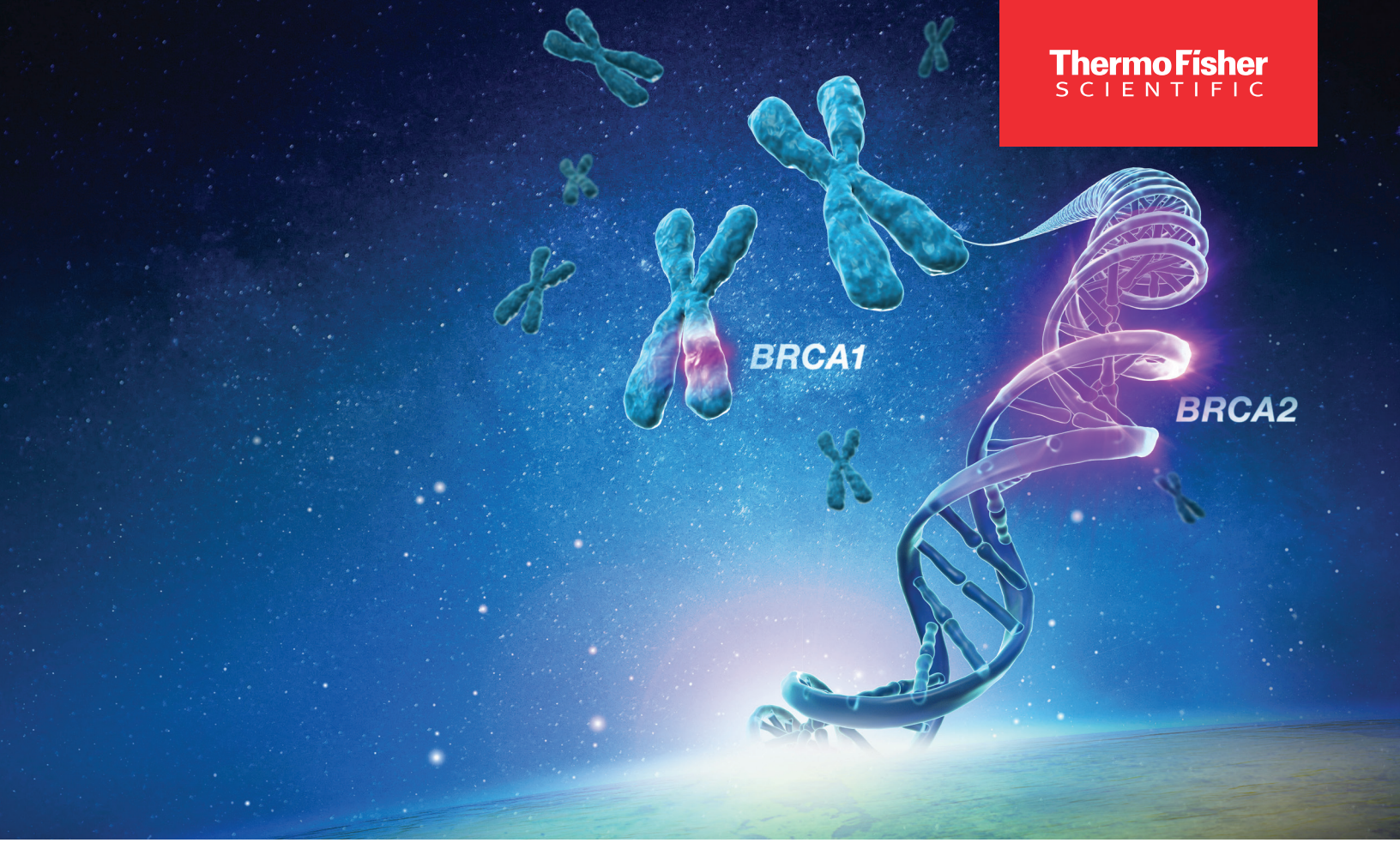

Oncomine BRCA Assay GX

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™ S5 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.

A

Control sample variants	Platform	Library	SNV		Indel	
			Sensitivity (%)	PPV (%)	Sensitivity (%)	PPV (%)
5% allele frequency	Ion GeneStudio S5 System	Manual Chef	100 100	98 92	98 99	92 99
	Genexus System	Genexus	100	100	98	100
50%, 100% allele frequency	Ion GeneStudio S5 System	Manual Chef	100 100	100 100	100 100	100 100
	Genexus System	Genexus	100	100	94	100

B

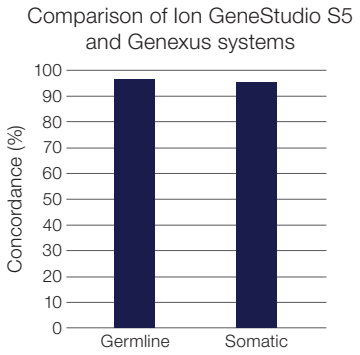

Figure 1. Superior accuracy in detecting somatic and germline variants, and high consistency, independent of the workflow. (A) Sensitivity and positive predictive value (PPV) for detecting single-nucleotide variants (SNV) and indels. Positive predictive value = true positives/total number of positives. 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.

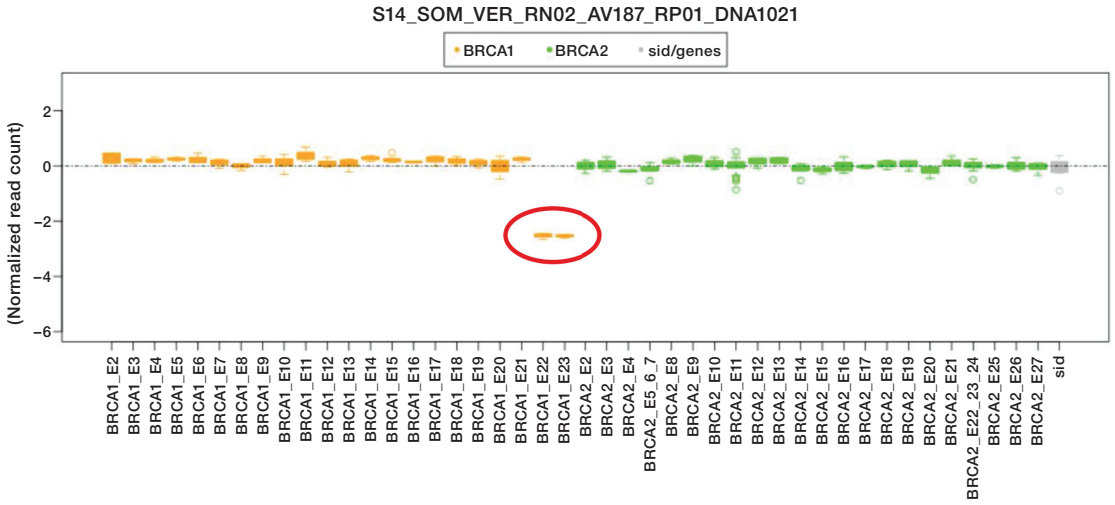

Figure 2. Relative abundance of *BRCA* exons is plotted. Shown are the data from a sample that has a deletion in *BRCA1* (gold) of exons 22 and 23 (red circle). *BRCA2* (green) has no copy number variation (CNV). The gray plot indicates the sample ID (sid) amplicons used for normalization.

All | SNVs/Indels | CNVs

Oncomine BRCA (5.16) Filter Chain Applied 95 of 103 Variants | Edit Filters | Export | Columns

Key Vari...	Type	Oncomine Gene Class	Oncomine Variant Class	Gene	Locus	AA Change	Ref	Alt	Call	Nuc Change	Allele Frequency (%)
Yes	snp	Loss-of-Function	Truncating	BRCA1	chr17:41234451	p.Arg1443Ter	G	A	PRESENT (HETEROZYGOUS)	c.4327C>T	31.6
Yes	del	Loss-of-Function	Truncating	BRCA2	chr13:32913558	p.Lys1691AsnfsTer15	CA	C	PRESENT (HETEROZYGOUS)	c.5073delA	35.7
Yes	ins	Loss-of-Function	Truncating	BRCA2	chr13:32937354	p.Ile2675AspfsTer6	T	TA	PRESENT (HETEROZYGOUS)	c.8021_8022insA	11.1
Yes	del	Loss-of-Function	Truncating	BRCA2	chr13:32913836	p.Asn1784ThrfsTer7	CAA	CA	PRESENT (HETEROZYGOUS)	c.5351delA	44.4

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.

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Ordering information

Product	Size	Cat. No.
For the Ion GeneStudio S5 System		
Oncomine BRCA Research Assay, Manual Library Preparation	24 reactions	A32840
Oncomine BRCA Research Assay, Chef-Ready Library Preparation	32 reactions	A32841
For the Genexus System		
Oncomine BRCA Assay GX	32 reactions	A47912

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