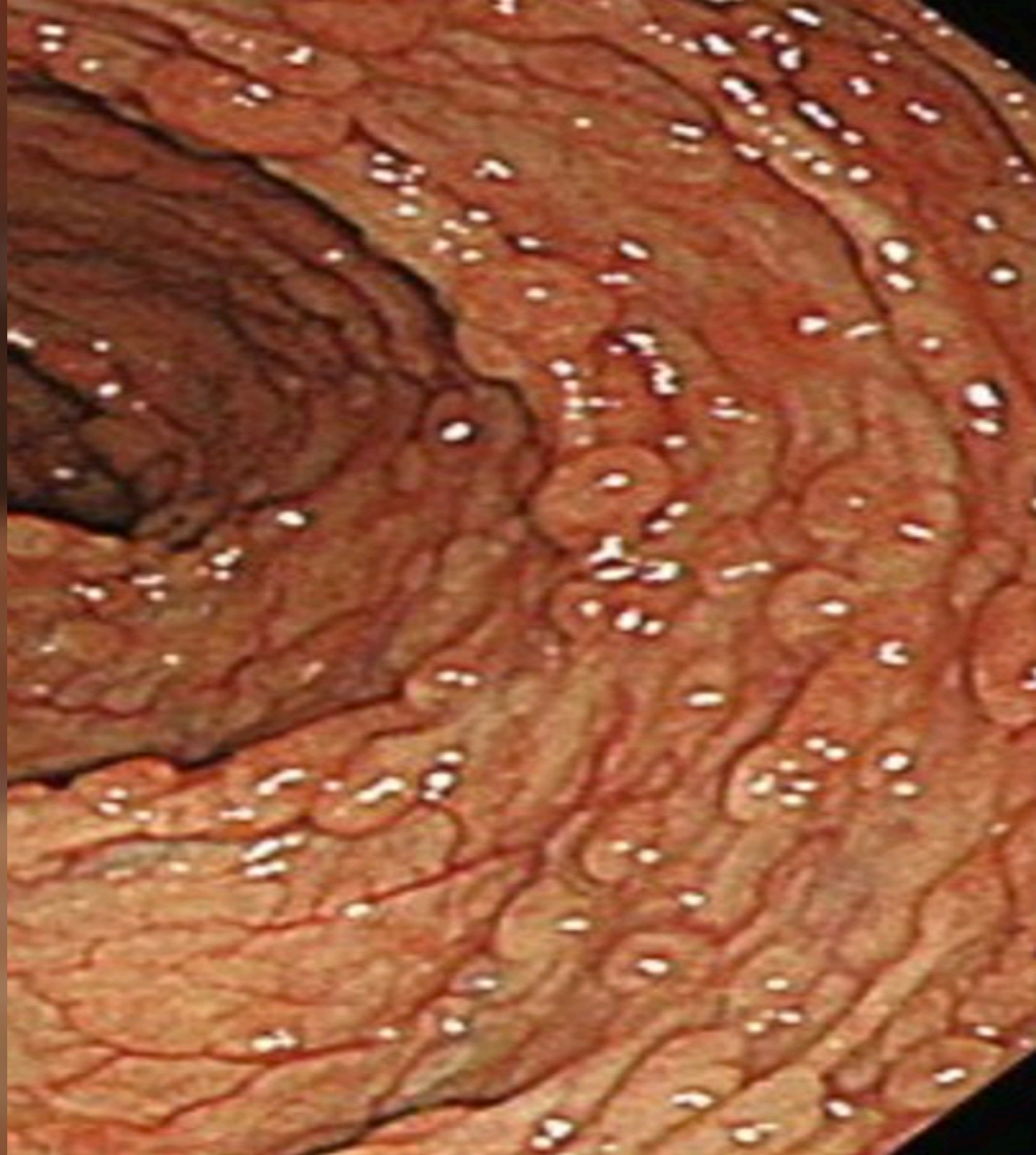
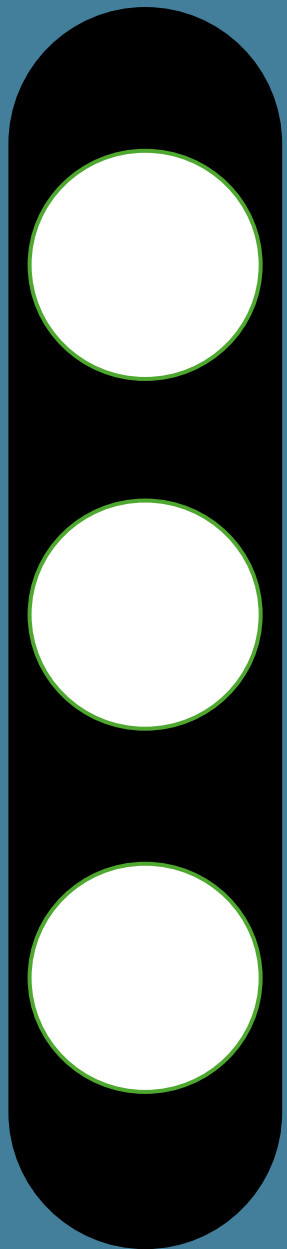


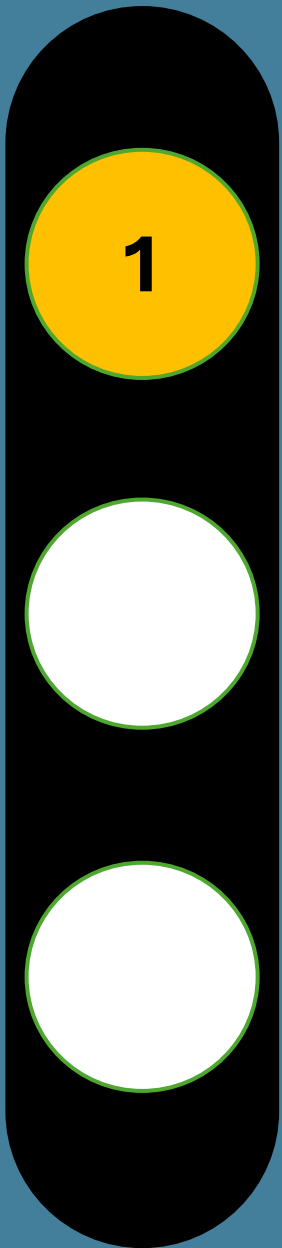
Polyposis Diagnosis

Christopher W. Deitch, MD
Cooper University Hospital





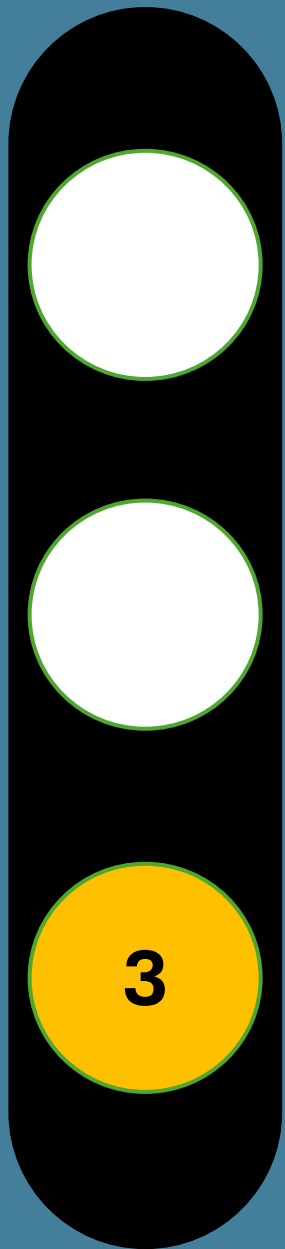
Objectives



Identifying risks especially
In younger populations



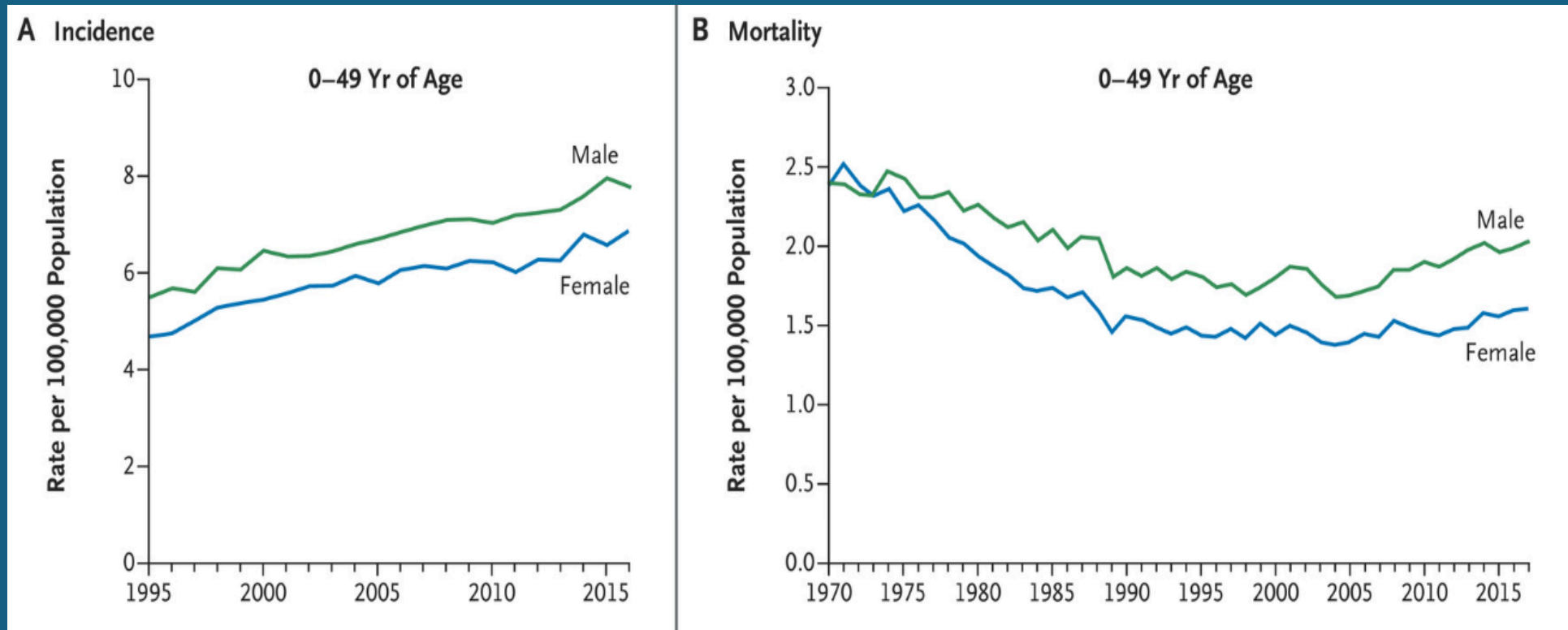
Recognize features of Lynch Syndrome



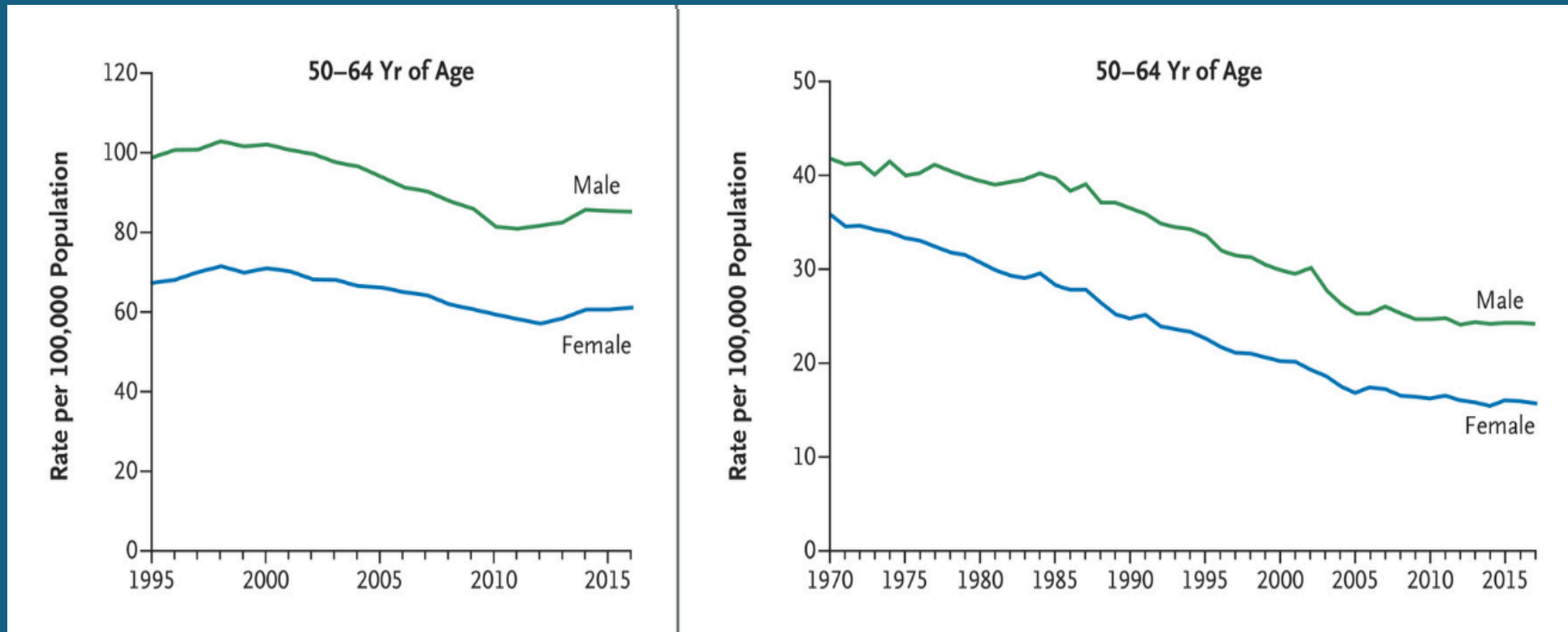
MUTYH Polyposis



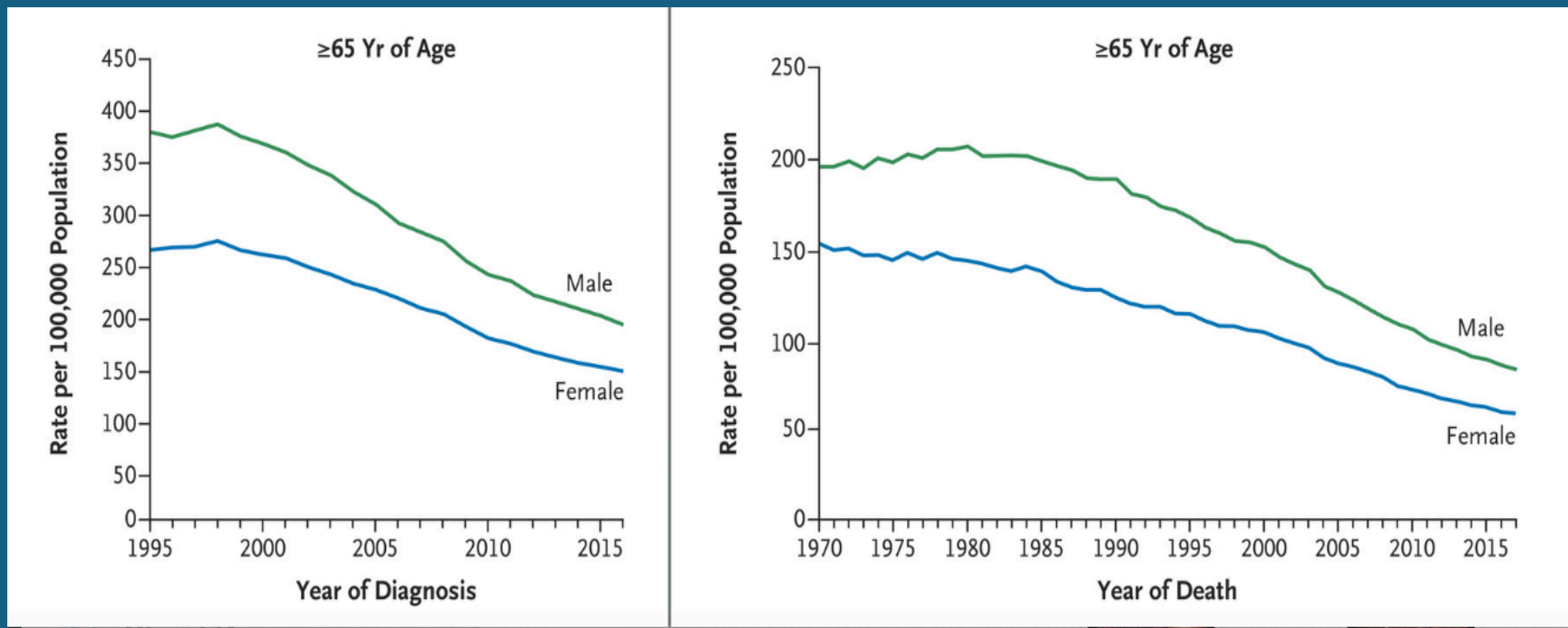
Trends in Colon Cancer Incidence and Mortality



Trends in Colon Cancer Incidence and Mortality



Trends in Colon Cancer Incidence and Mortality



Trends in Colon Cancer Incidence and Mortality

1 FDR	~2.5 X risk
FDR < 45 years old	~3.9X risk
> 1 FDR with CRC	~4.25X risk

~25% patients with CRC have a significant FH
FDR with advanced adenoma: Screen as a FDR
with colon cancer

Trends of early onset colorectal cancer

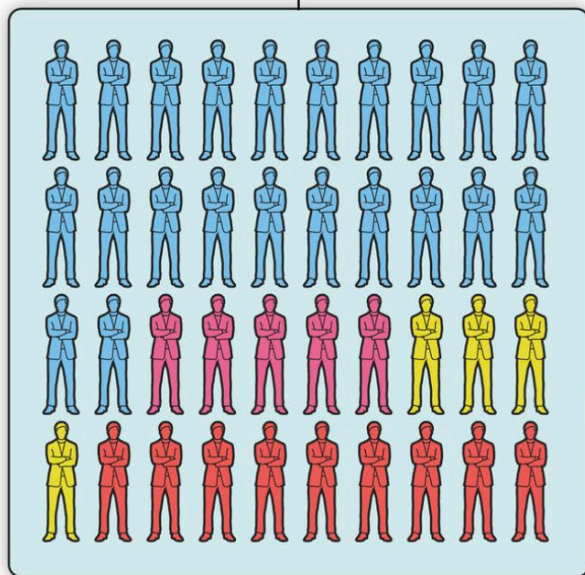
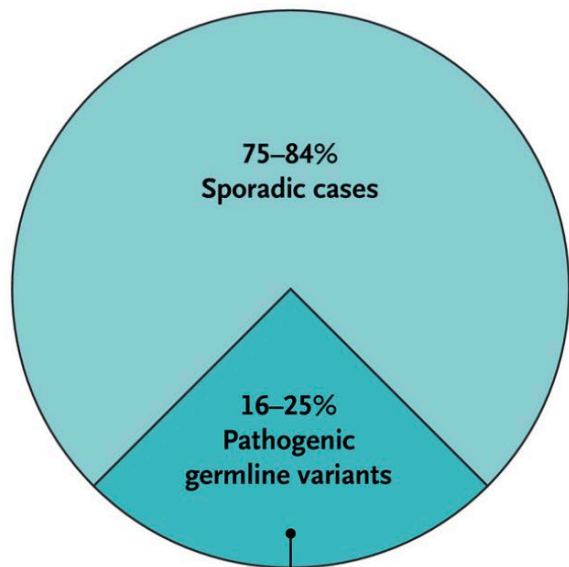
70% left colon

More advanced on presentation

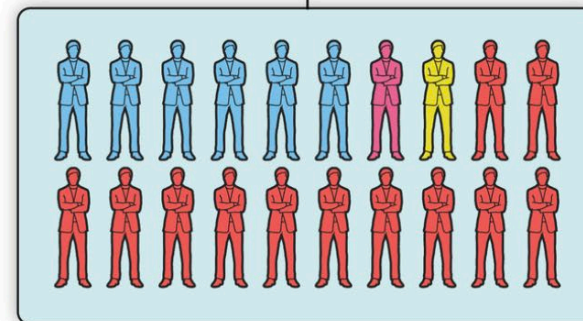
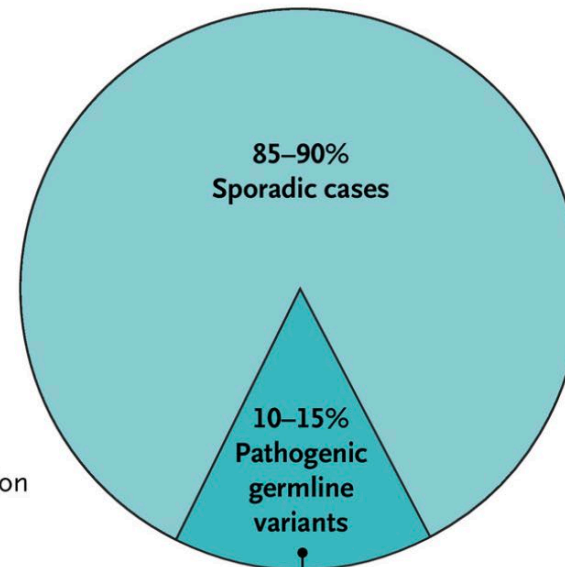
MSI-High (microsatellite Instability)

Microbiome

CRC: Early onset



CRC: All ages



- Lynch syndrome
- APC mutation
- Biallelic *MUTYH* mutation
- Other mutations

Polyp Syndromes

Familial Adenomatous polyposis

Lynch Syndrome

Li Fraumeni

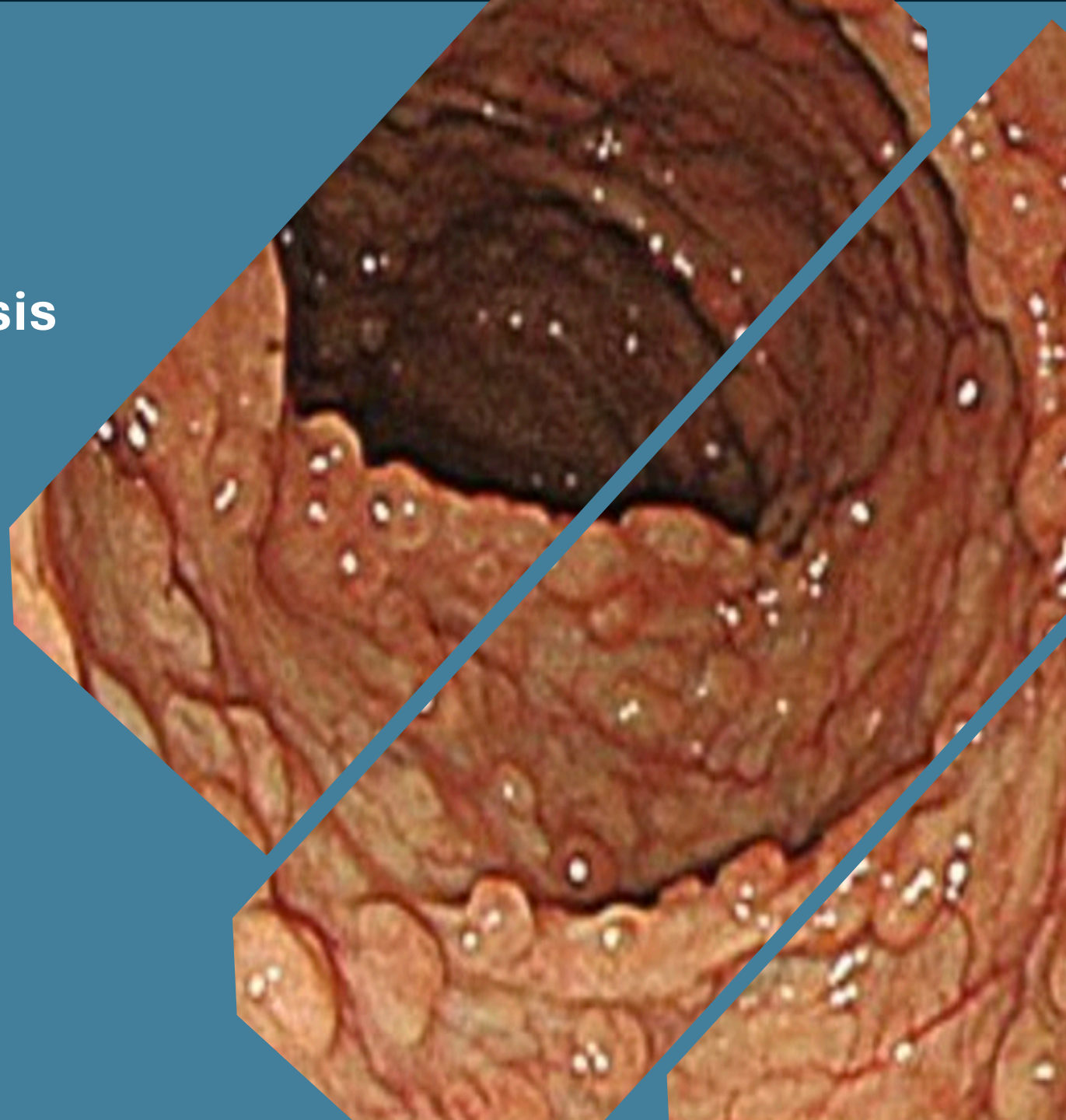
MUTYH associated polyposis

Serrated Polyposis

Juvenile Polyposis

Peutz-Jeghers

PTEN associated polyposis



Illustrations

39 year old woman undergoes a colonoscopy for bloating and rare passage of blood

38 year old woman reads a pamphlet awaiting her chemotherapy for uterine sarcoma while sitting in her GYN/ONC waiting room realizes the significance of her mother, maternal uncle and maternal grandfather dying from colon cancer all before the age of 55

Illustrations

39 year old found to have a stage 2A sigmoid colon cancer. A detailed family history fails to reveal any relatives afflicted with cancer

38 year old woman walks over to the oncology office and asks for a colonoscopy. A right sided ulcerated mass is discovered

Amsterdam I Criteria

3 Family Members with colon cancer
2 Affected generations
1 under age 50





Amsterdam II Criteria

**3 Family members with any HNPCC
related cancer**

2 Affected generations

1 under age 50





Bethesda Criteria

Colorectal cancer < 50

**Synchronous or metachronous
colon or HNPCC related cancer
regardless of age**

CRC MSI-H <60 years old

**CRC in first degree relative with HNPCC
related cancer diagnosed < 50**

**CRC in 2 FDR or SDR with HNPCC
related cancer of any age**





PREMM (Predicted Model for gene Mutations)

93% Sensitivity

99% Specific

Outperforms MMRpro and MMRpredict



Mutations



MLH 1



MSH 2



MSH 6



PMS 2



EPCAM

Screening our Lynch Population

Colonoscopy / Chromoendoscopy

Age 20-25

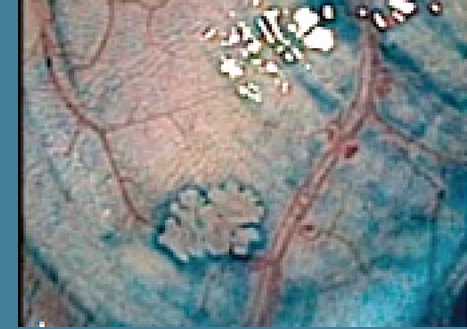
Every 1-2 years

Screening our Lynch Population

Colonoscopy / Chromoendoscopy

Age 20-25

Every 1-2 years



Screening our Lynch Population

Colonoscopy / Chromoendoscopy

Age 20-25

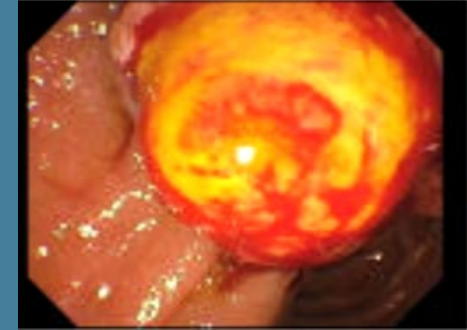
Every 1-2 years



Upper Endoscopy

Age 20-25

Every 2-3 years



Screening our Lynch Population

Colonoscopy / Chromoendoscopy

Age 20-25

Every 1-2 years

Upper Endoscopy

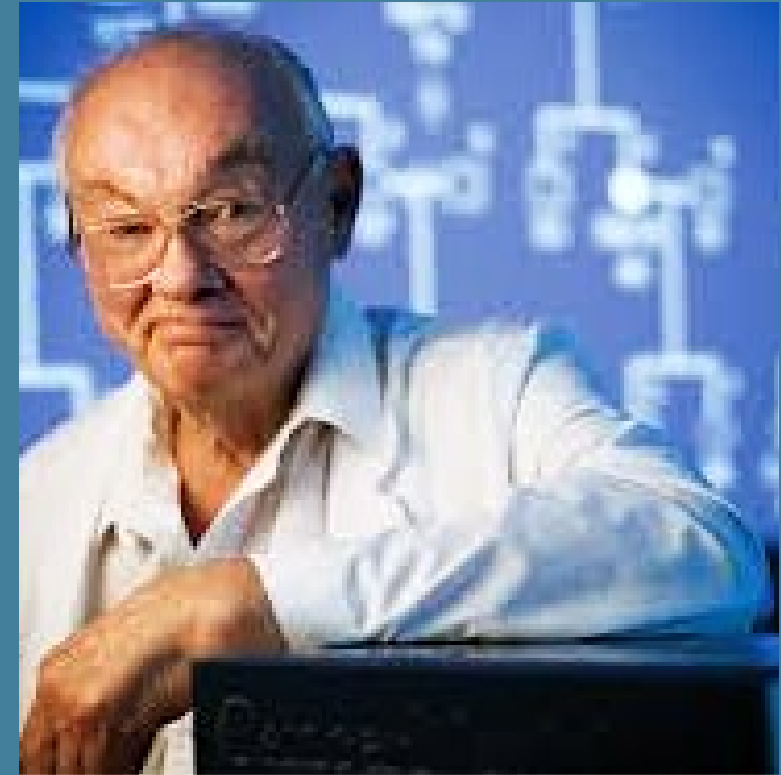
Age 20-25

Every 2-3 years

Small Bowel Screening

Pancreatic Cancer Screening

Urologic Screening



Familial Colorectal Cancer Syndrome X

Fulfill criteria for possible genetic mutations

Patients have no identified genetic alteration

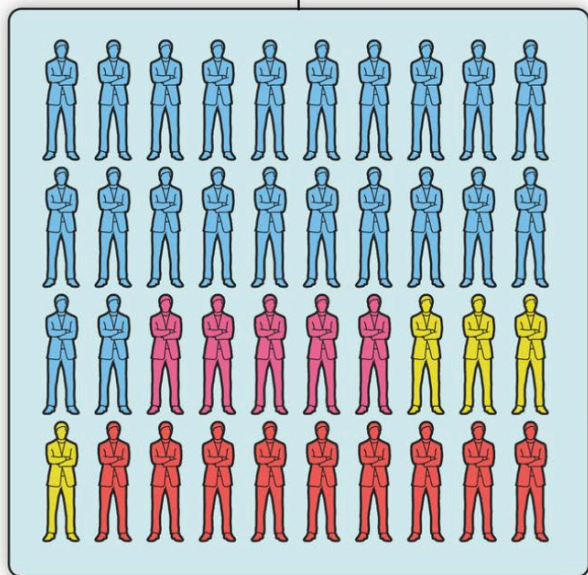
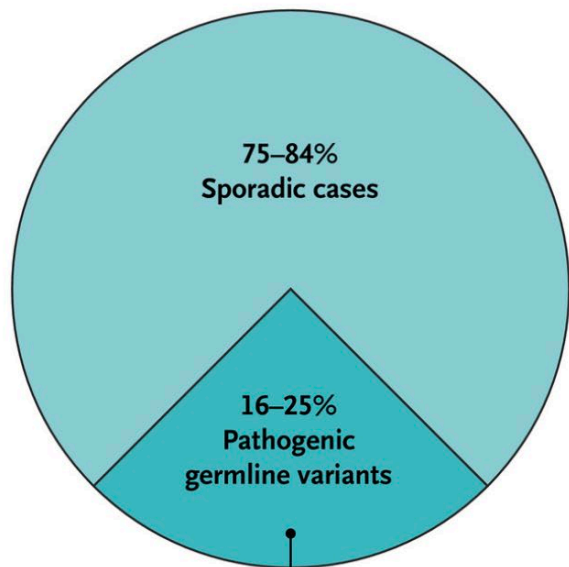
Cancers develop later than Lynch Syndrome

Fewer extracolonic cancers

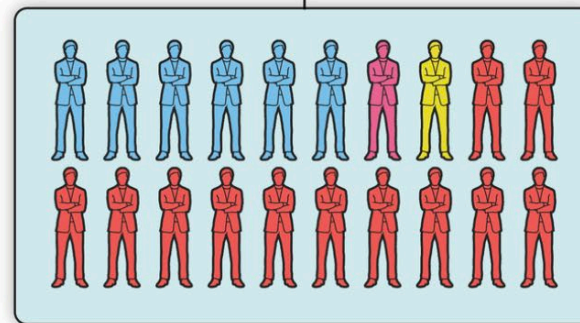
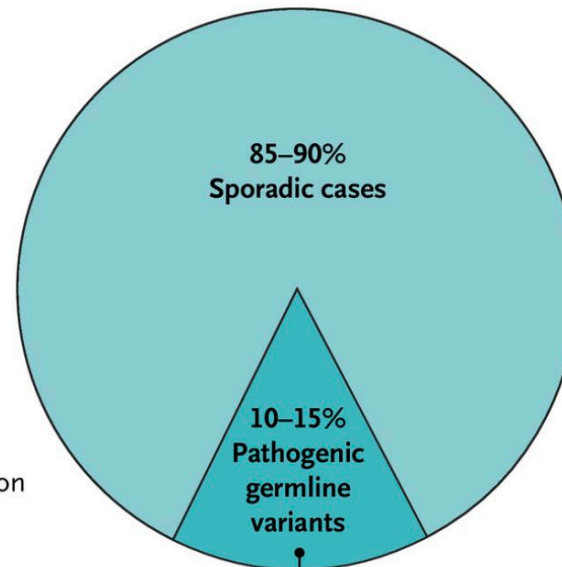
Left side > Right side

Pathologically distinct

CRC: Early onset

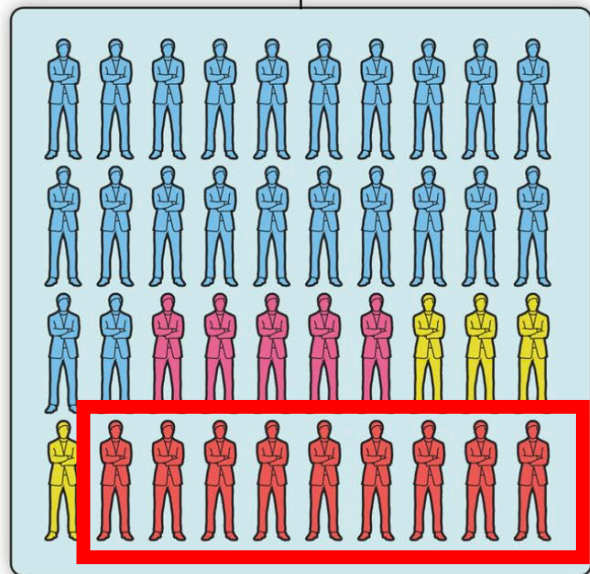
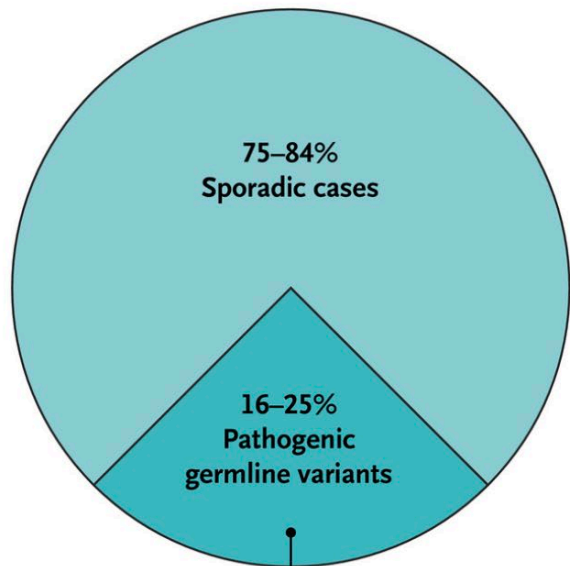


CRC: All ages

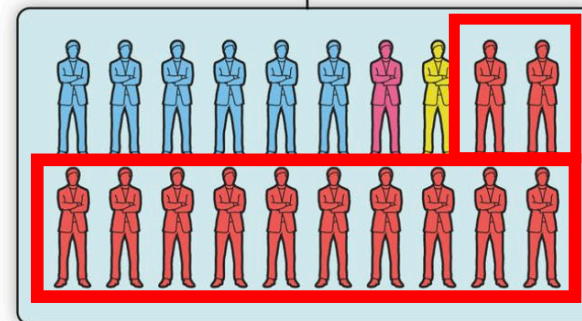
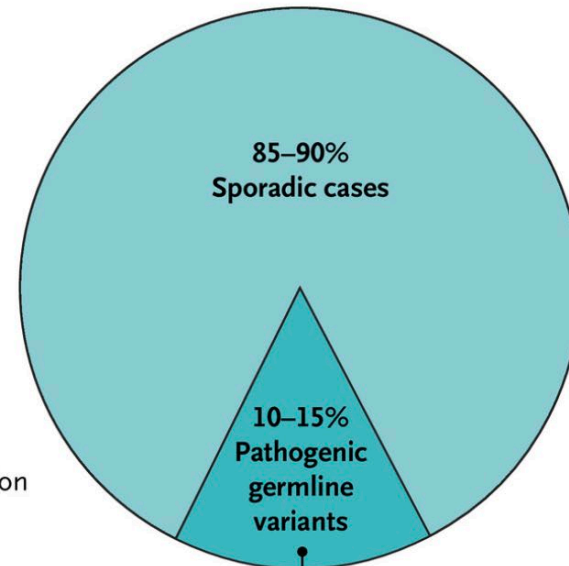


- Lynch syndrome
- APC mutation
- Biallelic *MUTYH* mutation
- Other mutations

CRC: Early onset

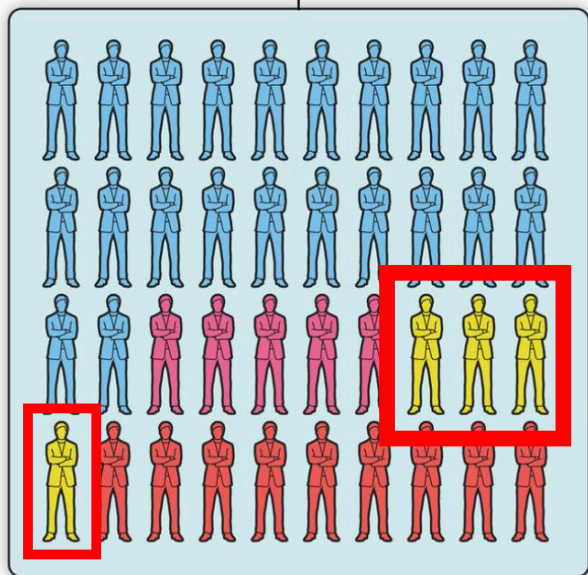
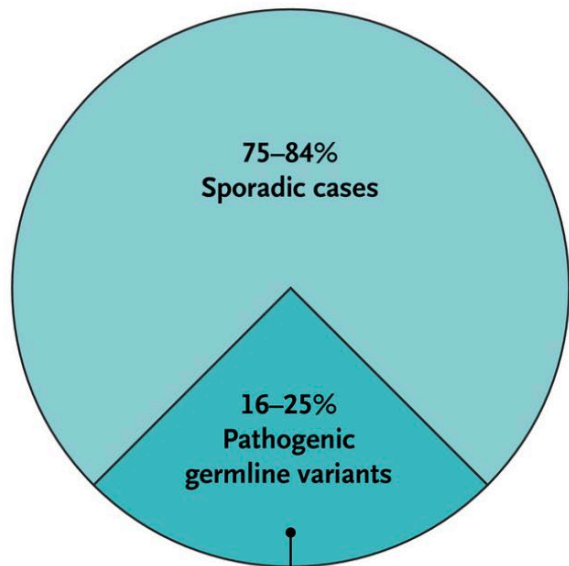


CRC: All ages

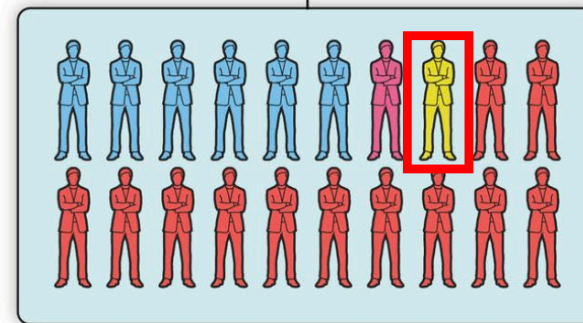
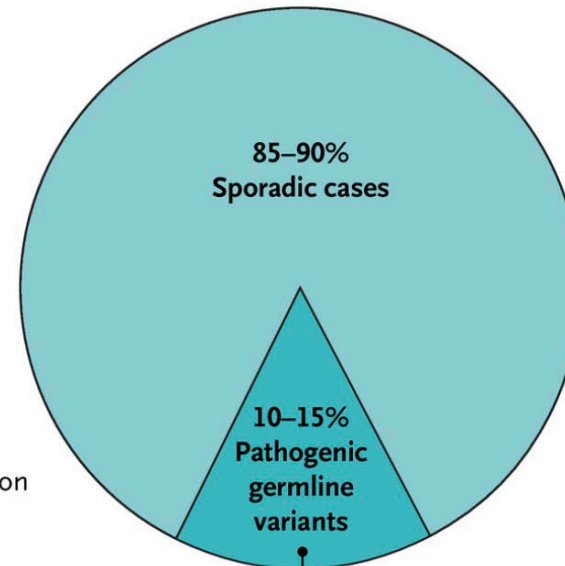


- Lynch syndrome
- APC mutation
- Biallelic *MUTYH* mutation
- Other mutations

CRC: Early onset



CRC: All ages



- Lynch syndrome
- APC mutation
- Biallelic *MUTYH* mutation
- Other mutations

MUTYH Associated Polyposis

10-100 Adenomatous or sessile serrated polyps

Gastric and Duodenal Polyps

Increase risk of duodenal, bladder, ovarian and thyroid cancers

26% Patients with multiple adenomas

Concomitantly found with other genetic mutations (APC)

MUTYH Associated Polyposis

When to consider MUTYH

10 adenomas on one exam

10 adenomas AND colon cancer

Cumulative > 15 adenomas

Adenomas, Traditional Serrated, Sessile Serrated or Hyperplastic

Colon cancer risk 40-65%

Older onset than Lynch Syndrome

Colonoscopy every 1-2 years

OK, What did I forget?



"Thanks for almost everything, Dad."

OK, What did I forget?

Overlap of MUTYH with Serrated Polyposis Syndrome

Pay attention to the other, less common polyp syndromes

Be a diligent endoscopist

Adenoma Detection Rates

Have suspicion

Thank you, organizers

Thank you, colleagues and friends

References

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JAMA 2021; 4(11)e 2134308
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