HONORHEALTH

HonorHealth Research and Innovation Institute, Scottsdale, Arizona

# Importance of Germline Testing in Colorectal Cancer Patients

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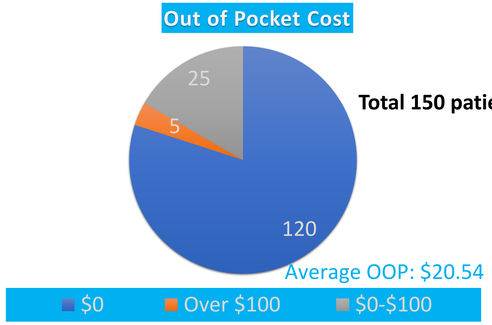
## Background

- Most commonly, colorectal cancer (CRC) occurs sporadically. However, well acknowledged familiar cancer syndromes are also identified.
- Genetic susceptibility to CRC includes Lynch Syndrome, familiar adenomatous polyposis, MUTYH associated polyposis, and others.
- There are individuals that do not meet certain criteria, by lack of family history, polyposis or age for genetic testing. These individuals are considered at average risk for developing CRC.
- Due to lack of national guidelines, identification of genetic testing in average risk individuals who have a diagnosis of colorectal cancer is not the standard of care. **Identification of a germline pathogenic variant in already diagnosed cancer patients is usually not done.**
- By identifying a germline pathogenic variant, it can lead to potential novel treatment options for patients, along with cascade testing of at-risk family members.
- Cascade testing can lead to early identification of a mutation in a family member which is essential for cancer prevention and early detection.

## Method

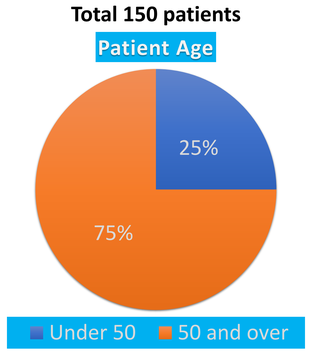
At HonorHealth Research and Innovation Institute, most patients with CRC are referred to us to discuss clinical trial options. Regardless of patient's age or family history, all patients undergo germline testing. For 5 years, we have been using the same laboratory/Ambry Genetics for the germline testing. As their technology advanced, their testing panel increases from 64-91 genes.

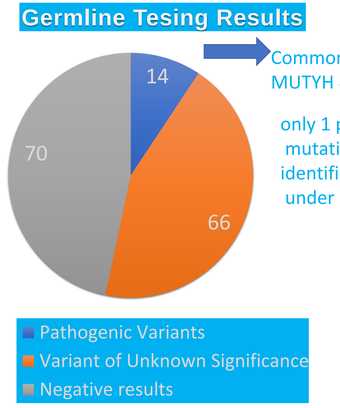



**All** patients with a diagnosis of colorectal cancer should undergo **germline testing despite family history.**

Almost 10% of patients with no significant family history of CRC had pathogenic mutations.


## Results




Common mutations: MUTYH & CHEK2

only 1 pathogenic mutation was identified in a patient under 50 years old

**Cascade testing of family members within 90 days of a patient's test report was completed free of charge this cascade testing resulted in identification of mutations within family members.**

## Conclusion/recommendations

All Advanced Practice Providers should encourage patients with a diagnosis of CRC to complete germline testing, regardless of age and family history.