Oncomine BRCA Assay GX

Enabling large genomic rearrangement detection in *BRCA1* and *BRCA2* genes in as little as 24 hours

The Ion Torrent™ Oncomine™ BRCA Assay detects *BRCA* somatic and germline mutations from formalin-fixed, paraffin-embedded (FFPE) tissue and whole blood, and has been widely adopted in molecular pathology laboratories across the world and referenced in several publications [1–9]. In addition to the Ion GeneStudio™ SS System, the assay is now available on the Ion Torrent™ Genexus™ System.

Robust, rapid, and consistent performance
- Fully verified on clinical research samples
- Based on proven Ion AmpliSeq™ technology
- Requires only 20 ng of DNA input
- 100% exonic coverage with large intronic flanking regions
- Detects large genomic rearrangement, such as large insertions and deletions (indels) and exon-level duplication and deletion, removing the need to employ multiple technologies
- Enables detection of all relevant variant types with high confidence

The Oncomine BRCA Assay GX is now available on the Genexus System
- One-day specimen-to-report workflow, operated on Ion Torrent™ Genexus™ Software
- Unmatched ease of use with only two user touchpoints and 20 minutes of hands-on time
Exceptional performance

Figures 1, 2, and 3 demonstrate the exceptional performance of the Oncomine BRCA Assay. All exons are 100% covered, with an average of 64 bases of flanking sequence into the introns upstream and downstream of each exon, allowing for over 99% confidence of detecting 5% somatic variants across both genes. The uniformity and high read counts help ensure high sensitivity and accuracy of both somatic and germline mutation detection, demonstrated with different workflows and sequencers.

Figure 1. Superior accuracy in detecting somatic and germline variants, and high consistency, independent of the workflow. (A) Sensitivity and positive predictive value (PPV) for detecting single-nucleotide variants (SNV) and indels. Positive predictive value = true positives/total number of positives. Sensitivity = true positives/(true positives + false positives). (B) The percent of concordance for germline analysis and somatic cell analysis with the Ion GeneStudio S5 and Genexus systems.

Figure 2. Relative abundance of BRCA exons is plotted. Shown are the data from a sample that has a deletion in BRCA1 (gold) of exons 22 and 23 (red circle). BRCA2 (green) has no copy number variation (CNV). The gray plot indicates the sample ID (sid) amplicons used for normalization.

Figure 3. Example of a Genexus System report showing the results for a sample with an SNV on the BRCA1 gene, and two deletions and one insertion on the BRCA2 gene.
References


Ordering information

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