

Lynch Syndrome in Young Hispanic Male

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OBJECTIVES

- Inform clinicians about the rising incidence of early onset colorectal cancer (EO-CRC).
- EO-CRC is a rising concern but is disproportionately affecting young Hispanic adults when compared to non-Hispanic Whites (NHW).

BACKGROUND

- There has been nearly a 40% increase in the incidence of colorectal cancer (CRC) in young adults from 1992-2014.
- There has yet to be any identifiable causes of the increase incidence of EO-CRC.
- In 2018, there were around 26.5 million Hispanics in the U.S. between the ages of 20-29-year-old age group and CRC incidence increased 90% within this age group.

THE CASE

- A 25-year-old Hispanic male with a prior medical history of childhood seizures presented with sharp mid-epigastric abdominal pain that radiated to his right lower quadrant. An abdominal CT-scan was completed with findings consistent with perforated appendicitis and reactive thickening of the terminal ileum and cecum. He was discharged on oral antibiotics with directions to obtain a repeat CT-scan.
- The patient was readmitted two months later for worsening abdominal pain and 10-pound weight loss. A repeat abdominal CT-scan showed increased inflammation of the terminal ileum. Gastroenterology was consulted and thought his CT-findings were due to prior appendicitis or a new diagnosis of inflammatory bowel disease, but a colonoscopy was not pursued due concern for perforation. The patient was prescribed antibiotics with plans to undergo colonoscopy as an outpatient, which was delayed secondary to the COVID-19 pandemic and lack of insurance.

IMAGING AND FIGURES

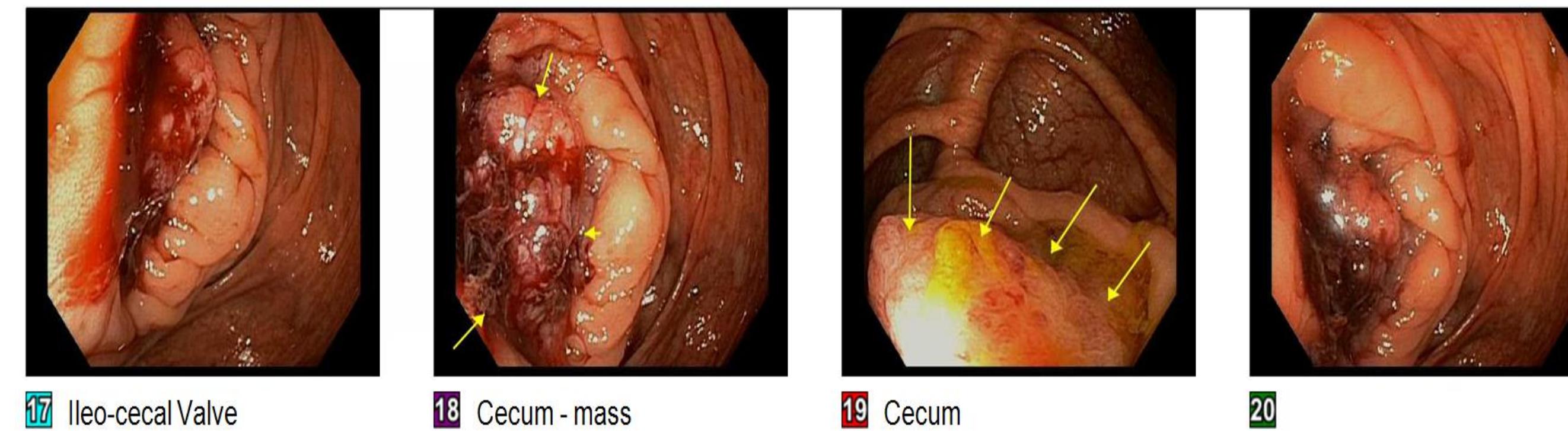


Figure 1: Colonoscopy showing obstructing, fungating mass in the distal cecum.

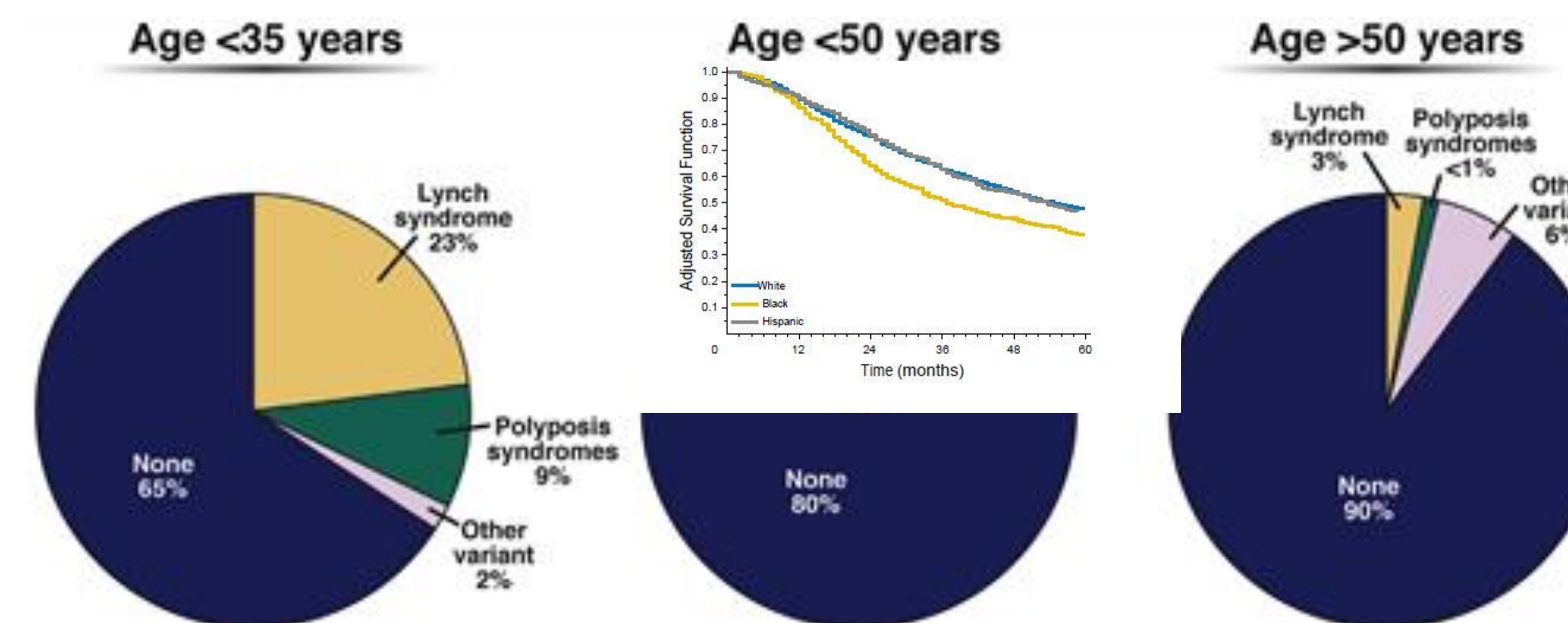


Figure 2: Prevalence of pathogenic variants by age at CRC diagnosis (Image from Stoffel EM, Murphy CC. Epidemiology and Mechanisms of the Increasing Incidence of Colon and Rectal Cancers in Young Adults. *Gastroenterology*. 2020;158(2):341-353.)

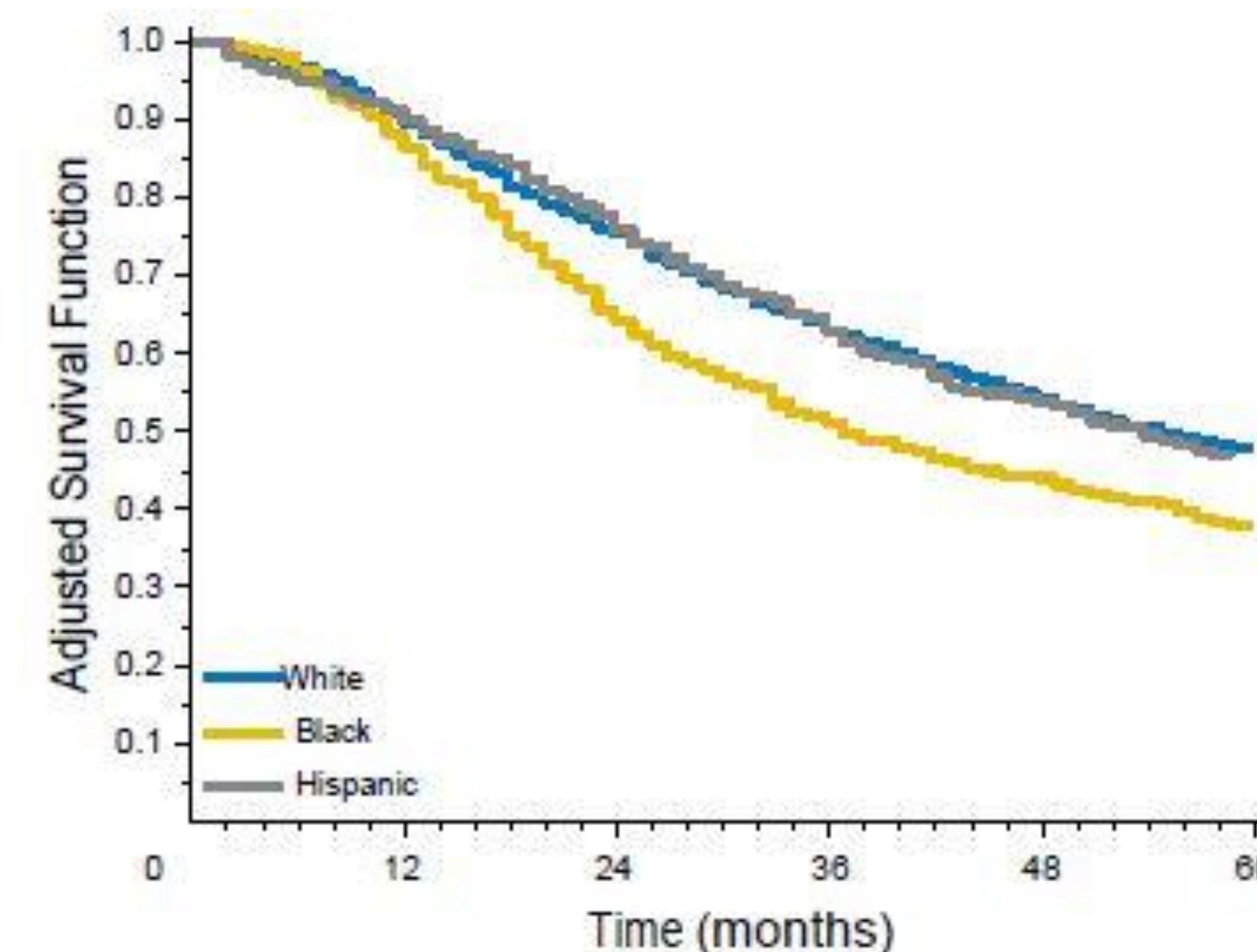


Figure 3: Adjusted overall survival curves for young-onset for stage III CRC. (Image from Holowatyj AN, Ruterbusch JJ, Rozek LS, Cote ML, Stoffel EM. Racial/Ethnic Disparities in Survival Among Patients With Young-Onset Colorectal Cancer. *JCO*. 2016;34(18):2148-2156.)

CLINICAL COURSE

- One month after discharge, he was admitted to our institution for recurrence of his abdominal pain. CT of the abdomen again showed inflammation and thickening of the cecum and terminal ileum.
- The patient underwent colonoscopy which found an obstructing, fungating mass in the distal cecum. Tissue pathology confirmed moderately differentiated adenocarcinoma Stage III (PT3PN2b).
- He underwent right hemi-colectomy with surgical pathology confirming metastatic moderately differentiated adenocarcinoma
- Germline testing was positive for mutation in MLH1, consistent with Lynch Syndrome.

DISCUSSION

- Younger patients have nearly double the prevalence of pathogenic germline variants, with the most frequent being Lynch Syndrome (LS).
- Around half of the patients diagnosed with LS are the first in their family to be diagnosed with a predisposing cancer syndrome.
- Survival data is variable on patients with EO-CRC; however current consensus is that patients diagnosed under 30 years old have the worst survival.
- Young Hispanics have trended to a worse survival probability compared to NHW.
- In conclusion, due to the rising young Hispanic population in conjunction with EO-CRC incidence, clinicians will need to be cognizant of this as a potential diagnosis as well as the ramifications in survivability and screening.

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