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Oncomine *BRCA* Research Assay

A new NGS-based tool to detect somatic and germline mutations from FFPE tissue and blood

New to the Ion Torrent™ Oncomine™ family of products is the Oncomine™ *BRCA* Research Assay for the detection of *BRCA* somatic and germline mutations from formalin-fixed, paraffin-embedded (FFPE) tissue and whole blood. Based on powerful Ion AmpliSeq™ technology, and with 100% exon coverage of both genes, the assay enables robust *BRCA* gene analysis even from challenging FFPE samples.

Robust, rapid, and consistent performance:

- Fully verified on clinical research samples
- Based on proven Ion AmpliSeq technology
- Requires as low as 10 ng DNA input
- 100% exonic coverage with large intronic flanking regions
- Produced with enhanced manufacturing quality control

Easy implementation and use in every laboratory:

- Combined with a tailored bioinformatics solution in Ion Reporter™ Software
- A flexible workflow enabling multi-sample runs
- Allows a choice of Ion Torrent™ platforms and multiplexing levels

"The Oncomine *BRCA* Research Assay is allowing us to get robust and fast results even from small FFPE tissue samples."

Markus Möbs, Senior Research Fellow
Molecular Pathology Laboratory and Institute of Pathology
Charité Medical University Berlin

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Figure 1. The Oncomine *BRCA* Research Assay workflow is flexible, allowing manual or automated template preparation using the Ion OneTouch™ 2 System or the Ion Chef™ System and the Ion PGM™ or Ion S5™ System for highly consistent sequencing.

Oncomine *BRCA* Research Assay: exceptional performance

Figures 2 and 3 demonstrate the exceptional performance of the new Oncomine *BRCA* Research Assay. All exons are covered 100%, 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 (templating and sequencers).

gDNA variants	Platform	Library	Templating	SNV		Indel	
				Sensitivity	PPV	Sensitivity	PPV
5% allele frequency	Ion PGM System/Ion 318™ Chip	Manual	OT2	100	100	99	99
		Ion Chef	Ion Chef	100	99	99	98
	Ion S5 System/Ion 530™ Chip	Manual	Ion Chef	100	98	98	92
		Ion Chef	Ion Chef	100	92	99	99
50%, 100% allele frequency	Ion PGM System/Ion 318™ Chip	Manual	OT2	100	100	100	100
		Ion Chef	Ion Chef	100	100	100	99
	Ion S5 System/Ion 530™ Chip	Manual	Ion Chef	100	100	100	100
		Ion Chef	Ion Chef	100	100	100	100

Figure 2. Superior accuracy in detecting somatic and germline variants, and high consistency independent of the workflow. At 5% allele frequency, >1,000 SNV and >600 indel variants measured. At 50%, 100% allele frequency, >4,000 SNV and >200 indel variants measured. Positive predictive value = true positives/total number of positives. Sensitivity = true positives/(true positives + false positives). OT2 = Ion OneTouch 2

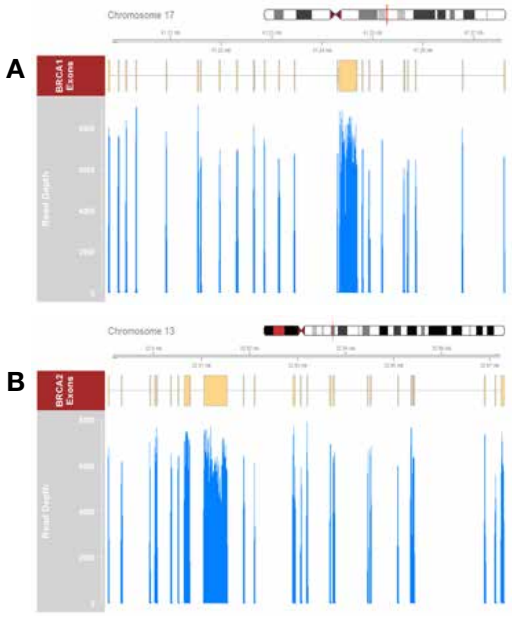

Figure 3. 100% exon coverage across both (A) *BRCA1* and (B) *BRCA2* genes, with high uniformity and read counts across all exons, allowing for over 99% confidence of detecting 5% somatic variants.

Ordering information

Product	Quantity	Cat. No.
Oncomine *BRCA* Research Assay, Manual Library Preparation	24 tests	A32840
Oncomine *BRCA* Research Assay, Chef Ready Library Preparation	32 tests	A32841

For more information, go to
thermofisher.com/oncominebrca

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