CGAT - Hereditary Breast and Ovarian Cancer v1.0 (05/2025)

Panel Description

This hereditary breast and ovarian cancer (HBOC) panel covers 19 genes implicated in hereditary predisposition to breast and ovarian cancer. This panel is useful for evaluating hereditary cancer risk for patients with a personal or family history suggestive of a hereditary breast and ovarian cancer syndrome, allowing for targeted cancer surveillance based on associated risks as well as allowing for predictive testing, and appropriate screening of at-risk family members. Therapeutic eligibility with certain targeted therapies may also be assessed, for example: eligibility for treatment with poly adenosine diphosphate-ribose polymerase (PARP) inhibitors based on certain gene alterations (eg, BRCA1, BRCA2) in selected tumor types. Besides the full HBOC panel, a single gene or subset of genes can be selected from this list as part of a custom panel.

Panel Gene Content

ATM	BRIP1	MLH1	PALB2	RAD51D
BARD1	CDH1	MSH2	PMS2*	STK11
BRCA1	CHEK2	MSH6	PTEN	TP53
BRCA2	EPCAM^	NF1	RAD51C	

^ only copy number events, and not SNVs, will be evaluated in EPCAM
*variation confined to exons 11-15 of PSM2 will not be assessed. See below

Coverage and Limitations

- All RefSeq coding exons and up to +/- 10bp of adjacent intronic sequence of the genes listed are covered by at least 20 reads with an expected MQ >15.
- Due to pseudogene interference, it is not possible to accurately detect variant calls in PMS2 exons 11-15. Therefore, small nucleotide variant calls in these exons, as well as copy number alterations confined to these exons will not be analyzed.
- Copy number events affecting MSH6 exon 1 are expected to be detected, but this has not been reliably verified due to lack of positive controls.
- Only copy number alterations affecting the 3’ end of EPCAM are relevant to hereditary cancer testing. Therefore, SNVs in EPCAM will not be assessed.