color

# High-Quality, Accurate Results

Color's laboratory, team, and processes generate results you can trust.


## Coverage and Accuracy

- Color performed a blinded study to assess the validity of our test. Over 700 samples were studied, and all genetic variants were detected with ≥99.5% accuracy.
- Our CAP-accredited and CLIA-certified laboratory uses the newest technology, including 2D barcoded tubes and advanced liquid-handling robots, to ensure the integrity of every result.
- The quality of every sample is checked multiple times as it moves through the sequencing and interpretation process.
- A certified medical professional reviews every result before it is released.
- Specifications:
    - Full sequencing and large rearrangements of all 30 genes
    - Minimum read depth: 20X (>99% at >50X)
    - Median read depth: 250X (up to >1000X)
    - Intronic coverage: +/- 20bp, as well as intronic tiling

## Variant Classification

- Our Ph.D. and M.D. scientists use state-of-the-art tools to classify variants according to ACMG guidelines.
- Likely pathogenic and pathogenic variants, copy number variations, insertions and inversions are confirmed by an alternative technology according to Color's internal protocols.
- VUS and likely pathogenic variants are re-reviewed every six months, as available medical literature and scientific knowledge are updated. Most VUS's are eventually found to be harmless, and when there is more information we will contact you and your patient.

## Genetic Counseling

Color offers you and your patient complimentary access to our team of board-certified genetic counselors to answer any questions you may have about your patient's results.

## Turnaround Time

Our average turnaround time is currently 3-4 weeks from the day we receive your patient's activated sample in our laboratory, but the actual time will be subject to the data associated with each unique sample.

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# Genes Covered

Color's Hereditary Cancer Test analyzes the most relevant genes for mutations that could increase your patient's risk for breast, colorectal, melanoma, ovarian, pancreatic, prostate, stomach, and uterine cancers.

| 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* | • | | | | | • | | • |
| *NBN* | • | | | | | | | • |
| *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*: Exons 12-15 not analyzed.