Hereditary Cancer Gene Test

High-Quality, Accurate Results

Coverage and Accuracy

- In a blinded study to assess the validity of the test, over 500 samples were studied, and all genetic variants were detected with greater than 99% accuracy.
- The 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 scientists use state-of-the-art tools to classify variants according to ACMG guidelines.
- All clinically actionable variants (i.e. likely pathogenic and pathogenic) as well as all reported copy number variations, insertions and inversions are confirmed by a secondary technology such as Sanger sequencing, array CGH, or MLPA.
- 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 Is Available Upon Request

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

Turnaround Time

The 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 by the Hereditary Cancer Gene Test

The Hereditary Cancer Gene 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 laboratory 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.

E&OE
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