**To: Dartmouth Health Providers**

**From:** Nidhi Shah, MD; Wahab A. Khan, PhD; Joel A. Lefferts, PhD; Parth Shah, MD; Arief Suriawinata, MD

**Date:** 5/15/25

**Re:       DH Hereditary Breast and Ovarian Cancer (HBOC) Panel go-live**

The Clinical Genomics and Advanced Technologies (CGAT) Laboratory in the Department of Pathology at DHMC will be offering Hereditary Breast and Ovarian Cancer (HBOC) Panel, beginning 5/19/25. The panel analyzes the protein-coding regions of 19 genes to identify variants implicated in hereditary breast and ovarian cancer risk/predisposition.

Gene Content

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

^Note that variant detection is limited in specific regions of these genes. See specific limitations below.

**Turnaround time:** Results will typically be available within 4 weeks for HBOC panel.

**Specimen details:** The preferred specimen is EDTA anticoagulated peripheral whole blood. Buccal swab samples can also be accommodated.

- Blood Collection tube: Purple top tube with EDTA for whole blood; volume: 3-5 mL (minimum acceptable: 0.5-1 mL)

- Buccal Collection tube: ORAcollect (OCR-100) from DNA Genotek Inc. (provided by lab)

- Storage (blood and buccal): Ambient temperature for transport within 24 hours to CGAT lab or refrigerated over weekend

**Test ordering details: LAB2333**

**Limitations:** Due to pseudogene interference, variants will not be detected in exons 11-15 of PMS2. Copy number events affecting MSH6 exon 1 may not be reliably detected. In EPCAM, only copy-number alterations affecting the 3' end of the gene will be assessed.

**For questions or additional information, please contact:** Nidhi Shah, Wahab Khan, Joel Lefferts, Parth Shah, Heather Steinmetz (CGAT manager) or Samantha Allen (CGAT supervisor) in the Department of Pathology and Laboratory Medicine.