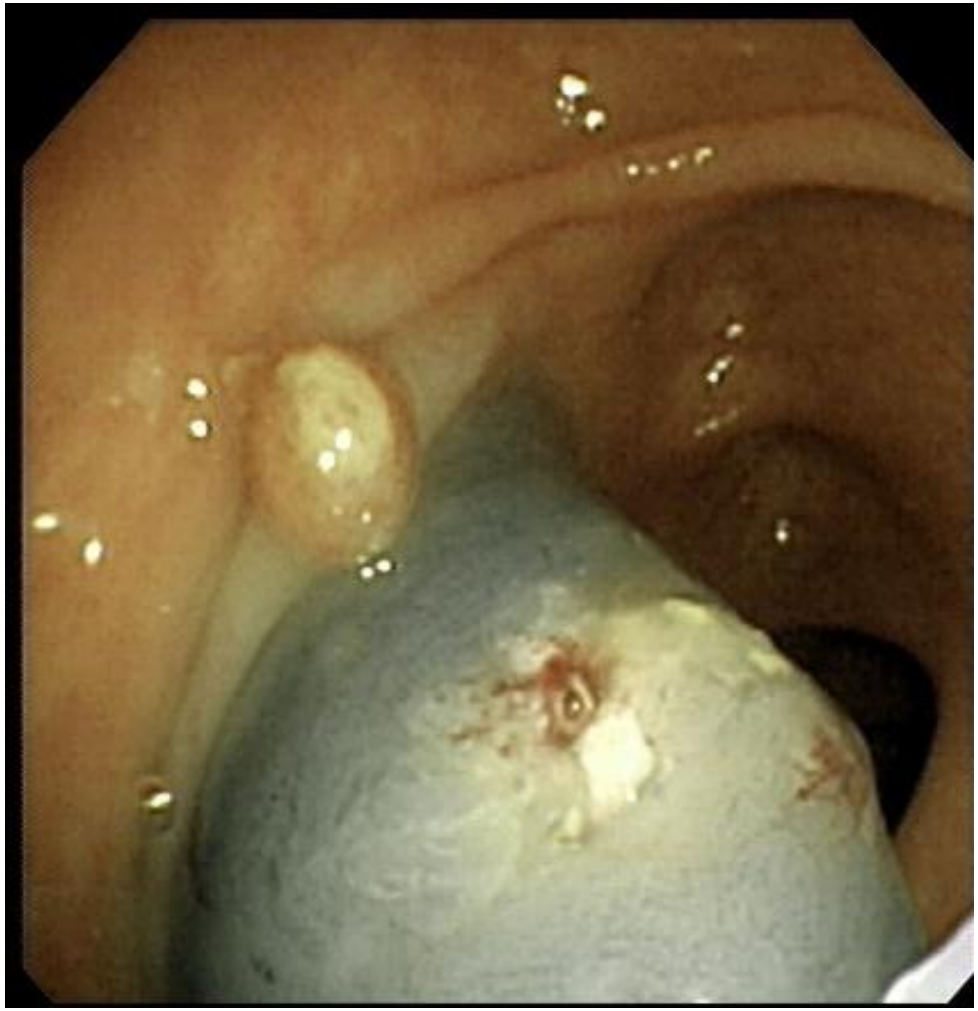
Symptomatic with polyp leading intussusception

Asymptomatic affected child
Start screening age 5-8 years
OGD,
VCE (or MRI)
and colonoscopy

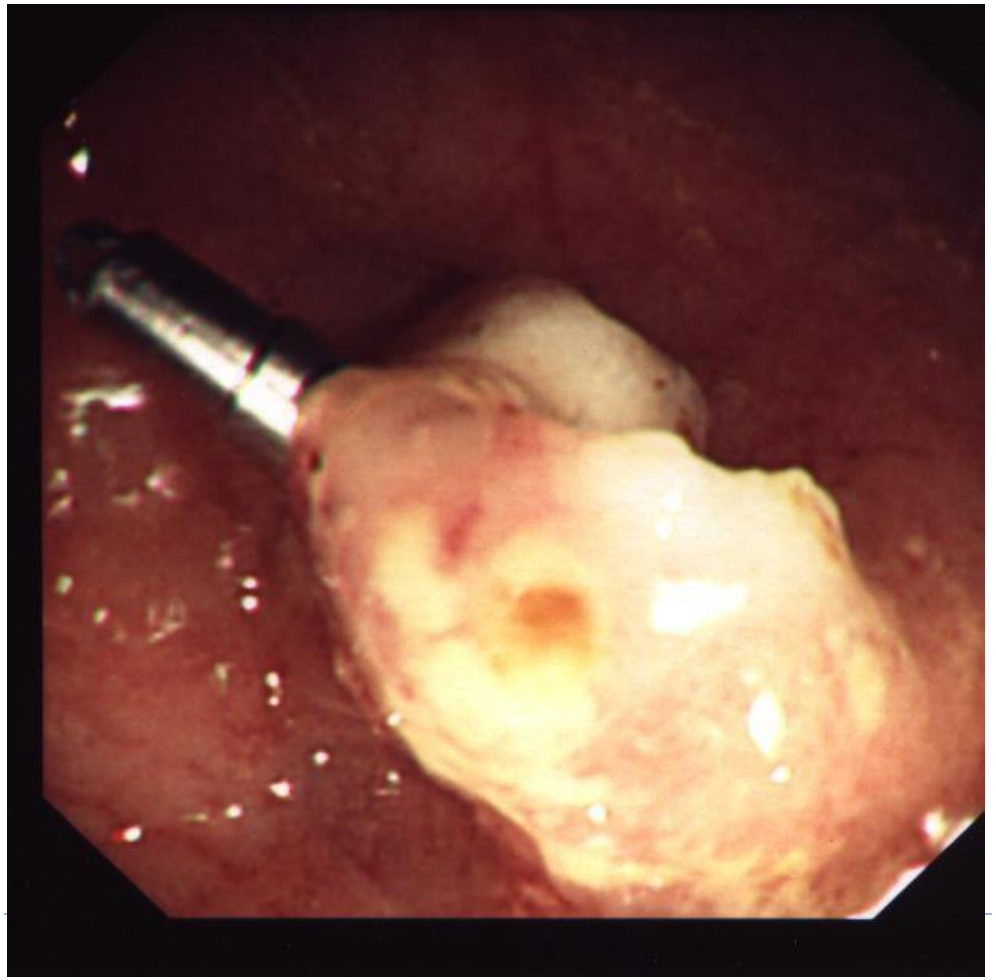
The risk from surveillance is the polypectomy



Post polypectomy – check the site



Clips and loops to reduce complication from polypectomy



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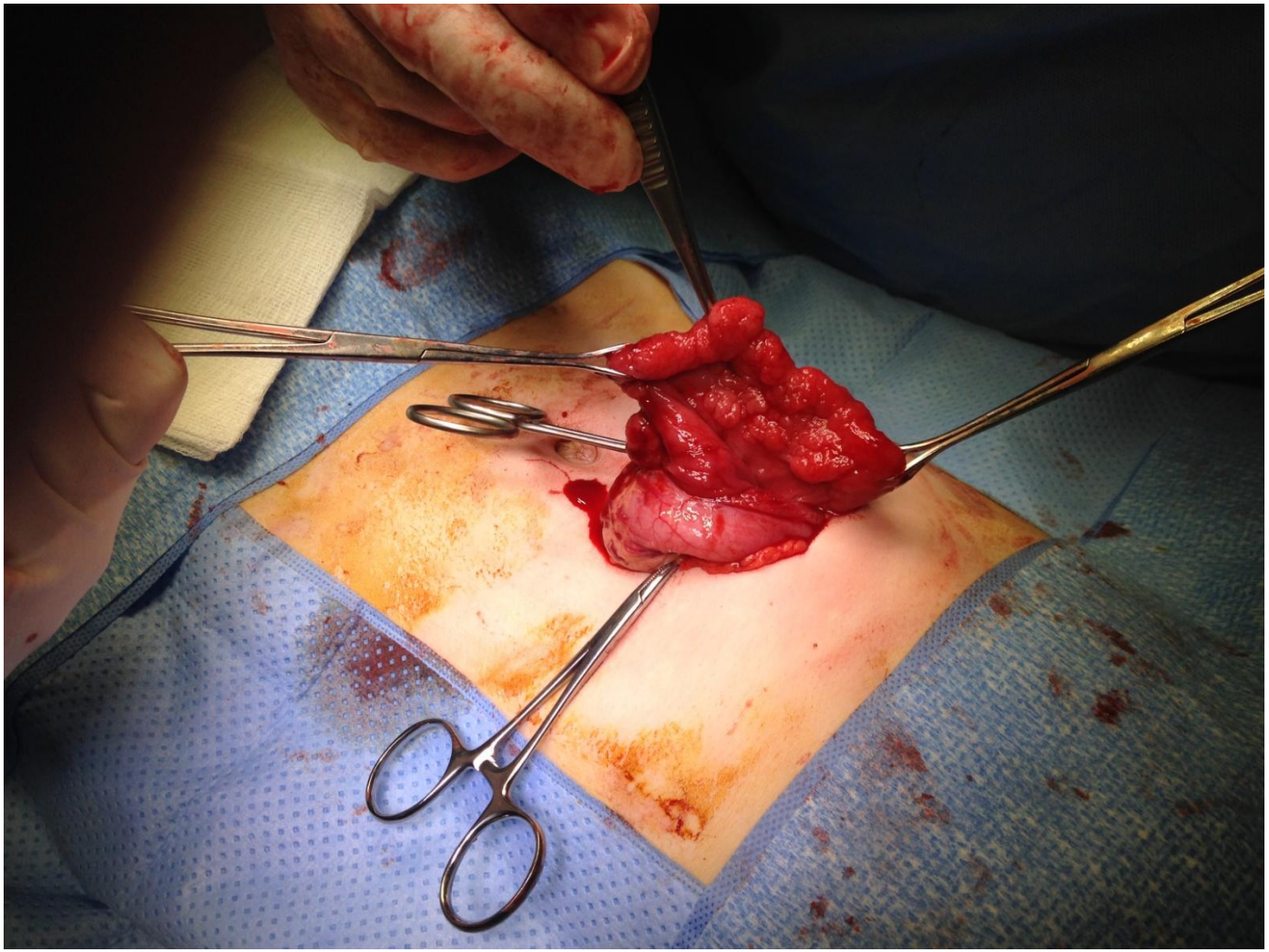
Many polyps are massive. Do not feel you should remove endoscopically



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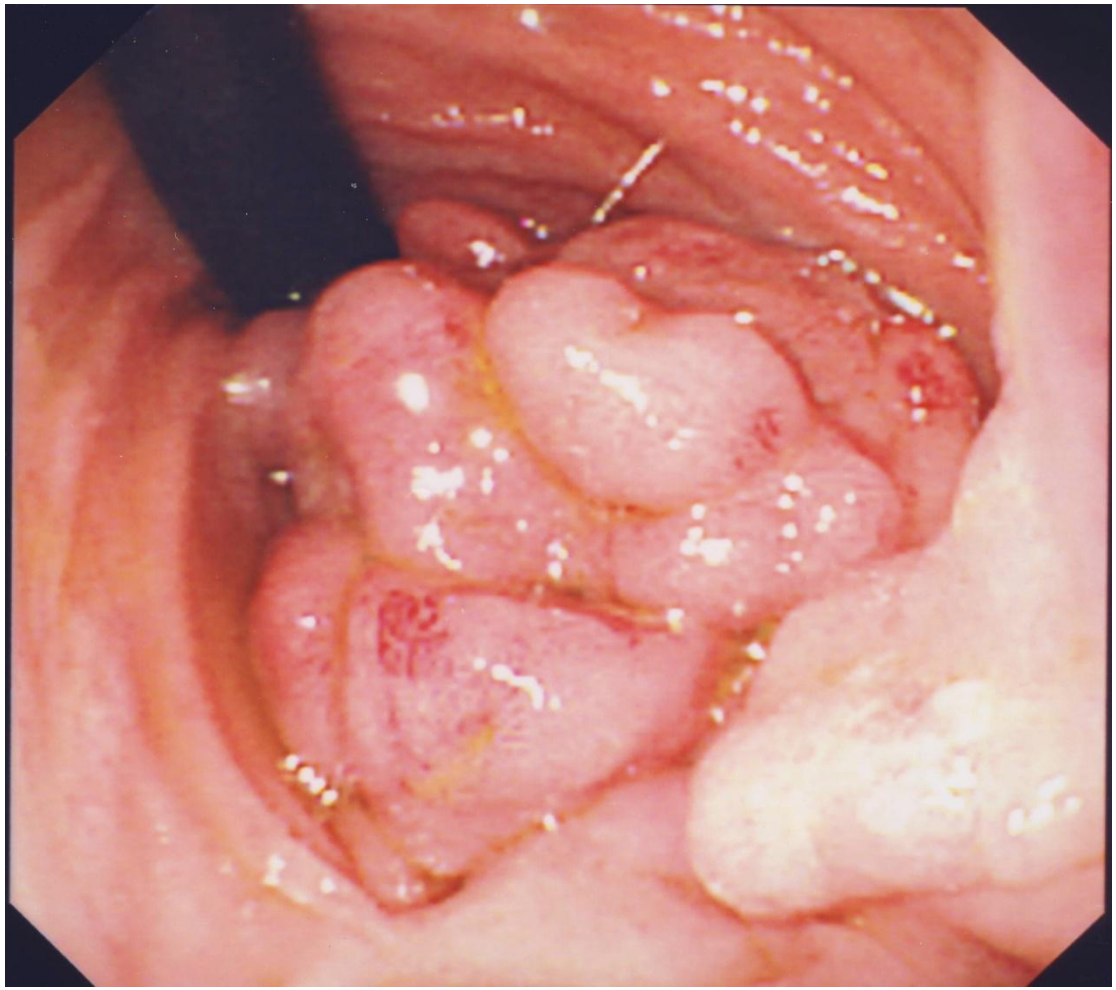
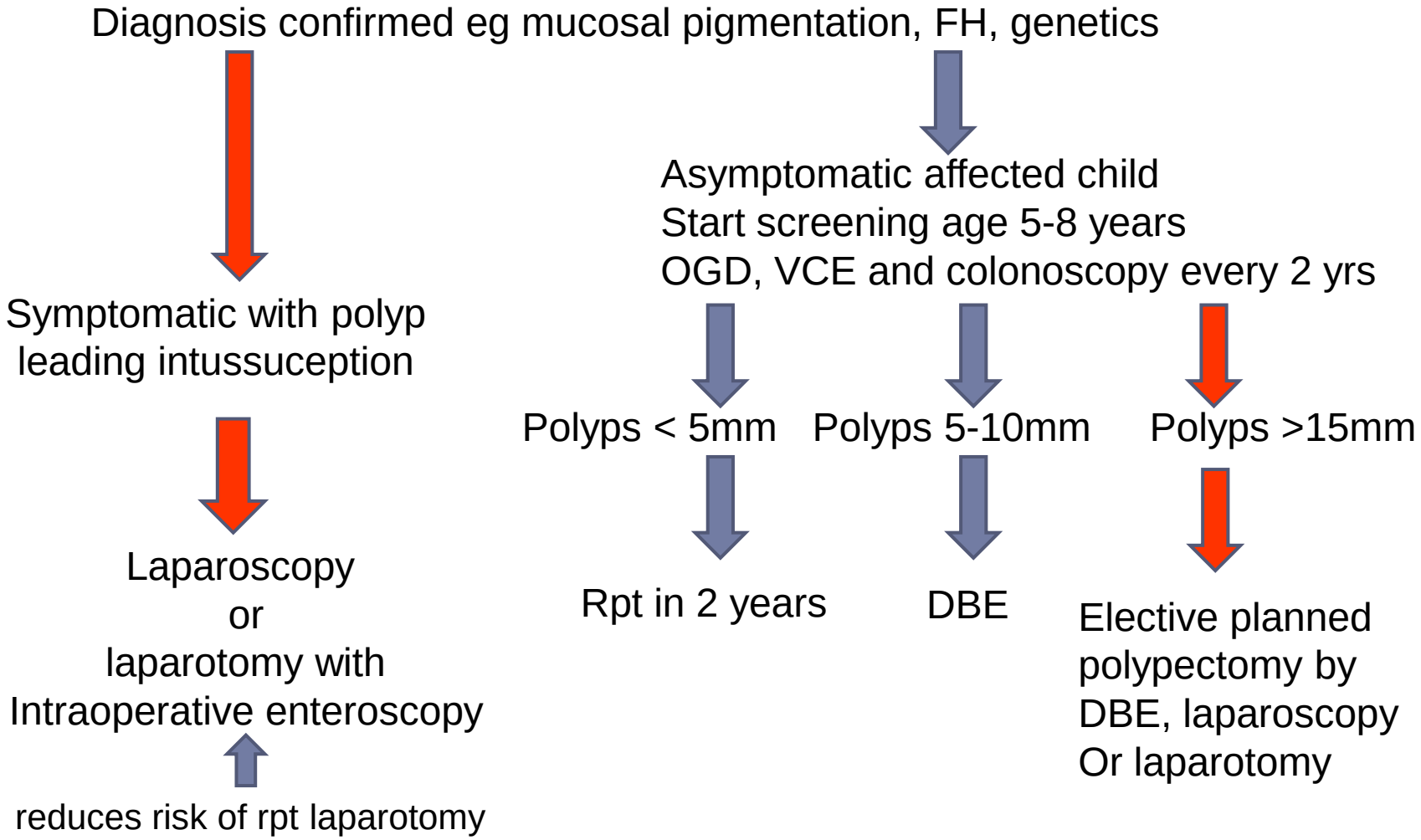
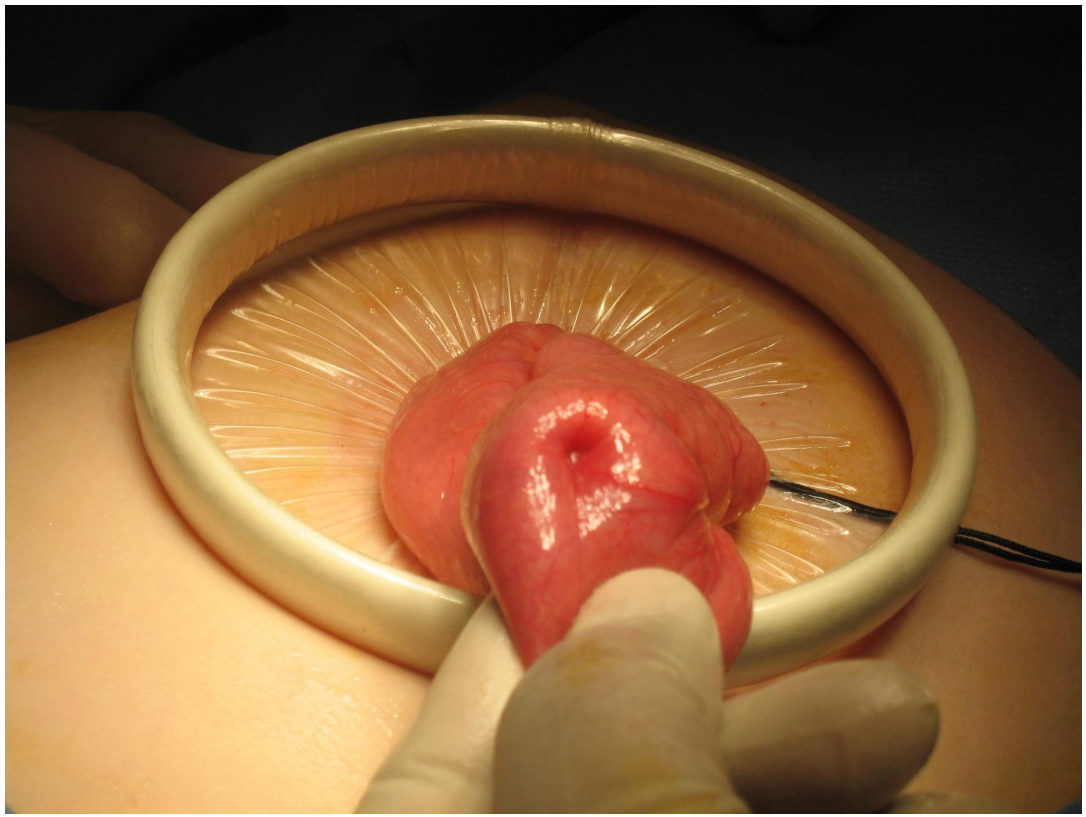
Beware duodenal polyps



Current screening PJS protocol





PJS Bottom Line

Symptomatic needs surgery

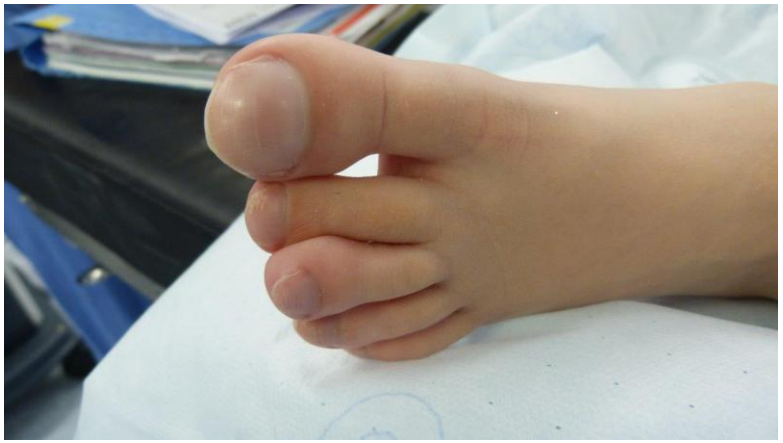
Small bowel surveillance starts at age 5-8 years but respect the risks of polypectomy

Classification of Intestinal Polyps

- Common polyps
  - Juvenile polyps (hamartoma)
- Inheritable polyposis
  - adenomatous polyps
    - familial adenomatous polyposis coli (FAP)
    - MYH-associated polyposis (MAP)
  - hamartomatous polyps
    - Peutz-Jegher's syndrome
    - PTEN hamartoma tumor syndromes
      - Cowden's disease
      - Bannayan-Riley-Ruvalcaba syndrome
    - Juvenile polyposis

Consider a polyposis syndrome in a child with a "juvenile polyp"……if

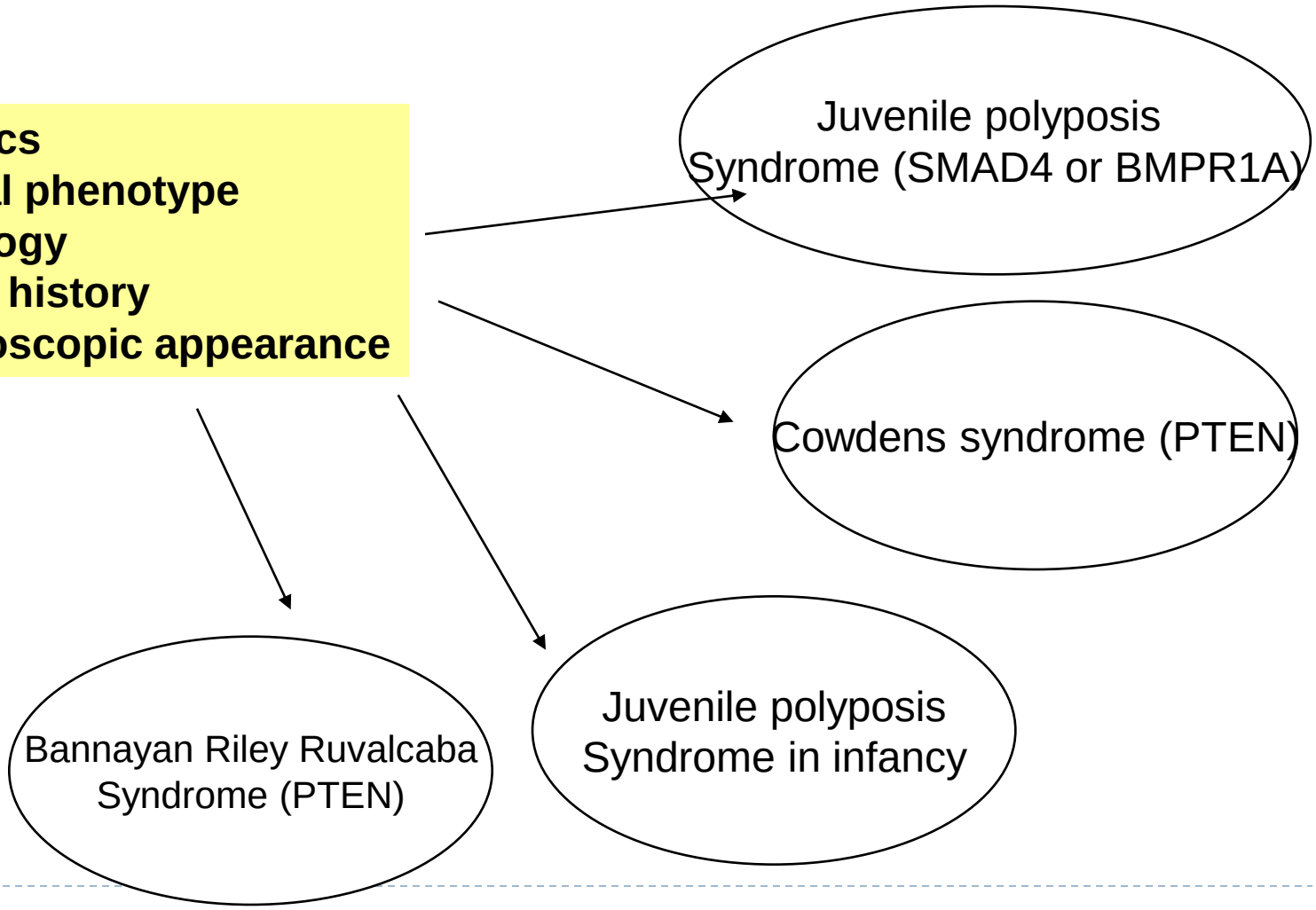
- >3-5 polyps cumulative
- Dysmorphic features – macrocephaly, digital clubbing



- Concerning family history
- Possibility of HHT (hereditary haemorrhagic telangiectasia)

Interpreting hamartomatous syndromes

Genetics
Clinical phenotype
Pathology
Family history
Colonoscopic appearance



Genetic mutation is not identified in 50% of cases

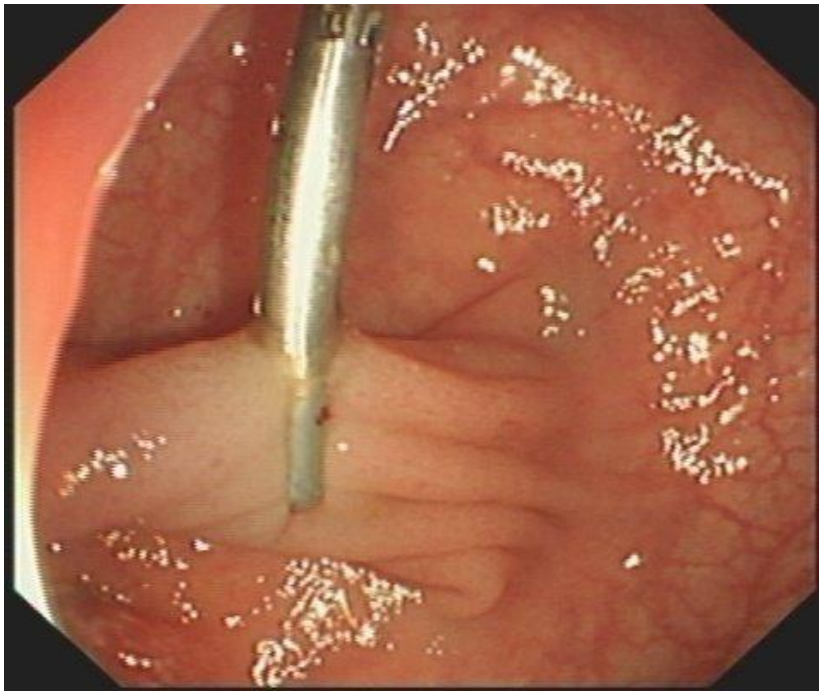
- PTEN
  - 85% of Cowden
  - 65% of Bannayan Riley Ruvalcaba syndrome
- SMAD 4 /HHT
  - 20-50% JPS
- BMPR1A
  - 20-40% of JPS
- ENG
  - 2-5% of JPS, HHT

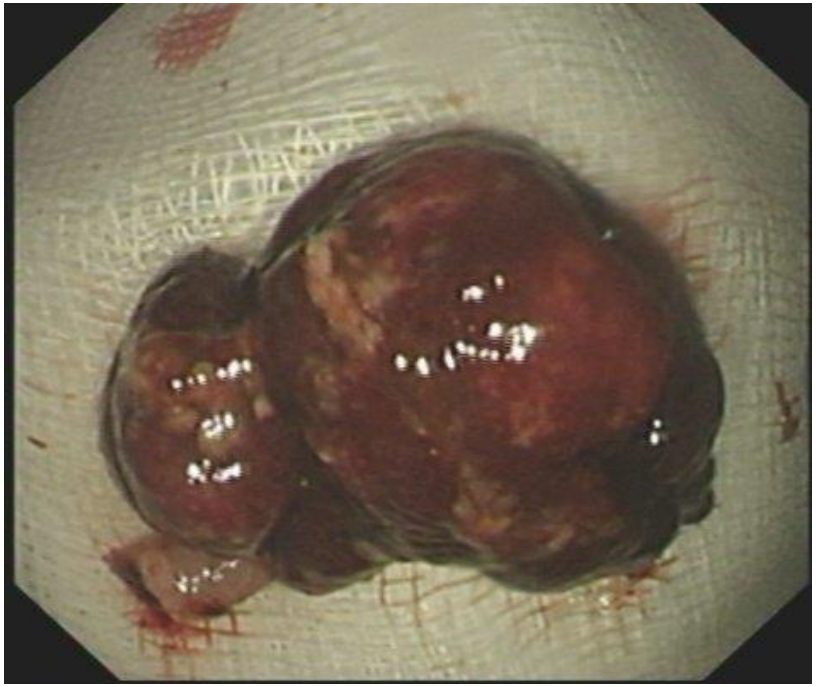
Endoscopic surveillance

- Juvenile polyposis syndrome (SMAD4/BMPR1A)
- 2-3 yearly colonoscopy with polypectomy if bleeding from age about 12-15 years
- Consider Ix for HHT
- GI malignancy risk is very low

- PTEN eg Cowden or BRRS
- Other non GI screening is necessary
- GI malignancy risk is very low

Again – don't underestimate the polypectomy risk





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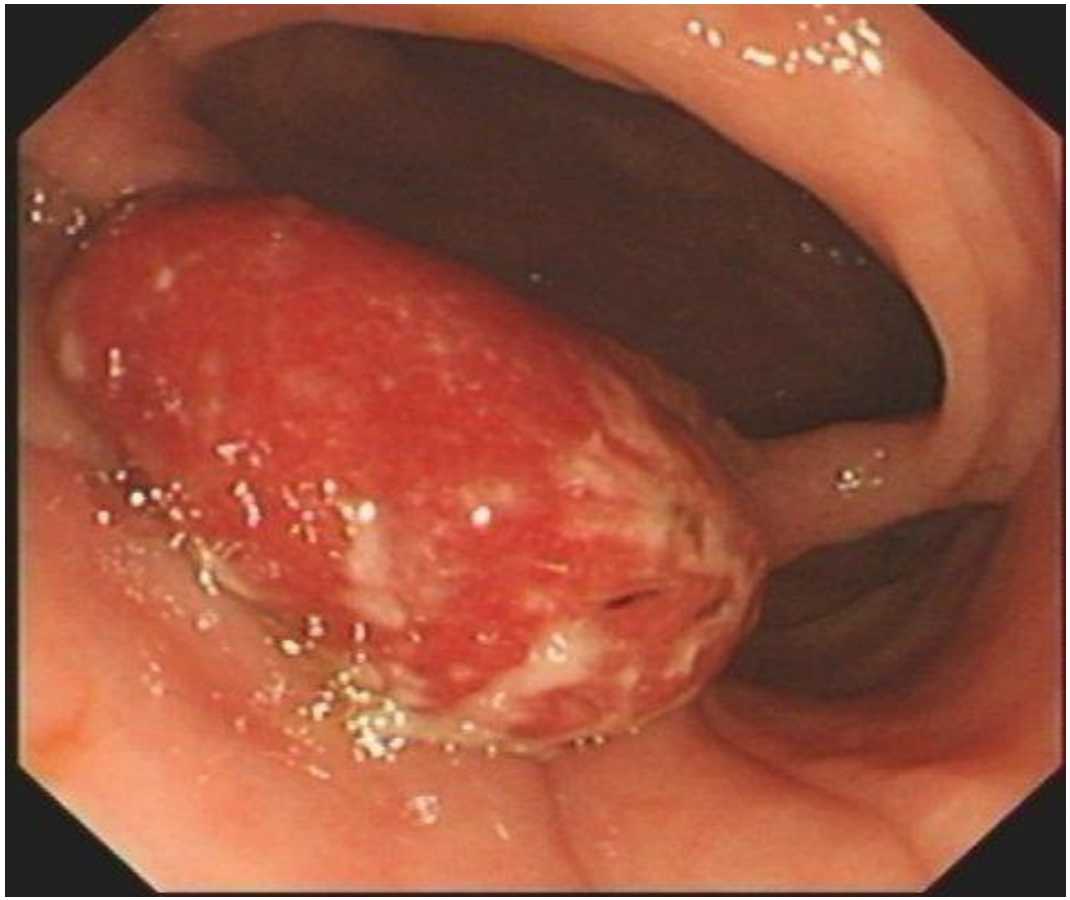
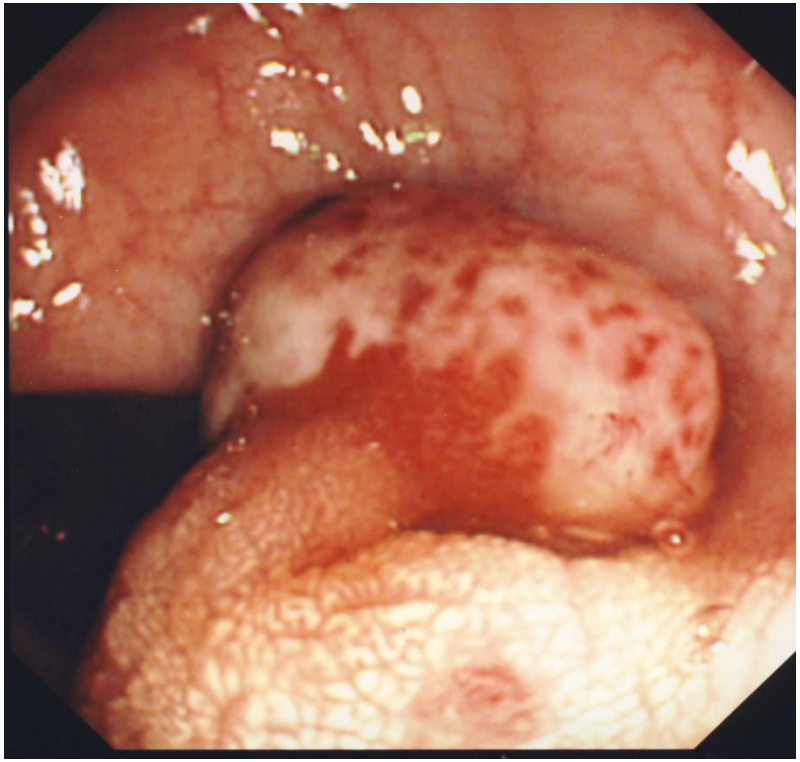


Juvenile polyposis syndrome

Careful assess the phenotype, and interpret genetics with a geneticist

CRC risk is still low

Modify the surveillance according to phenotype, and symptoms



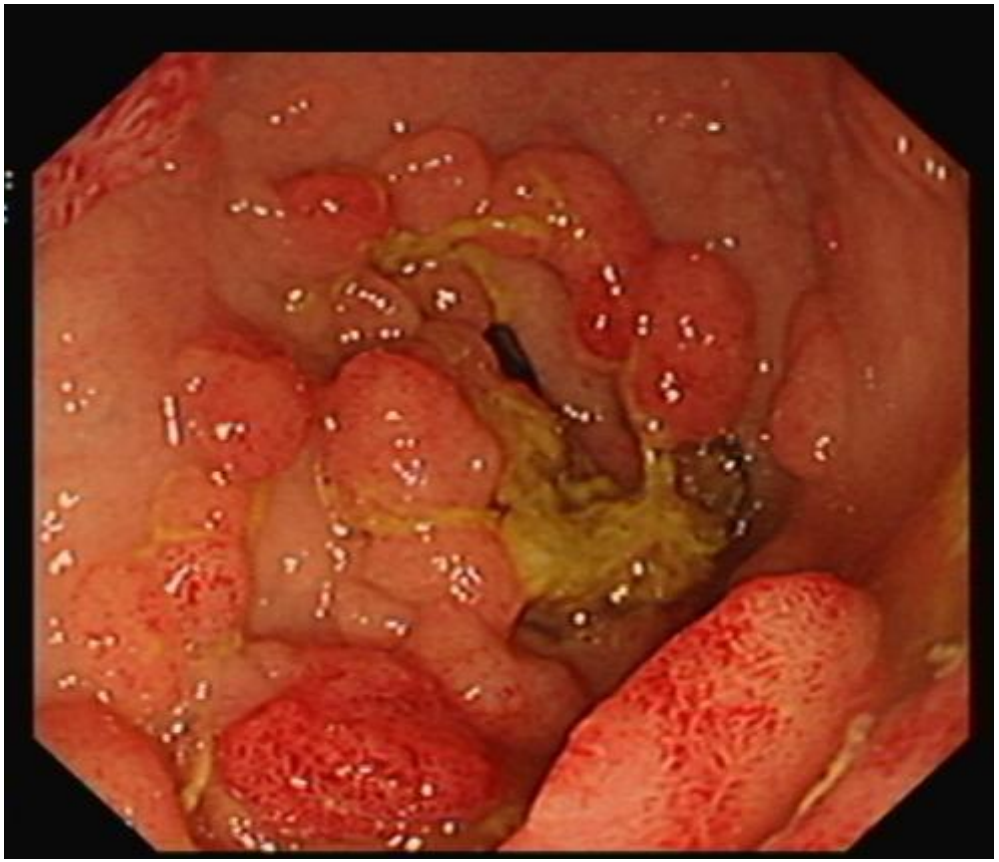
Bottom Line JP

The polyps are benign

Repeat colonoscopy to know if there are more later in life

Consider FH

Rescope if bleeding



Bottom Line FAP

Often wait until teenage years

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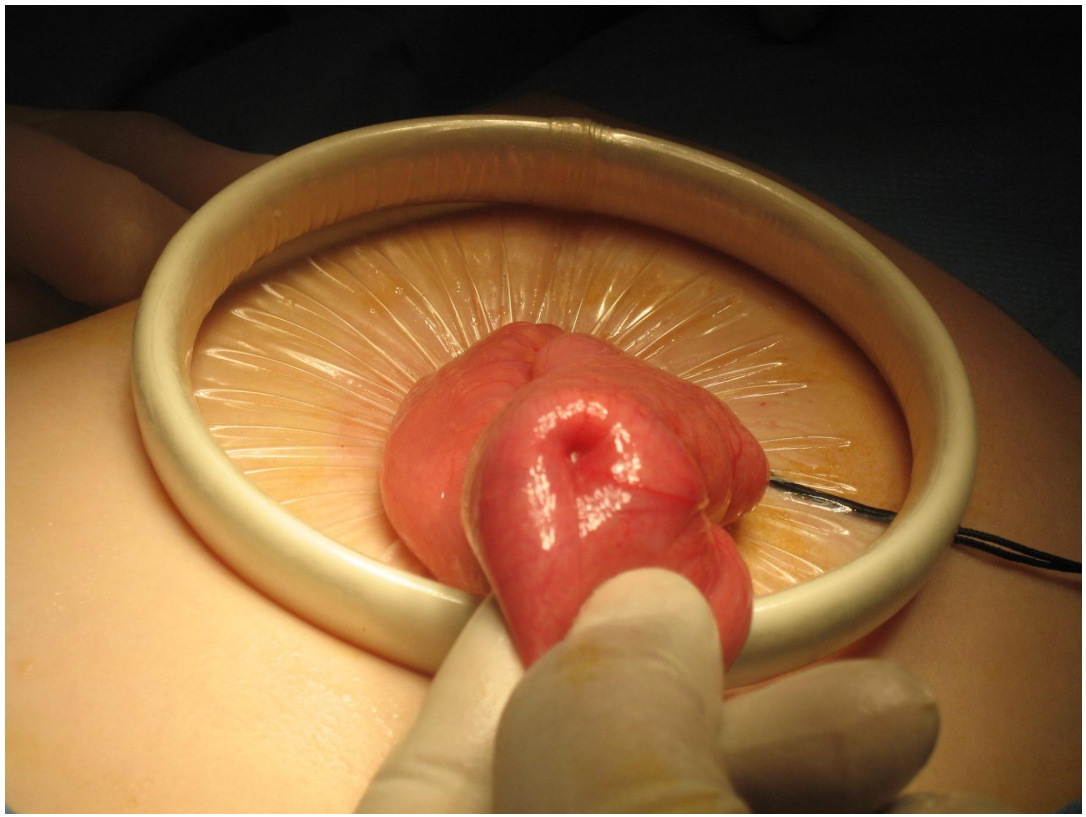
Understand the gene - start earlier if high risk genotype

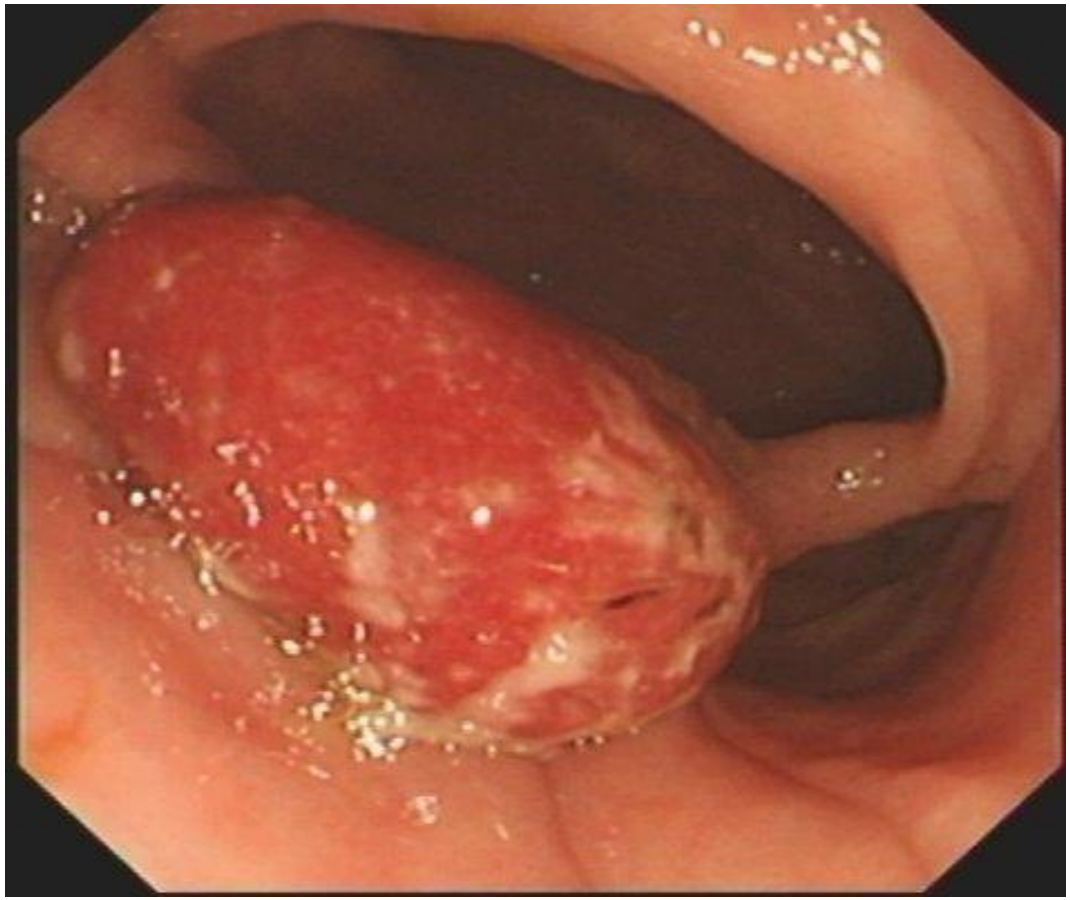
Chemoprevention does not alter timing nor surveillance



Bottom Line PJS

Symptomatic needs surgery

Small bowel surveillance starts at age 5-8 years but respect the risks of polypectomy



Juvenile polyposis syndrome

Careful assess the phenotype, and interpret genetics with a geneticist

CRC risk is still low

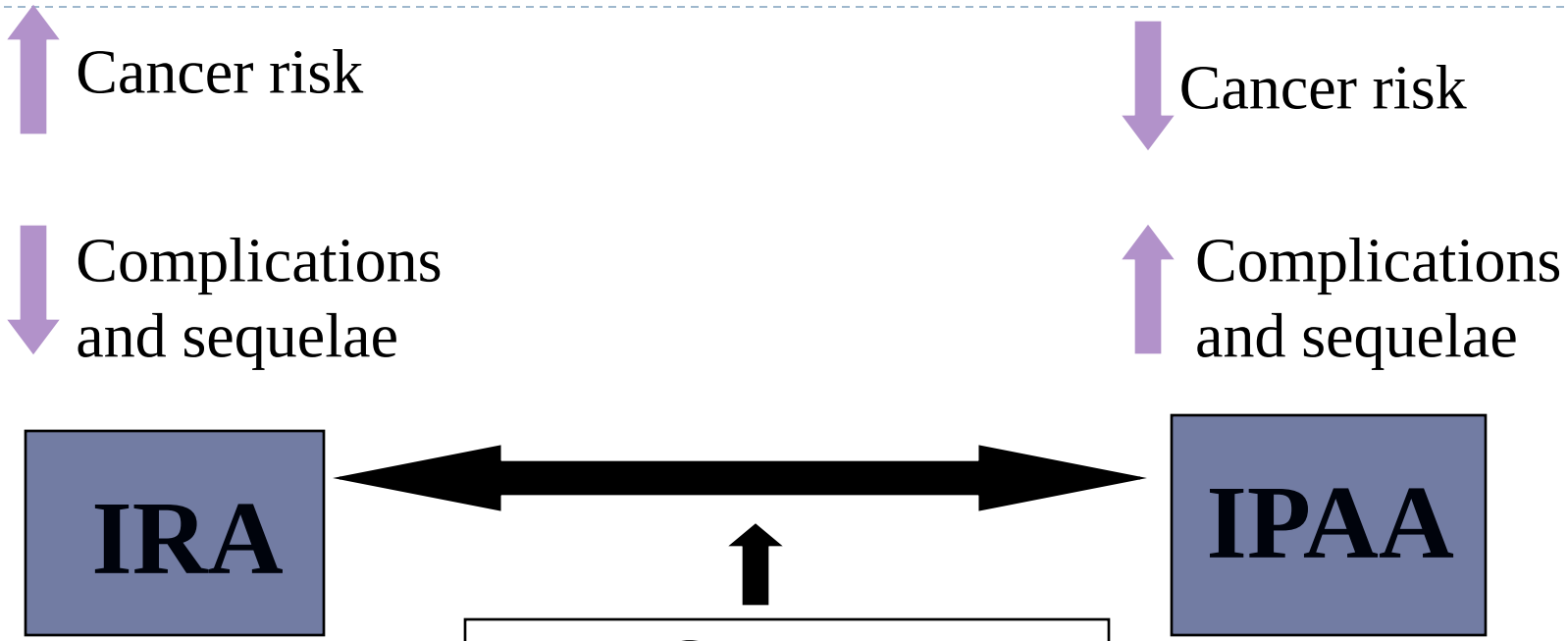
Modify the surveillance according to phenotype, and symptoms

Why surveillance

- Solitary colonic polyp – to avoid missing others
- FAP – to prevent a CRC
- PJS – to prevent a mid gut intussusception and laparotomy
- JPS – to characterise the condition

Be careful with polypectomy esp in PJS. Use the surgeon wisely

Annual colonoscopy impacts on surgical choices



Thank you

UK Polyposis team

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  - Kay Neale and Jackie Hawkins
  - Prof Sue Clark
  - Wolfson Academic Dept of Endoscopy,
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ST MARK'S HOSPITAL