myBRCA HiRisk Technical Information

VeritasGenetics

Targeted next generation sequencing and large rearrangement analysis in 26 genes.

TARGETED GENES

- *BRCA1* and *BRCA2* along with 24 other genes known to be associated with increased risk for hereditary breast and/or ovarian cancer.
- This multi-gene panel test also includes genes known to be associated with an increased risk for other cancer types such as colorectal, pancreatic, gastric, endometrial, and thyroid.
- Genes cover several hereditary cancer syndromes including Cowden syndrome, Li-Fraumeni syndrome, Peutz-Jeghers syndrome, and Lynch syndrome.

ATM	*BARD1*	*BLM*	*BRCA1*	*BRCA2*	*BRIP1*	*CDH1*	*CHEK2*	*EPCAM*	*FAM175A*	*MEN1*	*MLH1*	*MRE11A*
MSH2	*MSH6*	*MUTYH*	*NBN*	*PALB2*	*PMS2*	*PTEN*	*RAD50*	*RAD51C*	*RAD51D*	*STK11*	*TP53*	*XRCC2*

REPORTING

- *BRCA1* and *BRCA2* large rearrangement analysis included (>99.9% sensitivity and specificity).
- Large rearrangement analysis for remaining 24 genes includes multi-exon del/dups only.
- *EPCAM* mutation detection is limited to del/dup analysis of the 3' end of the gene.
- *PMS2* pseudogene analysis included.
- Point mutations and small insertions/deletions are detected (>99.9% sensitivity and specificity).
- Likely pathogenic or pathogenic intronic variants up to 10 base pairs from the coding region are always reported.
- Variant classification based on ACMG Guidelines.

HIGHEST ACCURACY

- Clinically validated based on CDC, CLIA, CAP, ACMG and CLSI guidelines.
- Analytical sensitivity and specificity over 99.9%.
- In validation studies, 100% reproducibility and repeatability across runs and operators with no false positives and negatives detected.
- Proprietary bioinformatics pipeline and variant assignment workbench used for data analysis.
- 33% of validation samples with known ethnicity were from non-white donors, including persons of Hispanic, African-American, East Asian, and Middle Eastern ancestry.

MOST COMPLETE VARIANT DATABASE

- More than 25,000 variants representing over 500,000 patients, classified based on concordance among multiple clinical labs and curated and annotated by a physician.

FREE THE DATA

- Veritas strongly believes in data sharing to allow peer review and strengthen our collective knowledge about risk variants. Therefore, we have joined the Global Alliance for Genomics and Health (GA4GH) and FreeTheData movements and are contributing high quality, de-identified variant information to academic databases.

Samples Accepted	Saliva, whole blood, extracted genomic DNA
Amplicon Coverage	The assay offers full coverage of 82,853 base pairs of genomic sequence, including the complete coding regions and splice sites.
Validation	87 samples (69 positives and 18 negatives) were used for validation. All mutations were detected; there were no false positives and no false negatives.

ABOUT VERITAS GENETICS

Veritas Genetics aims to make high quality affordable genetic testing available to every patient who needs it. We believe that genetic knowledge can help improve patient care by providing recommendations that can lead to disease prevention and early detection. Veritas Genetics is founded and supported by clinicians and scientific leaders from the world's top academic institutions including Harvard Medical School and MIT.

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