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# Hereditary Cancer Test

$249 self-pay or insurance billing

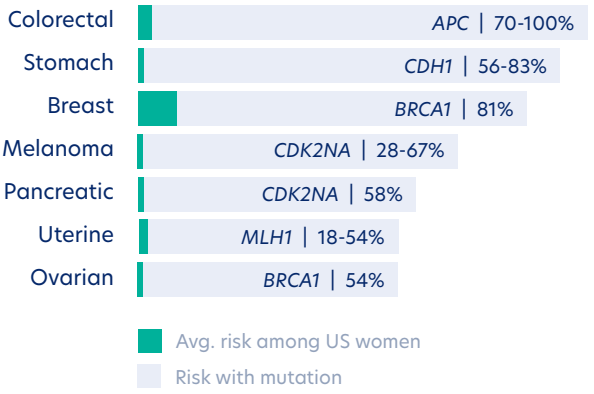
## Clinical-grade, affordable genetic testing

Color's Hereditary Cancer Test analyzes the genes relevant for mutations that can increase your patient's risk for common hereditary cancers, including breast, colorectal, melanoma, ovarian, and other cancers.

Multi-gene panel testing for patients with suspected hereditary breast and ovarian cancer risk identifies substantially more individuals with relevant cancer risk than does *BRCA1* and *BRCA2* testing alone.[1] In a Color study of over 23,000 individuals, 70% of genetic mutations associated with hereditary cancer were outside of the *BRCA1* and *BRCA2* genes.[2]

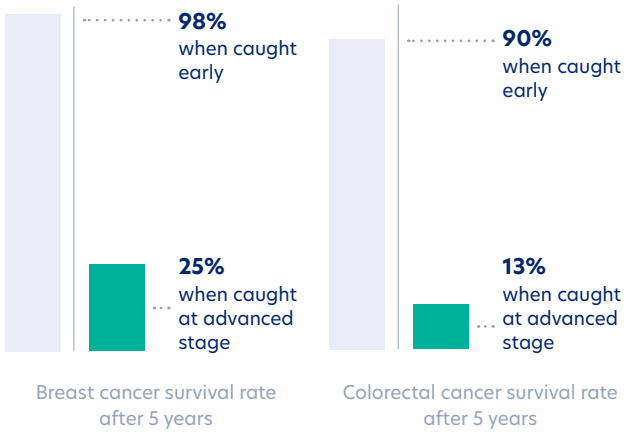
### A genetic mutation can greatly increase cancer risk.

The genes selected were based on their association with increased cancer risk. Mutations in the genes below may increase cancer risk as shown.[3-9]




### Early cancer detection improves the odds of survival.

The 5-year survival rates for the cancers covered by the Hereditary Cancer Test increase dramatically when caught at an earlier and more treatable stage.[10-11]




### Coverage and accuracy

Color performed blinded validation studies and all genetic variants were detected with >99% sensitivity and 100% concordance.

**Specifications:**

- Sequence of the complete coding and adjacent intronic sequences for the genes relevant for mutations
- Analysis of single nucleotide variants, small insertions/deletions, and structural variants such as copy number variants, insertions and inversions
- Minimum read depth: 20X (>99% at >50X)
- Median read depth: 250X

### Variant classification

- Reported variants are confirmed by alternate technologies, including Sanger sequencing, MLPA or aCGH according to Color's internal protocols.*
- Variants of Unknown Significance (VUSs) and likely pathogenic variants are re-reviewed every six months as available medical literature and scientific knowledge are updated. Most VUSs are eventually found to be harmless. We will update you and your patient if a reported variant is reclassified.

*Certain exceptions apply. Variants will not be confirmed if, after testing, there is insufficient DNA available for secondary confirmation. Variants called at high confidence (color.com/variantconfidence) will be reported without secondary confirmation if the variant has been confirmed at least three times in previous carriers.

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Genes covered

| Gene | Breast | Ovarian | Uterine | Colorectal | Melanoma | Pancreatic | Stomach | Prostate* |
|---|---|---|---|---|---|---|---|---|
| BRCA1 | • | • | | | | • | | • |
| BRCA2 | • | • | | | • | • | | • |
| MLH1 | | • | • | • | | • | • | |
| MSH2 | | • | • | • | | • | • | |
| MSH6 | | • | • | • | | | • | |
| PMS2*** | | • | • | • | | | | |
| EPCAM** | | • | • | • | | • | • | |
| APC | | | | • | | • | • | |
| MUTYH | | | | • | | | | |
| MITF** | | | | | • | | | |
| BAP1 | | | | | • | | | |
| CDKN2A | | | | | • | • | | |
| CDK4** | | | | | • | | | |
| TP53 | • | • | • | • | • | • | • | • |
| PTEN | • | | • | • | • | | | |
| STK11 | • | • | • | • | | • | • | |
| CDH1 | • | | | | | | • | |
| BMPR1A | | | | • | | • | • | |
| SMAD4 | | | | • | | • | • | |
| GREM1** | | | | • | | | | |
| POLD1** | | | | • | | | | |
| POLE** | | | | • | | | | |
| PALB2 | • | • | | | | • | | |
| CHEK2 | • | | | • | | | | • |
| ATM | • | | | | | • | | |
| BARD1 | • | | | | | | | |
| BRIP1 | • | • | | | | | | |
| RAD51C | | • | | | | | | |
| RAD51D | | • | | | | | | |

* Please note that research and screening guidelines for genes associated with hereditary prostate cancer are still in their early stages. It is part of the Color service to keep you updated if any information related to your results changes.

** Only positions known to impact cancer risk analyzed: CDK4: only chr12:g.58145429-58145431 (codon 24) analyzed, EPCAM: only large deletions and duplications including 3' end of the gene analyzed, GREM1: only duplications in the upstream regulatory region analyzed, MITF: only chr3:g.70014091 (including c.952G>A) analyzed, POLD1: only chr19:g.50909713 (including c.1433G>A) analyzed, POLE: only chr12:g.133250250 (including c.1270C>G analyzed.

*** PMS2: Variants of uncertain significance are not reported for exons 12-15. Analysis excludes three variants commonly observed in the pseudogene PMS2CL: c.2182_2184delinsG, c.2243_2246delAGAA and deletion of exons 13-14 (chr7:g.6015768_6018727del).



**color.com/clinicians-tests** | (844) 362-6567 or +1 (650) 651-7130 outside the US | **providers@color.com**