INVITAE

# Genetic testing in colorectal cancer care: Earlier detection leads to better care for patients and lower costs

**Colorectal cancer** is the second deadliest cancer in the US,[1] causing more than

**53,000**

**deaths each year.[2]**

Nearly

**150,000**

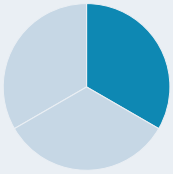
**people are diagnosed**

each year, over half of them Medicare enrollees.[2]

It's also the **fourth most common cancer** in the US.

---

**Genetics** can play an essential role in effecive treatment of colorectal cancer, with familial or inherited factors contrinuting to nearly **one-third of cases.[3]**



NCCN Clinical Practice Guidelines (NCCN Guidelines®) **recommend germline genetic testing for all patients with colorectal cancer** diagnosed before age 50, and in all patients with colorectal cancer that is mismatch repair (MMR) deficient. Testing **can be considered for patients** without MMR deficiency who are **diagnosed at or after age 50.*[4]**

## The average lifetime risk of colorectal cancer increases dramatically for people with genetic changes:

On average, **1 in 25** people develop colorectal cancer.[5]

That increases to...

Nearly

**1 in 6**

for people with genetic variants associated with familial colorectal cancer.[6]

More than

**2 in 3**

for people with genetic variants associated with Lynch syndrome.[6]

Up to

**100%**

for untreated people with variants associated with familial adenomatous polyposis.[7]

**Genetic testing for colorectal cancer patients and their families can help provide the right care for the right patients at the right time, while reducing healthcare costs and saving lives.[8]**

## Genetic information can help physicians improve care with precision medicine by:

- **Identifying treatment and management strategies that are more likely to work:**[9]
  As many as **67% of colorectal cancer patients** with an inherited genetic variant could be eligible for precision therapy and/or clinical trials based on their genetics.[10] Additionally, more frequent screenings (e.g. colonoscopies) may also be used to detect cancer earlier for those patients with an increased risk for cancer.
- **Evaluating a person's risk of developing future cancers:**[11]
  The same genetic variants that can increase a person's risk of colorectal cancer can also increase their risk of other types of cancer.[10] If that's known ahead of time, patients can take steps to reduce their risk of a second cancer.
- **Identifying at-risk relatives who may also have inherited the genetic change:**[12]
  Recent studies show that the US could **save $4.2 billion† from early diagnosis** of colorectal cancer.[13] When a diagnosis is made at stage I rather than stage IV, **an estimated $60,000 is saved per Medicare patient** in the first year.[13] Genetic testing for family members of colorectal cancer patients helps make earlier diagnosis possible.

## Yet too few patients receive testing today.

A recent study utilizing data from a large US national health plan reported the use of genetic testing in 8% of eligible colorectal cancer patients with commercial insurance and only 4% of eligible colorectal cancer patients with Medicare.[9] Of the patients who did get testing, **18% had an inherited genetic variant**[10] that could confer eligibility for precision therapy, clinical treatment trials, and/or clinical management changes and recommendations. The INTERCEPT study demonstrated that universal testing for patients with colorectal cancer can impact patient treatment.[14] That same opportunity for genetics-informed care planning was not available for the ~90% of colorectal cancer patients who did not receive genetic testing.

In a recent study, researchers found that **if all colorectal cancer patients received genetic testing, it would identify approximately 64% more patients who could potentially benefit from targeted treatments.**[15]

*Germline multigene testing in patients with CRC aged 50 y and older without a known MMR deficiency in the tumor is a category 2B recommendation. For patients with CRC aged 50 y and older with a known MMR deficiency in the tumor, additional tumor testing may be recommended instead of germline multigene testing.

† Inflation-adjusted from 2011 to 2021 dollars.


1. SEER Cancer Stat Facts: Colorectal Cancer. National Cancer Institute. Bethesda, MD. Date accessed August 10, 2021. Retrieved from https://seer.cancer.gov/statfacts/html/colorect.html
2. American Cancer Society. (2020). Colorectal cancer facts & figures 2020-2022. Published online, 48.
3. Jasperson *et al.* (2010). Hereditary and familial colon cancer. *Gastroenterology.* 138(6):2044-58.
4. **Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCNGuidelines®) Genetic/Familial High-Risk Assessment: Colorectal V.1.2022. ©National Comprehensive Cancer Network,Inc. 2022. All rights reserved. Accessed 09/08/2022. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.**
5. American Cancer Society. Key Statistics for Colorectal Cancer. Date accessed August 10, 2021. Retrieved from https://www.cancer.org/cancer/colon-rectal-cancer/about/key-statistics.html
6. Veldwijk J *et al.* (2016). Preferences for genetic testing for colorectal cancer within a population-based screening program: a discrete choice experiment. *European Journal of Human Genetics.* 24(3):361–366.
7. Jasperson KW, Patel SG, Ahnen DJ. APC-Associated Polyposis Conditions. 1998 Dec 18 [Updated 2017 Feb 2]. In: Adam MP, Ardinger HH, Pagon RA, *et al.*, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1345/
8. Ladabaum, U. *et al.* (2011). Strategies to identify the Lynch syndrome among patients with colorectal cancer: a cost-effectiveness analysis. *Annals of internal medicine,* 155(2), 69-79.
9. National Cancer Institute. Genetics of Colorectal Cancer (PDQ®) - Health Professional Version. Date accessed August 10, 2021. Retrieved from https://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq
10. Nielsen et al. (2021). Clinical impact of medical policy supporting universal germline testing for patients with colorectal cancer. *Journal of Clinical Oncology.* 39:15_suppl, 10514-10514. https://ascopubs.org/doi/abs/10.1200/JCO.2021.39.15_suppl.10514
11. Patel, S. G., & Ahnen, D. J. (2012). Familial colon cancer syndromes: an update of a rapidly evolving field. *Current gastroenterology reports,* 14(5), 428-438.
12. Burt, R. W., Cannon, J. A., & David, D. S. (2013). National Comprehensive Cancer Network. *J Nat Compr Canc Netw,* 11, 1538-1575.
13. Kakushadze, Z., Raghubanshi, R., & Yu, W. (2017). Estimating cost savings from early cancer diagnosis. Data, 2(3), 30.
14. Uson Jr, P. L., Riegert-Johnson, D., Boardman, L., Kisiel, J., Mountjoy, L., Patel, N., ... & Samadder, N. J. (2021). Germline Cancer Susceptibility Gene Testing in Unselected Patients With Colorectal Adenocarcinoma: A Multicenter Prospective Study. *Clinical Gastroenterology and Hepatology.*
15. Muller, C. *et al.* (2021). Underdiagnosis of Hereditary Colorectal Cancers Among Medicare Patients: Genetic Testing Criteria for Lynch Syndrome Miss the Mark. *JCO Precision Oncology, 5,* 1103-1111.