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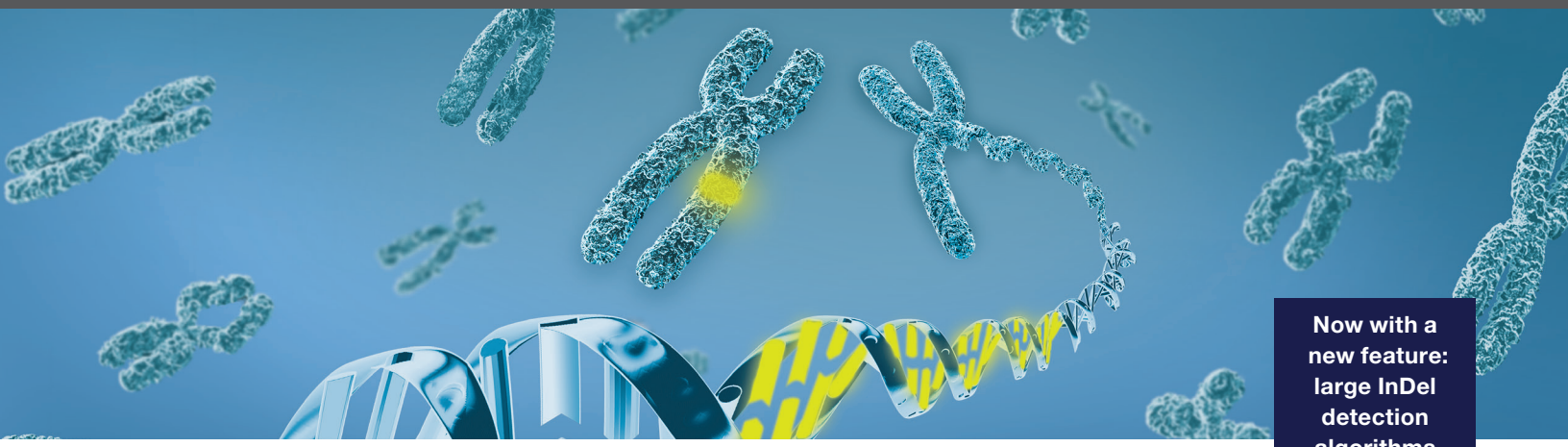

Now with a new feature: large InDel detection algorithms

Oncomine *BRCA* Research Assay

Now with a new feature—large InDel & exon/gene deletion/duplication detection algorithms

The Ion Torrent™ Oncomine™ *BRCA* Research Assay for the detection of *BRCA* somatic and germline mutations from formalin-fixed, paraffin-embedded (FFPE) tissue and whole blood has been widely adopted in molecular pathology laboratories across the world. The Assay can now detect large InDels and exon or whole gene deletion or duplication events, uniquely empowering laboratories to detect all classes of mutations in one NGS workflow, removing the need to employ multiple technologies.

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
- Enables detection of all relevant variant types with high confidence

Easy implementation and use in every laboratory:

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

"The Oncomine *BRCA* Research Assay is allowing us to get fast and robust results on both somatic and germline variants including large InDel detection."

Professor Enrico Tagliafico, MD, PhD
Head of Clinical Genomics Laboratory
Modena University Hospital, Modena, Italy

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Exceptional performance:

Figures 1, 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	Ion Chef	100	99	99	98
	Ion S5™ System/ Ion 530™ Chip	Ion Chef	100	92	99	99
50% and 100% allele frequency	Ion PGM System/ Ion 318 Chip	Ion Chef	100	100	100	99
	Ion S5 System/ Ion 530 Chip	Ion Chef	100	100	100	100

Figure 1. 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 were measured. At 50% and 100% allele frequency, >4,000 SNV and >200 indel variants were measured. Positive predictive value = true positives/total number of positives. Sensitivity = true positives/(true positives + false positives).

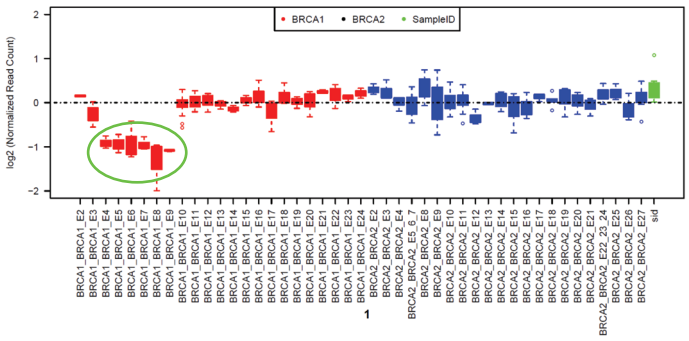

Figure 2. Relative abundance of *BRCA* exons are plotted. Above you can see a sample which has a deletion in *BRCA1* (red) of exons 4–9 (green circle). *BRCA2* (blue) has no CNV. The green plot indicates the sample ID amplicons used for normalization.

Locus	Oncomine Variant Class	Oncomine Gene Cla...	Genes	Copy Number	Type	CNV Subtype	Call	CNV Confidence	Variant ID
chr13:32890490			BRCA2	2	CNV	REF	EXON2-20	100	BRCA2
chr13:32950743	ExonDeletion	Loss-of-function	BRCA2	1	CNV	BigDel	EXON21-27	100	BRCA2
chr17:41197601			BRCA1	2	CNV	REF	EXON2-24	100	BRCA1

Figure 3. Example IR report of a sample showing (bottom row) normal copy number for the *BRCA1* gene, a deletion of exons 21–27 of the *BRCA2* gene (middle row), and normal copy number (top row) for the rest of the *BRCA2* gene (exons 2–20).

Ordering information

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

Find out more at **thermofisher.com/oncominebrca**

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