Hereditary breast and ovarian cancer syndrome associated with *BRCA1* or *BRCA2* gene defect is characterized by an increased risk for male and female breast cancer, ovarian cancer (includes fallopian tube and primary peritoneal cancers), and to a lesser extent other cancers such as prostate cancer, pancreatic cancer or melanoma.

The SD Genomics BRCACancerDISCOVERY™ tests *BRCA1* and *BRCA2* genes and provides the degree of cancer risk following from the identification and management guidelines based on identified variants.
In addition, the BRCACancerDISCOVERY™ is helpful to decide poly (ADP–ribose) polymerase (PARP) inhibitors as treatment for ovarian or breast cancers. This analytic service is recommended for the proactive or diagnostic testing.

REPORTABLE RANGE

Identified variants are classified according to 2015 guideline by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology (ACMG/AMP; Genet Med 2015;17:405–24, PMID–25741868) following a five–tier terminology system using the following terms: pathogenic, likely pathogenic, variant of uncertain significance, likely benign, and benign.

- Every variant classified as "pathogenic" and "likely pathogenic in all analyzed genes will be reported.
- Variants classified as "uncertain significance" will be reported. Uncertain significance means that it is unclear if this change increases risk for hereditary breast and ovarian cancer. Variants are interpreted using the literatures and public databases at the time of reporting. To account for new medical or scientific findings, all reported variants of uncertain significance are reviewed and reclassified annually. If variants are reclassified, updated reports with new interpretation will be provided.
- Variants classified as "likely benign" and "benign" are not reported.

Variant classification	Definition
Pathogenic variant	A variant is certain of being disease–causing.
Likely pathogenic variant	A variant has a greater than 90% certainty of being disease–causing.
Variant of uncertain significance (VUS)	A variant is uncertain of being disease–causing or benign.
Likely benign variant	A variant has a greater than 90% certainty of being benign.
Benign variant	A variant is certain being benign.

POTENTIAL RESULTS

- Testing positive for a clinically relevant genetic variant in the tested genes means that there is an increased risk for developing hereditary breast and ovarian cancer syndrome in the future. This result does not necessarily mean that there will be a development of hereditary cancers in lifetime. If test results are positive, genetic counseling is strongly recommended. This result may be informative to biological relatives.
- Testing negative for a clinically relevant genetic variant in any of the tested genes means that there is a reduced, but not eliminated risk for developing hereditary breast and ovarian cancer syndrome in the future. An individual's risk of cancer depends not only hereditary genetic factor but also on acquired factors such as age, environment, lifestyle, etc. It is important to note that negative results do not guarantee that there won't be any cancer development in the future.

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LIMITATIONS

- This test is designed to analyze the coding exons and adjacent introns of genes tested. Note that the variants on the region such as deep intron, promotor, or UTR are unable to be identified.
- Specific genetic alterations are unable to be detected by this test. Such genetic alterations are as followed: chromosomal aneuploidy, large genomic rearrangement (translocation, large insertions, large deletion, or inversions), or mosaicism
- Specific regions of a genomic DNA may limit the accuracy of results. Such regions are as followed: pseudogenes with high homology to the gene of interest, genetic changes from repetitive sequences, homopolymeric regions, or genetic alterations caused by low-level mosaicism.
- In addition, the test results may be incorrect if contaminated specimen is provided; quality of extracted DNA is poor; or inaccurate or insufficient personal clinical information is provided.

WORDS OF CONSENT

- I confirm that I have read and understand the information provided in this informed consent document.
- I will not make any medical decisions based on the test results without genetic counseling provided by healthcare professionals such as physicians, genetic counselors, or geneticists.
- I acknowledge that my deidentified sample, genetic information, and test results may be used in SD Genomics' internal quality control, laboratory validation studies, and research & development.
- I hereby authorize SD Genomics to conduct the following genetic test for myself:

☐ BRCACancerDISCOVERY™

Printed patient name:	
Patient signature:	Date (DD/MM/YYYY):
Email address:	

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